Pulmonary atresia without cyanosis

Report of two cases with ventricular septal defect and increased pulmonary blood flow

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Two children with pulmonary atresia, ventricular septal defect, and increased pulmonary blood flow are presented. The absence of clinical cyanosis and polycythaemia in children with loud continuous murmurs and bounding pulses suggested the diagnosis of persistent ductus arteriosus. However, the absence of a main pulmonary artery segment and the presence of right ventricular hypertrophy prompted further evaluation. Both children had pulmonary atresia, ventricular septal defect, and a large anomalous vessel between the systemic and pulmonary circulations. One child also had a form of dextrocardia. The importance of recognizing this condition and the comparison of these two children with the more typical patients with pulmonary atresia are discussed.

In patients with pulmonary atresia and ventricular septal defect, severe cyanosis usually is present, and death commonly occurs before the age of 1 year (Keith, Rowe, and Vlad, 1958; Nadas, 1963; Gasul, Arcilla, and Lev, 1966). In a few patients, however, the bronchial collateral circulation is enlarged, and under these circumstances this anomaly may be compatible with a relatively long survival, even without palliative operation (Bach, 1928; East and Barnard, 1938). In addition, occasionally a persistent ductus arteriosus (Ongley *et al.*, 1966) or an anomaly of the aortic arch (Wheeler and Abbott, 1928) may result in prolonged survival, though in the majority of cases the ductus arteriosus is closed (Schad, Kunzler, and Onat, 1966). In addition to enlarged bronchial arteries originating from the aorta, dilated anomalous bronchial vessels may sometimes arise from the subclavian, internal mammary, or superior intercostal arteries (O'Rahilly, Debson, and King, 1950; Wagenvoort, Heath, and Edwards, 1964; Greenspon and Leaman, 1939). Despite obvious enlargement of these anomalous vessels, however, the clinical picture has usually been characterized by cyanosis and

symptoms of decreased pulmonary blood flow (Tynan and Gleeson, 1966).

The present report describes two children with pulmonary atresia who were not cyanosed and who were found to have increased pulmonary blood flow. The clinical and haemodynamic significance of this syndrome is discussed.

Case histories

Case 1 A white boy was referred to the National Heart Institute at age 13 months for evaluation of a murmur and poor development. The infant was the product of a normal, full-term pregnancy. There were no difficulties in the neonatal period, and the parents were first told of a cardiac murmur when the child was 8 months of age. At 9 months of age, weight loss and fatigue on exertion were noted, and though weight gain resumed subsequently, the infant remained below the third percentile in weight. Subsequently, except for occasional episodes of tachypnoea, there was no evidence of congestive heart failure, and cyanosis was never noticed.

On physical examination, the infant was small, well-proportioned, and without cyanosis or clubbing. The pulses were bounding in all extremities. The jugular venous pulse was normal and carotid upstroke was brisk. The point of maximal impulse was in the sixth left intercostal space, just inside the anterior axillary line, and the left and right ventricular impulses were prominent. A soft systolic thrill was palpable over the upper and mid-right sternal border. The first heart sound was normal, and the second sound was single. A loud systolic ejection click was heard along the left

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sternal border and a fourth heart sound was present at the apex. There was a grade 2/6 systolic ejection murmur, loudest at the upper left sternal border with minimal radiation, and a grade 4/6 continuous murmur over the entire praecordium, loudest at the upper and mid right sternal border. A grade 2/6 mid-diastolic rumble was present at the apex.

The haemoglobin was 12.9 g./100 ml. The electrocardiogram showed a mean QRS frontal plane vector of $+90^\circ$, biventricular hypertrophy, with left atrial and right atrial hypertrophy. The chest radiograph showed cardiomegaly, both ventricles and the left atrium being enlarged. The aortic arch was on the right side. The pulmonary vascularity appeared to be increased.

At cardiac catheterization, the femoral arterial oxygen saturation was 84 per cent. The catheter passed from the right ventricle across a ventricular septal defect into the aorta. The right and left ventricular pressures were equal and a bidirectional shunt was present at the ventricular level. A patent foramen ovale was crossed but there was no evidence of an interatrial shunt. The pulmonary/systemic flow ratio was 1.2:1 and the mean pulmonary venous wedge pressure was 30 mm. Hg. On biplane angiography, the right ventricular outflow tract formed a blind pocket (Fig. 1) and the pulmonary artery filled from the aorta through bronchial collateral vessels. One greatly enlarged collateral vessel arose from the right subclavian artery, followed the usual course of the internal mammary artery, and entered the right pulmonary artery near the hilum (Fig. 1). Contrast medium traversing this vessel filled the entire right lung and the left upper lobe, the left lower lobe being less well visualized. The right and left pulmonary arteries were well outlined and the presence of a main pulmonary artery was suspected on one frame of the angiogram. The infant was subsequently placed on digoxin and has been followed in the outpatient clinic. At 22 months of age, he continues to do well, with no increase in his symptoms.

Case 2 A 4½-year-old white girl was admitted for evaluation of a cardiac murmur present since birth. There was a history of frequent upper respiratory infections, and episodes of pneumonia had occurred at 2 and 3 years of age. Cyanosis had never been noted. Weight gain had been poor in the neonatal period, there was easy fatigability, and at 7 months of age the patient was placed on digitalis for congestive heart failure. The child's activity subsequently improved though she remained well below the third percentile in height and weight.

On physical examination, the child was small and there was no cyanosis or clubbing. The arterial pulses were full, and the jugular venous pulse appeared normal. There was no praecordial bulge. A faint systolic thrill was present over the mid-left sternal border, and the point of maximal impulse was in the fourth right intercostal space just beyond the mid-clavicular line. The first sound was normal and the second heart sound was loud and single. A fourth heart sound was present at the

FIG. 1 (Case 1) In panel A, contrast material is injected into the right ventricle (RV) in the PA projection. Simultaneous filling of an atretic infundibular area (Atr. Outflow) and the aorta (Ao) is seen.

In panel B, the posterior sweep of the ascending aorta (Ao) is clear, ruling out types I, II, or III truncus arteriosus. A bronchial collateral vessel can be identified.

In panel C, contrast material is filling a large collateral vessel apparently arising from the subclavian artery. This vessel subsequently emptied into the right pulmonary artery at the hilar region.

apex, and a loud systolic ejection murmur that did not radiate was heard at the base. A grade 4/6 continuous murmur was audible over the entire praecordium and back, loudest over the mid-left sternal border. A grade 1/6 mid-diastolic rumble was present at the cardiac apex.

The haemoglobin was 14.9 g./100 ml.; there were no Howell-Jolly bodies on peripheral blood

smear. The electrocardiogram exhibited a mean QRS vector of $+85^\circ$ in the frontal plane. Biventricular hypertrophy was present, left ventricular complexes being seen in leads V₄R and V₅R. The P waves were normal in one electrocardiogram and showed a pattern of P mitrale with a P wave axis of -90° in another. The chest x-ray showed dextrocardia¹, cardiomegaly, and a left-sided aortic arch. The pulmonary vascularity appeared moderately increased. The abdominal organs were in normal position and a splenic shadow was identified on the left side.

At cardiac catheterization performed under moderate sedation, the femoral arterial oxygen saturation was 81 per cent. The right and left ventricular pressures were equal, and there was a bidirectional shunt at the ventricular level. The left atrium was entered through a patent foramen ovale, and there was no interatrial shunt. The pulmonary/systemic flow ratio was 1.3:1 and the mean pulmonary venous wedge pressure was 25 mm. Hg. Biplane angiography showed the atria to be normally positioned and the ventricles to be inverted, the right atrium emptying into the left ventricle and the left atrium into the right ventricle. The aorta filled from both right and left ventricular injections of contrast medium, but there was no visualization of the pulmonary arteries until after filling of the aortic arch. Several bronchial collateral vessels were identified, and one greatly dilated collateral vessel originated from the descending aorta and entered the left pulmonary artery at the hilum (Fig. 2). This vessel also provided a branch to the right pulmonary artery, but the main pulmonary artery was not visualized. The child was discharged on digoxin to be followed in the clinic, and has continued to maintain a satisfactory status.

Discussion

Pulmonary atresia with ventricular septal defect ('pseudo-truncus arteriosus') is usually classified as a variant of severe tetrad of Fallot (Edwards *et al.*, 1965). Children born with this defect usually have their pulmonary blood flow supplied through a persistent ductus arteriosus, and closure of this pathway undoubtedly accounts for a significant number of early deaths (Taussig, 1960). Occasionally, the ductus may persist, but in most patients only the enlarged bronchial collateral circulation provides the pulmonary blood flow. Anomalous bronchial arterial vessels have been described at necropsy in subjects without cardiac or pulmonary disease (Cauldwell *et al.*, 1948). Therefore, it is possible that the enlarged vessels described in the present report represent either a specific congenital malformation or exaggeration of an anomaly that also occurs sporadically in otherwise normal subjects.

¹ The term dextrocardia is used only to signify a heart in the right hemithorax.

FIG. 2 (Case 2) In panel A, injection of contrast material is made into the inferior vena cava (IVC) and fills the right atrium and a ventricle that is relatively smooth walled. The ventricle ends in a smooth atric outflow area and contrast enters the left-sided aorta (Ao).

In panel B, injection is made into the arch of the aorta (Ao) and shows filling of the right and left pulmonary arteries (RPA, LPA) via a huge collateral vessel arising from the mid-thoracic aorta and feeding into the left pulmonary artery.

The Table summarizes the clinical, electrocardiographic, and radiological features that characterized these two clinically acyanotic patients, and contrasts them with the findings in the typical patient with pulmonary atresia and cyanosis. The major differences relate to pulmonary blood flow, degree of polycythaemia, and left atrial/ventricular size. On physical examination, the continuous murmur was more precisely localized, a mid-diastolic rumble was present, and the peripheral pulses were bounding.

Several considerations deserve mention relative to the diagnosis and management of patients with pulmonary atresia without cyanosis. The presence in these patients of features suggestive of a persisting ductus arteriosus might have led to attempted ligation without earlier cardiac catheterization.

TABLE Comparison of patients with and without cyanosis and pulmonary atresia

Findings	Case 1	Case 2	Patients with cyanosis
Failure to thrive	+	+	+
Congestive heart failure	± (9 mth.)	± (7 mth.)	o
Cyanosis, clubbing	o	o	+
Haemoglobin (g./100 ml.)	12.9	14.9	Moderate to severe polycythaemia
Electrocardiogram	Right and left ventricular hypertrophy; right and left atrial enlargement; mean QRS = +90	Right and left ventricular hypertrophy; left atrial enlargement; mean QRS = +85; dextrocardia	Right ventricular hypertrophy; right atrial enlargement; right axis deviation
Chest radiograph	Right and left ventricular hypertrophy; left atrial enlargement; right-sided aortic arch; absent main pulmonary artery with increased pulmonary vascularity	Right and left ventricular hypertrophy; left atrial enlargement; dextrocardia with left-sided aortic arch; absent main pulmonary artery with increased pulmonary vascularity	Right ventricular hypertrophy; right atrial enlargement; about 25 per cent have right-sided aortic arch; absent main pulmonary artery with decreased pulmonary vascularity
Cardiac examination	4/6 continuous murmur mid-right sternal border; single S2; loud aortic ejection click; mid-diastolic rumble; bounding pulses	4/6 continuous murmur mid-left sternal border; single S2; loud aortic ejection click; mid-diastolic rumble; bounding pulses	No significant murmur or soft diffuse continuous murmur; single S2; occasional aortic ejection click; normal pulses

+ = present; o = absent; ± = equivocal.

However, the atypical location of the continuous murmur and absence of the pulmonary second sound should offer clues to the correct diagnosis or at least stimulate investigation before operative referral. The development of pulmonary arterial hypertension appears unlikely, though the clinical histories suggested that the shunt through the large collateral vessels had led to early congestive failure and pulmonary infections. Ongley *et al.* (1966) emphasized the importance of recognizing tetrad of Fallot and pulmonary atresia with ventricular septal defect when these malformations occur in patients with cyanosis and a continuous murmur. These are both potentially correctable lesions, and the natural history of acyanotic patients like the two reported here seems associated with a lower morbidity and mortality than either truncus arteriosus or a severe tetrad of Fallot. The need for early operation or palliation would probably be less likely and total correction could often be carried out at an older age.

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