ISOLATED RIGHT VENTRICULAR HYPOPLASIA WITH ATRIAL SEPTAL DEFECT

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Isolated Hypoplasia of the right ventricle is a rare congenital cardiac anomaly which has been infrequently reported in the literature. Less than fifteen cases have so far been recorded. We describe here the clinical, haemodynamic and angiographic findings in two cases, and briefly review the literature.

Case Reports

Case 1

J.F. was the second child of healthy parents; she was a full term normal hospital delivery, following a normal pregnancy. There was no difficulty in onset of respiration. B.W. 5 lb. 13 oz. She had two cyanotic attacks on the first day and was noted to be cyanosed on crying during the first week of life. On general examination she was a peculiar looking baby with a small head (circumference 12½″), receding brow, and long thin limbs with long fingers and toes. There were no heart murmurs and the peripheral pulses were normal. X-ray of the chest was normal and the ECG showed left ventricular hypertrophy. After the first day she had no further cyanotic attacks.

She subsequently thrived, but showed slight cyanosis at rest and a heart murmur was first noted at 10 months of age.

She was referred to the Brompton Hospital and at 2 years of age was admitted for further investigation. Her general condition was good. There was probable cyanosis at rest, which became obvious on crying; there was no differential cyanosis. The peripheral pulses were normal. The blood pressure was 80 mm Hg by palpation in upper limbs. The fingers and toes were clubbed. There was no chest bulge and no thrill. The heart was quiet with a grade of 1-2/6 ejection murmur best heard at the second left intercostal space. The second sound was single. The lungs were clear on auscultation and the liver was not palpable.

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Table 1. Haemodynamic data of Case 1 and Case 2

	CASE 1		CASE 2	
	Pressure mm.Hg.	O_2 Satn. %	Pressure mm.Hg.	O_2 Satn. %
Mixed Venous	-	58		49
Right Atrium	a=6 v=5	56	a=7 x=4 v=5 y=4 mean=5	47
Right Ventricle	11/0-5 presystolic wave=6	56	13/2-6 presystolic wave=7	45
Pulmonary Artery	11/5	56	-	-
Wedge	mean=7		-	-
Pulmonary Vein	-	96	-	98
Left Atrium	mean=2	87	a=4 x=2 v=3 mean=3	75-84
Left Ventricle	70/0-3	81-90	65/6-7	73-80
Axillary Artery	75/55	81	63/46	80
QP	2.4 L./min./Sq.M.		1.4 L./min./Sq.M.	
QS	3.3 L./min./Sq.M.		2.2 L./min./Sq.M.	
QP/QS	0.73		0.64	

Investigation: Hb 14.6 g%, P.C.V. 53%. Chest X-ray showed slight to moderate cardiomegaly with some probable reduction in lung vascularity. E.C.G. Axis + 100° and an adult R/S pattern, indicative of mild left ventricular hypertrophy. A clinical diagnosis of tricuspid atresia was made.

Cardiac catheterization: (right axillary approach) was carried out under general anaesthesia, breathing 25% oxygen. Both right and left pulmonary arteries were entered. There was a presystolic wave in the R.V. pressure tracing equal to the right atrial 'a' wave and a narrow ejection peak of the right ventricular pressure curve. *(Table 1)* The presystolic wave was also seen in the pulmonary artery pressure curve. The left atrium was entered from the right atrium, and the left ventricle from the left atrium. Oxygen saturation data revealed systemic arterial desaturation (81%) and a right to left shunt at atrial level.

Selective angiocardiography, with injection of 75% Triosil into the right atrium showed opacification of both the left atrium and an anteriorly placed right ventricle from which arose a normally situated pulmonary artery. The pulmonary valve was not well seen. *(Fig. 1)* Injection of contrast medium into the left ventricle showed no abnormality of this chamber or of the aorta, and no ventricular septal defect *(Fig. 2).*

Case 2

P.M. The mother had taken Nitrazepam in the last 4 months of pregnancy, which had been otherwise normal. He was the second of dissimilar twins, delivered by breech with forceps application to the after-coming head, at 39 weeks' gestation (pitocin drip for prolapse of cord). Birth weight 7 lb. His condition was poor at birth and he was placed in an incubator for three days. Cyanosis and a heart murmur were noted during the first few days. However, he subsequently thrived and development was normal.

A brother aged 6 years, a sister aged 8 years, and another brother and sister both aged 9 years (twins), besides his twin brother are all normal. His parents are healthy.

He was admitted to the Brompton Hospital for further assessment when aged 6 months. On examination he looked well with height (64 cm) and weight 6.300 kg) both around the 10th percentile. He was mildly cyanosed at rest but there was no clubbing. His peripheral pulses were normal. There was no heart murmur and the second sound was single and of normal intensity. The lungs were clear to auscultation and the liver was not enlarged. There was no other abnormality on routine examination. Relevant *investigations* showed Hb 16.5g%, P.C.V. 54 %. Chest X-ray showed normal size heart with diminished pulmonary vasculature. E.C.G. showed an indeterminate axis with normal P waves; adult R/S pattern indicative of left ventricular hypertrophy. A clinical diagnosis of tricuspid atresia was made.

At *cardiac catheterization* (Dr. G. A. H. Miller) the systemic saturation was 80%. The atrial septum was crossed by catheter and there was a right to left shunt at atrial level. There was again a presystolic wave transmitted to the R.V. The pulmonary artery was not entered. *(Table 1).*

Selective angiocardiography with injection of 75% Triosil into R.A. showed passage of contrast medium into a smooth-walled small anterior right ventricle, and thence into the pulmonary artery. The right outflow tract was not well seen. There was also some opacification of the left atrium. Injection of contrast medium into the left ventricle demonstrated no abnormality.

Discussion

Taussig (1936) drew attention to the association between defective development of the right ventricle and anomalous tricuspid and pulmonary valves. However, isolated hypoplasia of the right ventricle in the absence of valve atresia is very rare. The condition may be associated with hypoplasia or stenosis of the tricuspid valve, and an atrial septal defect or a patent foramen ovale is often present.

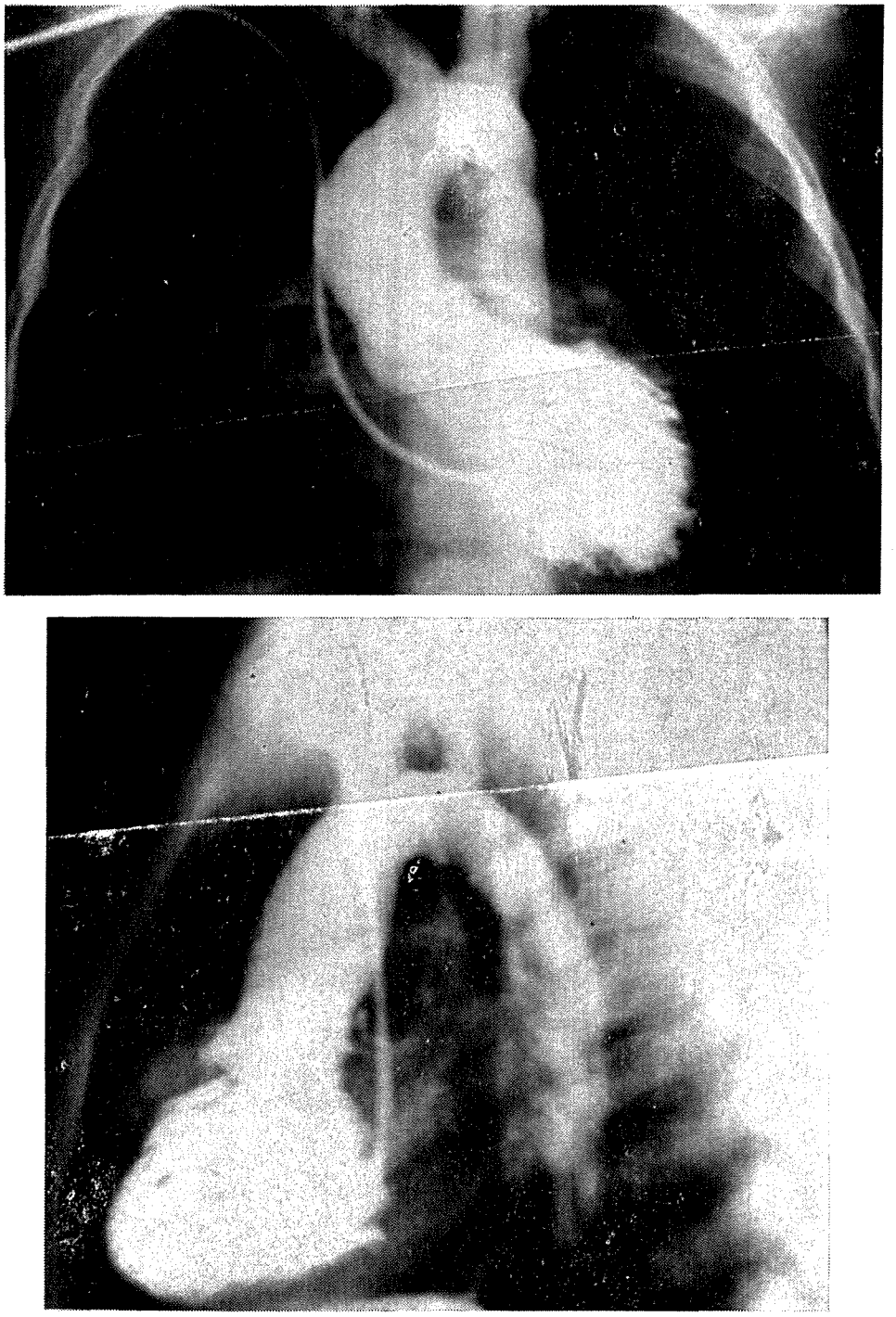

Fig. 1

Case 1

Angiogram with injection of contrast medium in the right atrium. There is opacification of right and left atrium, right ventricle, pulmonary artery and aorta.

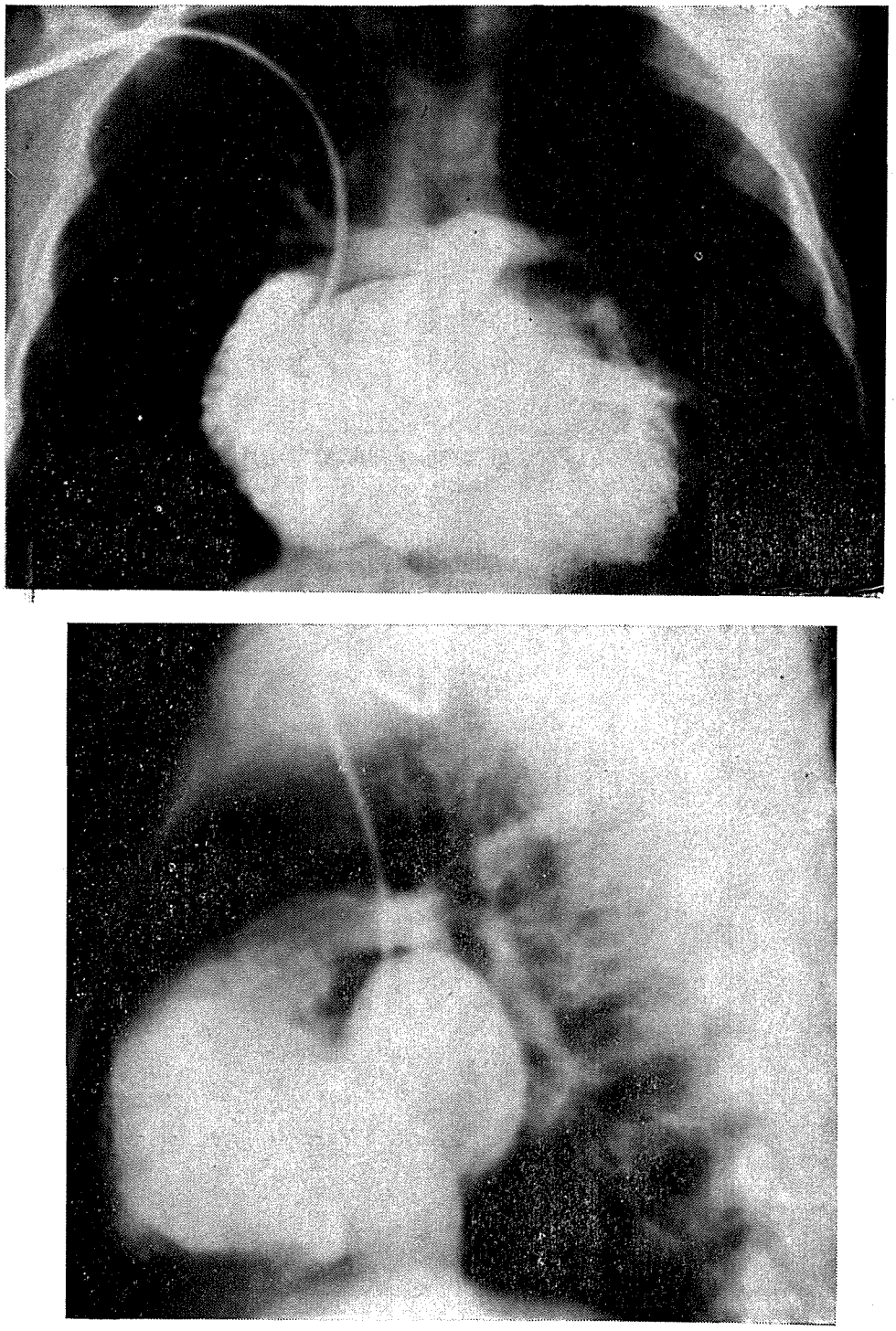

Fig. 2

Case 1

Angiogram with injection of contrast medium in left ventricle. Normal appearances.

Cooley *et al.* (1950) diagnosed isolated hypoplasia of the right ventricle on angiographic appearances, which was confirmed at necropsy. Gasul *et al.* (1959) diagnosed a similar case at thoracotomy for a Glenn procedure on a child with suspected tricuspid stenosis. Medd *et al.* (1961) gave the detailed anatomical findings at necropsy in two siblings with isolated hypoplasia of the right ventricle and tricuspid valve. Sachner *et al.* (1961) described the clinical and haemodynamic features in three adults with this condition, all in the same pedigree, and presenting with progressive right heart failure. They also gave the necropsy findings in a two month old infant. Fay and Lynn (1963) reported a six year old with hypoplastic right ventricle associated with atrial septal defect and supravalvar pulmonary artery stenosis; there was marked clinical improvement following anastomosis of the right pulmonary artery to the superior vena cava (Glenn procedure). Stoerner and Apitz (1965) gave the clinical, haemodynamic and angiographic findings in a 4 year old boy with this anomaly, a Blalock-Taussig anastomosis was performed but he died a few hours after operation. Raghib *et al.* (1965) reported a male infant with hypoplasia of the right ventricle and tricuspid valve, and Davachi *et al.* (1967) described this patient's sister who died with an identical malformation. Overy *et al.* (1966) recorded a case of anomalous systemic venous drainage with hypoplasia of the right ventricular myocardium and right to left shunt at atrial level.

Aetiology

The aetiology is obscure. A familial factor has been prominent in some reported cases (Sachner *et al.*, 1961; Medd *et al.*, 1961; Davachi *et al.*, 1967). Wood (1958) noted that congenital heart disease recurring in more than one member of a family is usually of the same type. The siblings and parents of the cases reported here have no clinical evidence of congenital heart disease. A primary failure of development of the muscle of the free wall of the right ventricle has been postulated (Medd *et al.*, 1961). Overy *et al.* (1966) suggested that deviation of blood from SVC which normally flows to the RV in foetal life (as in their case of anomalous systemic venous drainage to left atrium) might result in underdevelopment of the right ventricular myocardium. This is similar to LV hypoplasia which occurs when premature narrowing or closure of the foramen ovale results in reduction of flow into the left ventricle (Lev *et al.*, 1963). The case reported by Cooley *et al.* (1950) was attributed to absence of the right coronary artery, though this was found to be normal in others. In Uhl's case (Uhl, 1952) there was almost total absence of the myocardium from the right ventricular wall, with marked dilation of the chamber, which contained a large laminated mural thrombus; microscopic examination of the right ventricular wall showed epicardium and endocardium adjacent to each other with no intervening cardiac muscle; the coronary arteries were normal. It is here suggested that the determing factor for development of the myocardium is the blood flow into the chamber during foetal life rather than the coronary artery supply.

Clinical Features

The clinical features resemble those of tricuspid atresia. Cyanosis is a constant feature and is often recorded as being present from birth. Heart murmurs are usually absent in the straightforward case of hypoplasia of the right ventricle with a patent foramen ovale or ASD, and when present an associated anomaly such as pulmonary stenosis or frank tricuspid incompetence should be suspected. The E.C.G. typically shows left axis deviation but the axis was + 110° in one case of Medd *et al.* (1961). The praecordial lead pattern is that of left ventricular dominance, and there is usually less positive deflection of the QRS complex over the right chest leads than one would expect. Indeed ,if in the presence of signs of pulmonary stenosis there is little or no evidence of RV activity in the ECG the diagnosis of hypoplasia of the right ventricle should be borne in mind. Bi-atrial or right atrial hypertrophy is a feature, and the

PR interval is usually prolonged due to right atrial dilatation. The chest X-ray shows a normal sized heart with underfilling of the lung fields. The presence of cardiomegaly, without other evidence of heart failure, would be in favour of a diagnosis of Uhl's anomaly (Aplasia of the myocardium of the right ventricle) (Uhl, 1952).

Catheterization and Angiocardiography

At cardiac catheterization the passage of the catheter from RA to RV excludes tricuspid atresia. The tricuspid valve is normally situated. The pressure trace may be similar in pulmonary artery, right ventricle and right atrium, with a presystolic wave, possibly transmitted as far as the pulmonary artery, and a narrow right ventricular ejection peak. A right to left shunt is demonstrable at atrial level. On angiocardiography, injection of the contrast medium in the right ventricle shows this chamber to be normally situated but usually small and the pulmonary artery may be normal or hypoplastic. Injection of the contrast medium in the right atrium will demonstrate the right to left shunt at this level. The left ventricle is normal and the ventricular septum is intact. In Uhl's anomaly the right ventricular cavity is usually enormous, but it may be difficult to distinguish from hypoplasia of the right ventricle if a clot has formed in the right ventricle thus diminishing the size of this cavity. In Ebstein's malformation the RV wall is normal or thin but the ventricular cavity is diminished by a downward displacement of the tricuspid valve.

Both the cases reported here presented with a clinical picture of tricuspid atresia. In both cases the correct diagnosis was suspected during cardiac catheterization from the characteristics of the pressure tracings.

The angiocardiographical appearances in both cases excluded malposition of the tricuspid valve; the size of the cavity of the right ventricle was compatible with hypoplasia of the muscle wall rather than the total aplasia first described by Uhl.

Gasul *et al.* (1959) carried out successful palliative surgery in one case, who survived, anastomosing the superior vena cava to the right main pulmonary artery (Glenn operation). If there is a concomitant atrial septal defect, this should be closed at the same time (Gasul *et al.*, 1966; Overy *et al.*, 1966) but heart failure may supervene. On the whole, treatment of this anomaly has been disappointing.

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