Pediatr Radiol (2006) 36: 71–75
DOI 10.1007/s00247-005-0018-9

HISTORICAL PERSPECTIVE

Benjamin H. Taragin · Walter E. Berdon · Beth Printz

MRI assessment of bronchial compression in absent pulmonary valve syndrome and review of the syndrome

Received: 25 May 2005 / Revised: 16 August 2005 / Accepted: 25 August 2005 / Published online: 9 November 2005

Abstract Absent pulmonary valve syndrome (APVS) is a rare cardiac malformation with massive pulmonary insufficiency that presents with short-term and long-term respiratory problems secondary to severe bronchial compression from enlarged central and hilar pulmonary arteries. Association with chromosome 22q11 deletions and DiGeorge syndrome is common. This historical review illustrates the airway disease with emphasis on assessment of the bronchial compression in patients with persistent respiratory difficulties after valvular repair. Cases that had MRI for cardiac assessment are used to illustrate the pattern of airway disease.

Keywords Absent pulmonary valve syndrome · Airway abnormalities · MRI

Introduction

APVS is a rare but important cardiac cause of respiratory distress in the newborn. In the 1950s and '60s during the development of pediatric cardiology and cardiac surgery, absent pulmonary valve syndrome (APVS) was often referred to as "a variant of tetralogy of Fallot characterized by absent or rudimentary pulmonary valve tissue, severe pulmonary regurgitation, and aneurysmal dilatation of the main and mediastinal branch arteries" (Fig. 1) [1, 2].

B. H. Taragin (✉) · W. E. Berdon
Department of Radiology, Children's Hospital of New York,
3959 Broadway CHN-3,
New York, NY 10032, USA
e-mail: bentaragin@hotmail.com
Tel.: +1-212-3421654
Fax: +1-212-3057233

B. Printz
Department of Cardiology, Children's Hospital of New York,
New York, NY, USA

The causes of airway problems in APVS are different from those encountered in patients with vascular rings. APVS has compliant bronchi and bronchial compression caused by adjacent pulmonary artery dilatation (Fig. 2). Commonly, patients with APVS will continue to have respiratory difficulty if there has been insufficient reduction of the dilated central and pulmonary arteries causing severe bronchial compression.

The usual case of APVS has absence of normally functioning pulmonary valve tissue, annular thickening, a VSD, absence of the ductus arteriosus, and aneurysmal dilatation of the main, right, and left pulmonary arteries. The patient's cardiac disease, primary pulmonary insufficiency, is often not very severe. The inherent disease of the airways, a result of long-standing in utero compression by the tortuous dilated pulmonary arteries, can lead to severe bronchial compression and can eventually be lethal. Several recent articles have appeared in the surgical and intensive care literature addressing the short-term and long-term airway problems [2–4].

In the past, conventional imaging was not a vital part of management of these patients. This was partly related to inability to image the airway appropriately. Plain films in the preoperative neonatal period showed hyperaeration or, rarely, obstructive emphysema (which could be fluid-filled), and occasionally a large hilar mass on either side of the mediastinum. Diagnosis was made by angiography and later echocardiography without direct concern or attention to the airway disease.

We report from our experience with APVS patients, all of whom were followed with plain films and who despite surgical valvular repair (with reduction of the main, right, and left pulmonary arteries) had persistent short-term and long-term airway problems of varying severity. Two of these patients eventually needed tracheotomy. After tracheotomy failed to alleviate the patients' symptoms, MRI was performed to assess cardiac function and revealed severe persistent right and left bronchial collapse caused by the enlarged hilar pulmonary arteries (Fig. 3). Eventually, repeat surgery with careful reduction of the hilar and central pulmonary arteries relieved the bronchial compression and

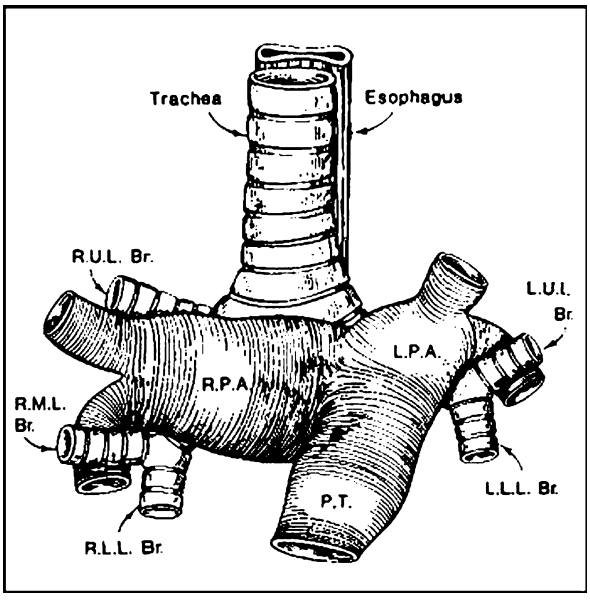

Fig. 1 Diagram illustrates absence of the pulmonary valve, with huge central pulmonary arteries compressing the main bronchi. *Br.* bronchus, *L.L.L.* left lower lobe, *L.P.A.* left pulmonary artery, *L.U.L.* left upper lobe, *P.T.* pulmonary trunk, *R.L.L.* right lower lobe, *R.M.L.* right middle lobe, *R.P.A.* right pulmonary artery, *R.U.L.* right upper lobe (reprinted with permission from ref. 2)

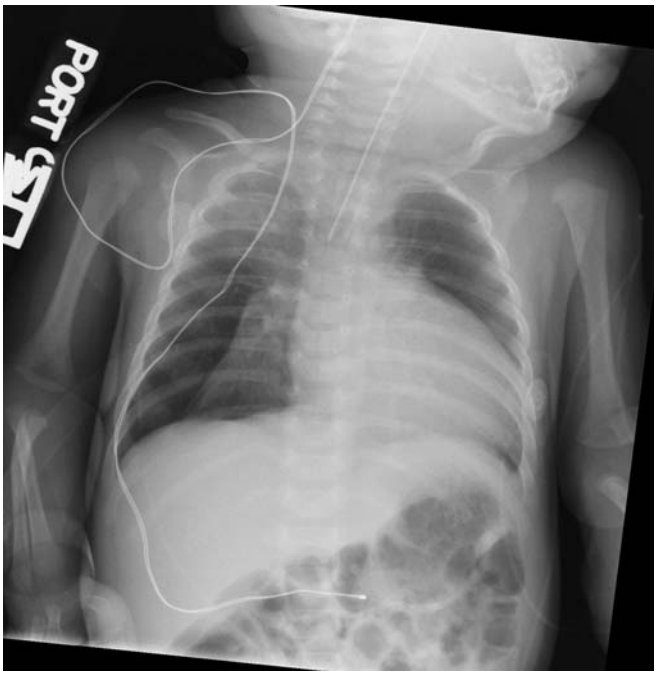

Fig. 2 Chest radiograph of a 2-month-old with APVS shows right lower and partial right upper lobe atelectasis from the obstructive bronchial compression

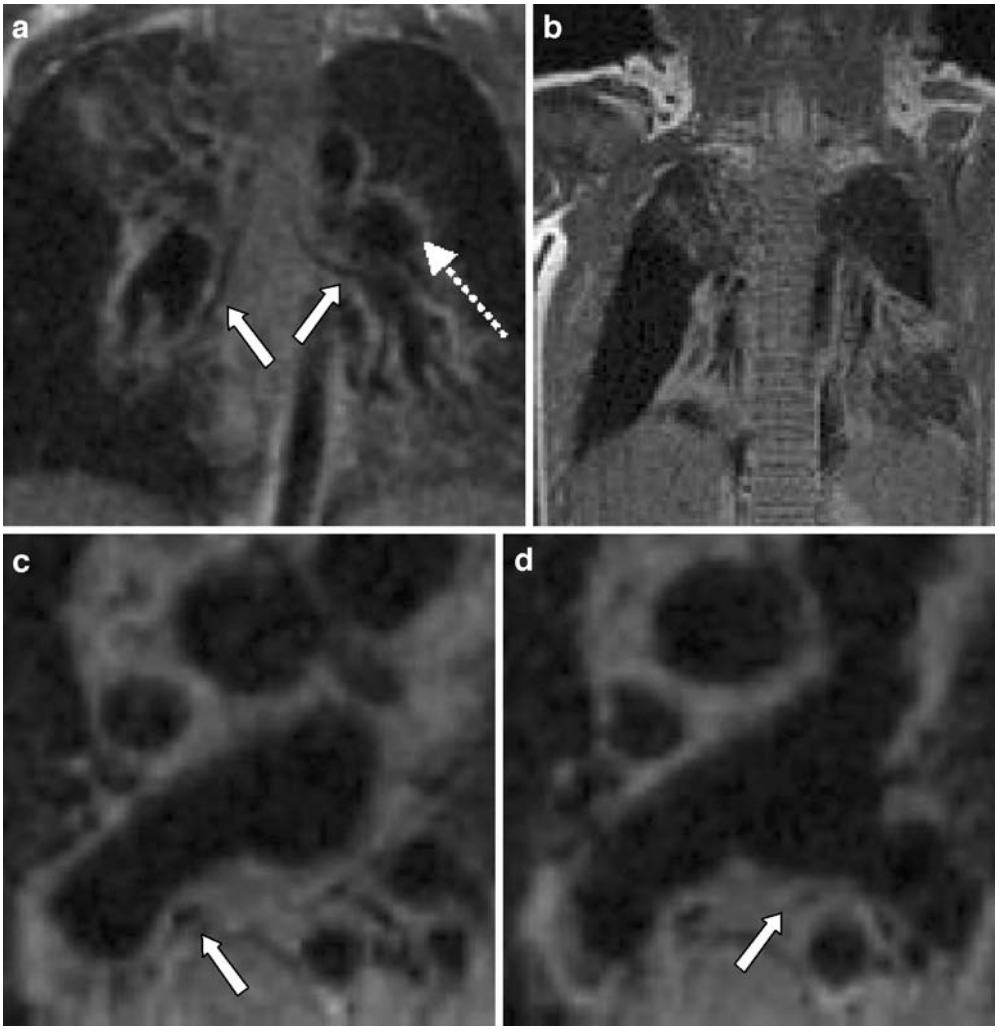

Fig. 3 Coronal (**a**, **b**) and axial (**c**, **d**) MRI images show bilateral areas of atelectasis, narrowed mainstem bronchi (*solid white arrows*) from the pulmonary artery dilatation (*dashed white arrow*) and resultant areas of atelectasis

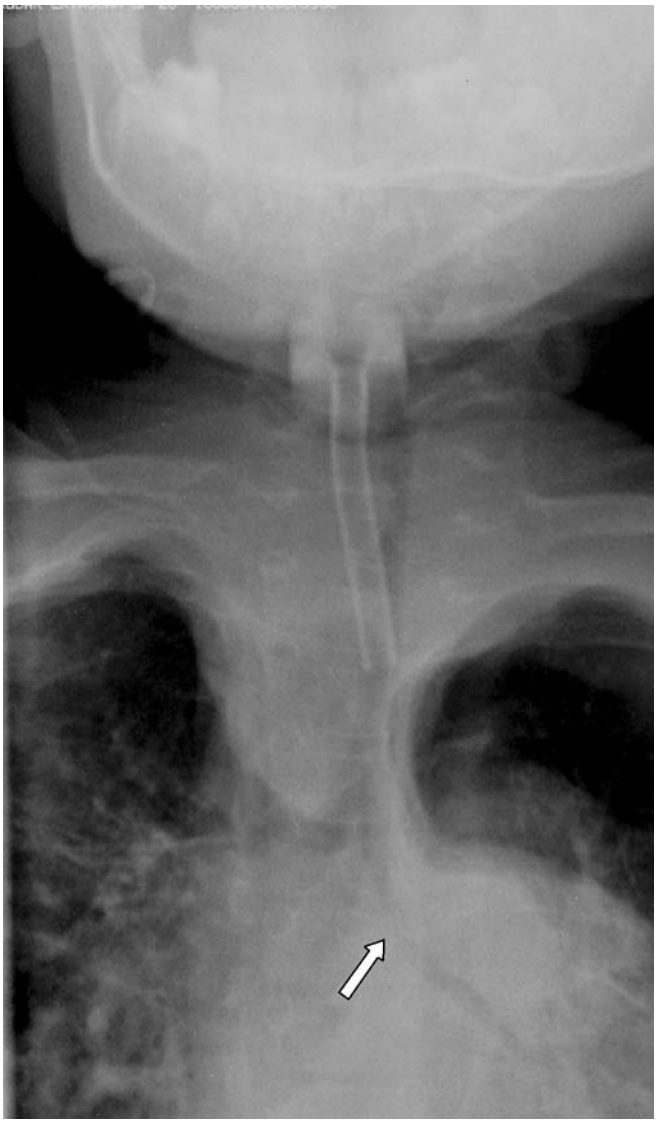

Fig. 4 Magnified airway images of a 2-year-old with APVS with marked narrowing of the mainstem bronchi (*white arrow*)

alleviated the airway disease. Two of the patients who had serious airway problems also had DiGeorge syndrome (22q11 deletions), which is present in many APVS patients.

Case reports

Case 1

A 2-month-old infant with APVS who had had surgical repair, including patching of the outflow tract and reduction plasty of the main right and left pulmonary arteries, could not be weaned off the ventilator. Chest radiography showed multiple areas of atelectasis (Fig. 2). MRI was performed to assess cardiac function and showed severe narrowing of the airways from the dilated pulmonary arteries (Fig. 3).

Case 2

A 2-year-old child born with APVS initially had surgical repair at an outside hospital with valve replacement and central pulmonary artery plasty. However, the patient could never be weaned off the ventilator after the initial surgery. Eventually, the patient required a tracheotomy. Magnification views of the airways performed with a Thoreus filter demonstrated the tracheotomy tube and marked bilateral bronchial narrowing (Fig. 4). MRI demonstrated severe narrowing by the dilated pulmonary arteries (Fig. 5). No thymus was identified. Incidentally noted were other DiGeorge syndrome anomalies, including a right-side aortic arch and a butterfly vertebral body. The second surgery performed at our institution focused on plication and reduction of the peripheral pulmonary arteries. After the second surgery, the patient was weaned off the ventilator and eventually discharged.

Discussion

Although the first reference to APVS is attributed to an article in 1847 by Chevers [5], the case reported by Kurtz et al. [6] in 1927 is really the first reference in the modern era of medical literature. It describes an 11-year-old boy who was cyanotic at birth, had recurrent life-long infections, and tired easily on exertion. After the patient died from ruptured appendicitis, autopsy revealed absence of the pulmonic valve and a 5-cm main pulmonary artery. In the 1950s, APVS was thought to be part of the spectrum including tetralogy of Fallot starting with the article by Rowe et al. [7] in 1955 describing a patient who was thought to have tetralogy of Fallot but had massively enlarged central pulmonary arteries that reverted to normal size at the hilum. By 1969, Durnin et al. [8] had collected 14 patients with absence of the pulmonary valve and ventricular septal defects, still referred to as tetralogy of Fallot variants. At this point, mortality for repaired tetralogy of Fallot was low, but 9 of these 14 APVS patients died. That paper's discussion makes no mention of airway disease. In 1976, Emmanoulides et al. [9] first noted agenesis of the ductus arteriosus as a common finding in patients with absent pulmonary valve, and hypothesized that the resultant in utero elevated right heart pressure from the absent ductus prevents development of a normal pulmonic valve. In the early 1970s, an association between APVS and DiGeorge syndrome was noted by Rose et al. [10]. Eventually, chromosome 22, which is responsible for DiGeorge syndrome and resultant hypocalcemia, was found in many of the patients with APVS. It is now confirmed that the syndrome is highly associated with DiGeorge syndrome and chromosome 22q11 deletions and can also have associated hypocalcemia [13]. Interestingly, the 11-year-old patient of Kurtz et al. [6] was noted to have an increased incidence of infections, raising the possibility of DiGeorge syndrome in that patient. In the 1980s and early 1990s, autopsy studies detailing the inherent abnor-

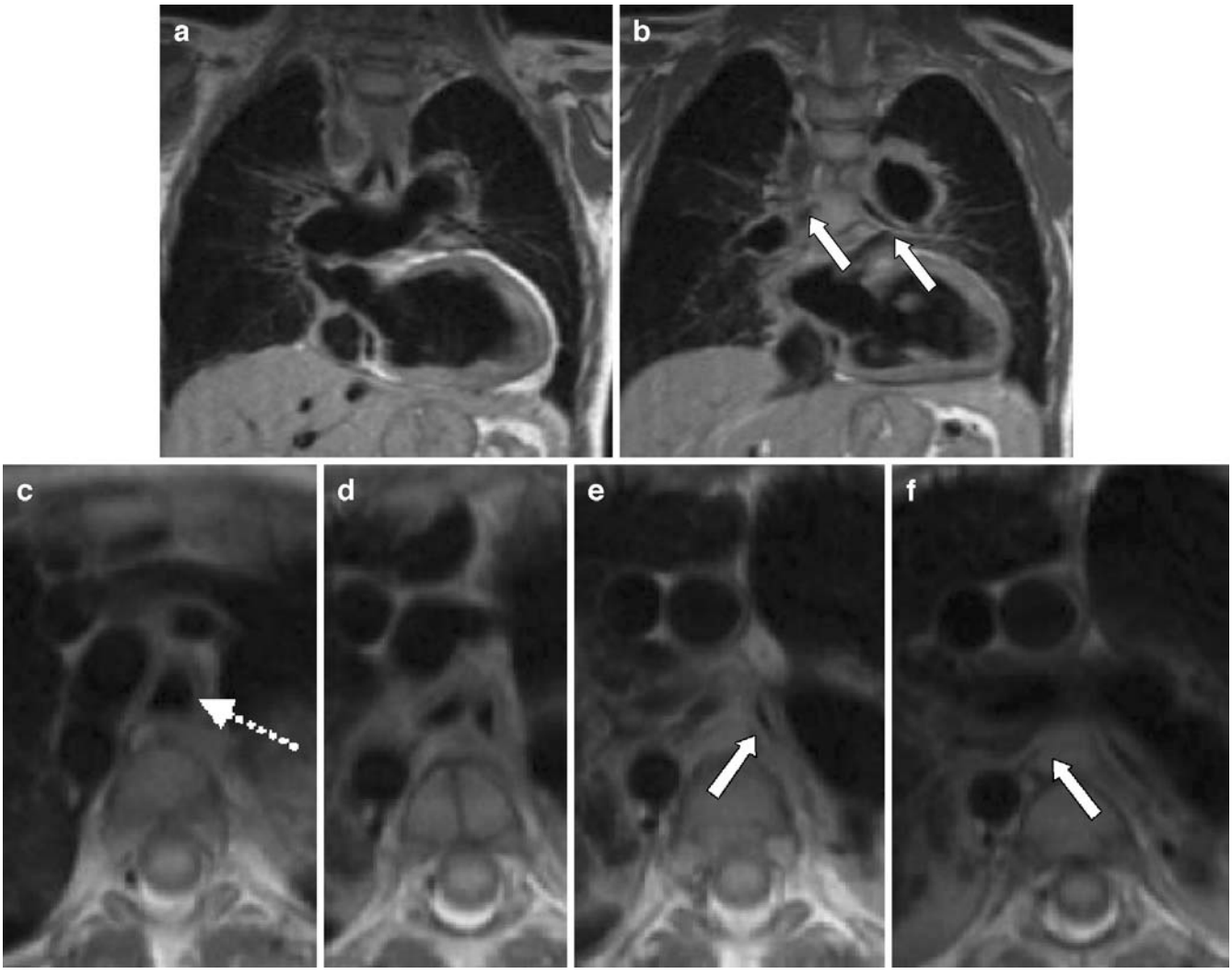

Fig. 5 Selected coronal (**a**, **b**) and sequential axial (**c**, **d**, **e**, **f**) MR images show the normal-caliber trachea (*dashed arrow*) and carina with marked narrowing of the right and left mainstem bronchi (*solid white arrows*). Additionally, the absence of a normal thymus is noted. Incidentally noted are a right-side aortic arch and a butterfly vertebral body

malities of the bronchi begin to appear [11]. In 1982, Rabinovitch et al. [11] described abnormal peripheral bronchial generations and decreased alveolar multiplication in patients with APVS. In 1990, Momma et al. [12] hypothesized and proved that a teratogenic agent, *N*, *N*′-bis-(dichloroacetyl)-1,8-octamethylenediamine (bis-diamine), which inhibits the normal action of neural crest cells in cardiovascular organogenesis, would cause absent pulmonary valve syndrome and absence of the ductus arteriosus, creating a rat model for this disease (Fig. 6).

Currently, patients with APVS are often diagnosed in utero, with prenatal US revealing aneurysmal dilatation of the pulmonary artery [13, 14]. After delivery, conventional preoperative radiographs show huge, massively dilated pulmonary arteries. Some patients develop obstructive emphysema and adjacent areas of atelectasis. There might be fluid retention noted on the initial neonatal chest radiograph [15].

Some infants with less severe airway disease survive with intubation, but many of these patients are difficult to extubate. A subgroup of these patients might eventually require tracheotomies, which do not alleviate the distal airway disease. Another group of patients might survive the neonatal period unaffected only to present later in life with airway difficulties and bronchospasm. Recently, Zucker et al. [16] have divided these patients, based on the presence of an intact ventricular septum, into an early presenting cohort and a cohort that presents later in life [16].

APVS is best thought of as a syndrome that has its own intricacies. Specifically, the inherent bronchial compression of APVS is distinct and can involve the mainstem bronchi well out into the hilar regions. Many patients are noted postoperatively to have difficulty ventilating or remain ventilator-dependent. This inherent airway disease is not seen in tetralogy of Fallot. Thus, though rare, APVS deserves mention as a vascular anomaly with associated bron-

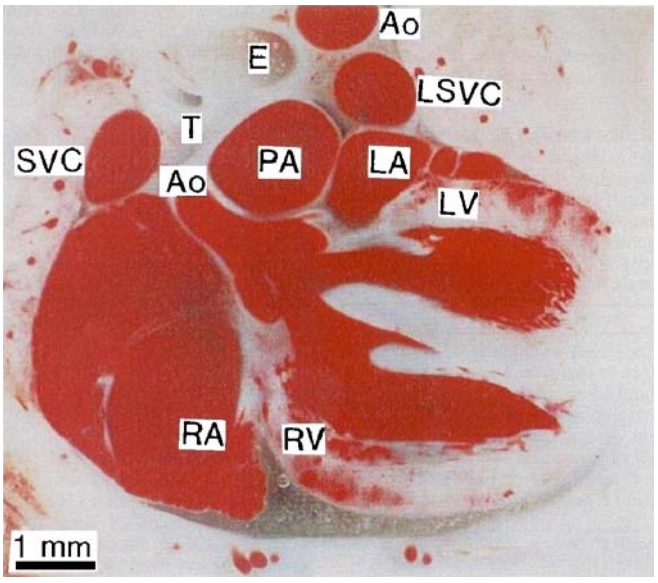

Fig. 6 Color photograph of a rat heart with teratogenic APVS. Note the massively dilated pulmonary artery (*PA*), in comparison to the adjacent aorta (reprinted with permission from ref. 10)

chial compression causing respiratory difficulties. It carries a significant risk of immediate mortality and morbidity.

This is in contrast to vascular rings, where surgical repair of the constriction relieves the airway symptoms, as the tracheal bronchial tree is inherently normal. In this group, mortality and morbidity is negligible. But it is also different from pulmonary slings (anomalous left pulmonary artery), which may be associated with complete cartilaginous tracheal and bronchial rings and is also associated with significant morbidity and mortality.

In APVS, surgical correction of the pulmonary insufficiency and valvular disease with limited reduction of the central arteries might ignore the distal pulmonary artery enlargement and resultant bronchial compression. Therefore, many pediatric cardiac surgeons recognize the importance of more extensive reduction plasty of the pulmonary arteries as far away from the hilum as possible as part of the same repair [2]. Others suggest a different approach—to correct the airway compression by translocation of the pulmonary artery anterior to the aorta and away from the airways. This technique has the potential to reduce or eliminate bronchial compression by the pulmonary artery [4]. A few papers and reports have advocated the use of airway stents to treat the bronchial compression [3].

In conclusion, pediatric cardiologists and radiologists performing MRI for evaluation of the postoperative cardiac anatomy and function must remember to assess inherent airway disease. The diagnosis of bronchial compression is often difficult on plain films and cannot be assessed on echocardiograms; however, it is often easily accomplished by cross-sectional imaging. In patients with APVS and ongoing respiratory difficulties, dedicated airway assessment should be performed. This can be obtained either by CT scanning or MRI [17]. Although both can provide excellent cross-sectional imaging and reconstruction techniques, MRI does not involve radiation and is better for assessing the patient's cardiac disease.

References

1. Knauth AL, Marshall AC, Geva T, et al (2004) Respiratory symptoms secondary to aortopulmonary collateral vessels in tetralogy of Fallot absent pulmonary valve syndrome. Am J Cardiol 93:503–505
2. Berlinger NT, Lucas RV, Foker J (1984) Pulmonary arteriopexy to relieve tracheobronchial compression by dilated pulmonary arteries. Ann Otol Rhinol Laryngol 93:473–476
3. Subramanian V, Anstead M, Cottrill CM, et al (1997) Tetralogy of Fallot with absent pulmonary valve and bronchial compression: treatment with endobronchial stents. J Pediatr Cardiol 18: 237–239
4. Hraska V (2005) Repair of tetralogy of Fallot with absent pulmonary valve using a new approach. Semin Thorac Cardiovasc Surg Pediatr Card Surg Annu 132–134
5. Chevers NA (1847) Collection of facts illustrative of the morbid conditions of the pulmonary artery, as bearing upon the treatment of cardiac and pulmonary diseases. Lond Med Gaz 38:828–835
6. Kurtz CM, Sprague HP, White PD (1927) Congenital heart disease: intraventricular septal defects with associated anomalies in a series of three cases examined postmortem and a living patient, 58-year-old with cyanosis and clubbing of the fingers. Am Heart J 3:77–90
7. Rowe RD, Vlad P, Keith JD (1955) Atypical tetralogy of Fallot: a noncyanotic form with increased lung vascularity; report of four cases. Circulation 12:230–238
8. Durnin RE, Willner R, Virmani S, et al (1969) Pulmonary regurgitation with ventricular septal defect and pulmonic stenosis —tetralogy of Fallot variant. Am J Roentgenol Radium Ther Nucl Med 106:42–51
9. Emmanoulides GC, Thanopoulos B, Siassi B, et al (1976) 'Agenesis' of ductus arteriosus associated with the syndrome of tetralogy of Fallot and absent pulmonary valve. Am J Cardiol 37:403–409
10. Rose JS, Levin DC, Goldstein S, et al (1974) Congenital absence of the pulmonary valve associated with congenital aplasia of the thymus (DiGeorge's syndrome). Am J Roentgenol Radium Ther Nucl Med 122:97–102
11. Rabinovitch M, Grady S, David I, et al (1982) Compression of intrapulmonary bronchi by abnormally branching pulmonary arteries associated with absent pulmonary valves. Am J Cardiol 50:804–813
12. Momma K, Ando M, Takao A (1990) Fetal cardiac morphology of tetralogy of Fallot with absent pulmonary valve in the rat. Circulation 82:1343–1351
13. Volpe P, Paladini D, Marasini M, et al (2004) Characteristics, associations and outcome of absent pulmonary valve syndrome in the fetus. Ultrasound Obstet Gynecol 24:623–628
14. Razavi RS, Sharland GK, Simpson JM (2003) Prenatal diagnosis by echocardiogram and outcome of absent pulmonary valve syndrome. Am J Cardiol 91:429–432
15. Fink AM, Edis B, Massie J (2005) The CT appearances of delayed amniotic fluid clearance from the lungs in an infant with absent pulmonary valve and congenital lobar emphysema. Pediatr Radiol 35:891–894
16. Zucker N, Rozin I, Levitas A, et al (2004) Clinical presentation, natural history, and outcome of patients with the absent pulmonary valve syndrome. Cardiol Young 14:402–408
17. Donnelly LF, Fleck RJ, Pacharn P, et al (2002) Aberrant subclavian arteries: cross-sectional imaging findings in infants and children referred for evaluation of extrinsic airway compression. AJR 178:1269–1274