Isolated Right Ventricular Hypoplasia with Atrial Septal Defect or Patent Foramen Ovale

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In 1936, Taussig stated that a diminutive right ventricle is always associated with underdevelopment or malformation of the tricuspid or pulmonic valves. Four cases of hypoplastic right ventricle unassociated with valvular defects have been reported in recent years. A patent foramen ovale or atrial septal defect has been an accompanying anomaly. All cases have been described in cyanotic infants or young children. In 1950, Cooley and his associates made the diagnosis by angiocardiography and confirmed it at necropsy. In 1959, Gasul and co-workers established the diagnosis in a 4-year-old child at thoracotomy. In 1961, Medd and associates defined the pathologic features in two siblings who died during infancy.

We have observed three adults with this disorder who were members of the same family. The purpose of this paper is to describe the clinical and hemodynamic features that are sufficiently characteristic to enable a definitive diagnosis. In addition, a fourth case obtained from the postmortem files of the Philadelphia General Hospital is briefly reported for historic interest.

Case Report

A 2-month-old white boy was admitted to the Philadelphia General Hospital in September 1935 because of fever and dyspnea of 1 week's duration.

Physical examination revealed an acutely ill cyanotic infant in respiratory distress. The pulse was 160, the respirations 70, and the temperature 105 F. The weight was 4.6 Kg., the height 55 cm. Erysipelas was present on the face and scalp. Both ear drums were perforated and discharging pus. Rhonchi were heard in both lung fields. The heart was of normal size by percussion. The rhythm was regular, and no murmurs were heard. The hemoglobin was 12 Gm. per 100 ml. and the white cell count 4,800. There was 2+ proteinuria. The patient was treated with immunotransfusions but died 6 days after admission.

The clinical diagnoses were bilateral otitis media, erysipelas, and bronchopneumonia. The necropsy diagnoses were isolated hypoplasia of the right ventricle, erysipelas, and bronchopneumonia.

The aorta, vena cavae, and pulmonary veins were normal. The ductus arteriosus was closed. The pulmonary artery was underdeveloped.

The heart weighed 20 Gm. (fig. 1). The right ventricular chamber was small: its lowermost portion reached less than one half the distance from the base to the true cardiac apex. The distance from the base of the tricuspid valve to the lowermost portion of the right ventricle was 1.2 cm. The distance from the base of the mitral valve to the lowermost portion of the left ventricle was 2.9 cm. The tricuspid valve was small, but of normal shape and insertion. The right atrium was dilated. The left ventricle showed hypertrophy. The foramen ovale was probe-patent.

Cases in Family E

The family pedigree is depicted in figure 2. Unfortunately, the unwillingness of many family members to be examined prevented full-scale genetic investigation. As far as can be determined, no consanguinity existed nor were there any abnormal pregnancies.

Case II-1

F.E., a 30-year-old Negro laborer was in good health until May 1953, when he fainted after a hot day's work in a coal mine. A company physician found a blood pressure of 120/95 and a pulse of 80. The cardiac impulse was diffuse with maximum intensity in the fourth interspace at the
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anterior axillary line. The rhythm was regular but frequent extrasystoles were present. There were blowing aortic and apical systolic murmurs. The hemoglobin was 18 Gm. per 100 ml., and the white cell count was normal. The urine showed 3+ proteinuria. The chest roentgenogram and electrocardiogram were consistent with left ventricular enlargement. A diagnosis of aortic stenosis was made and the patient was not allowed to return to work.

Subsequently, despite moderately strenuous work as a porter, he remained asymptomatic until November 1958. At this time, he noted the gradual onset of leg swelling. He was admitted to the Philadelphia General Hospital in December 1958. He denied dyspnea, orthopnea, hemoptysis, and chest pain. He had no complaints referable to the gastrointestinal or genitourinary systems. He denied rheumatic fever and syphilis.

On physical examination, except for marked pitting edema extending up to the level of the eleventh thoracic vertebra, he did not appear ill and had no orthopnea. The blood pressure was 110/80, the pulse 80, respirations 22, and temperature 98 F. The weight was 89 Kg., the height 200 cm., the arm span 206 cm., and the upper segment-to-lower segment ratio 0.89 (normal, 0.93; Marfan's gene, 0.85.9) The fingers were long and tapered. The nailbeds, lips, and mucous membranes were moderately cyanotic. The cervical veins were distended at 90° and filled from below. The lungs were clear. The heart was greatly enlarged to the left, the point of maximum impulse was at the sixth interspace in the anterior axillary line. The rhythm was regular, with a few extrasystoles. There were a presystolic gallop over the entire precordium, a soft nonradiating ejection murmur at the aortic area, and a systolic click over the remaining precordium. The liver was tender and palpable 5 cm. below the costal margin. Peripheral pulses were normal.

The patient was treated with hydrochlorothiazide, digoxin, and procaine amide and became edema free after 3 weeks. He had lost 24 Kg. of edema fluid. Following this diuresis, cardiac catheterizations were performed (table 1).

Throughout the hospital course, the hemoglobin ranged from 17 to 18 Gm. per 100 ml. The white cell count and differential were normal. Total serum protein was 4.8 Gm. on admission but rose to 6.2 Gm. per 100 ml. after diuresis. Paper electrophoresis showed an albumin of 2.7 Gm. and globulin of 3.5 Gm. with normal fractions. There was a persistent proteinuria ranging from 1.0 to 3.5 Gm. daily.

The electrocardiogram showed prolonged P-R interval, occasional premature ventricular contractions, bialtrial enlargement, and left ventricular

Figure 1
The dissected heart of case J.E. The right ventricular chamber is diminutive yet the myocardium is of normal thickness; the left ventricle is moderately hypertrophied (RA, right atrium; TV, tricuspid valve; RV, right ventricle; LA, left atrium; MV, mitral valve; LV, left ventricle).
hypertrophy. The mean QRS axis was -50° (fig. 3). The chest roentgenogram showed an enlarged, globular heart and increased vascular markings with a normal pulmonary conus (fig. 4).

The patient refused surgery and was discharged to the outpatient clinic after 8 weeks of hospitalization. He returned to work and did well except for transient peripheral edema that responded to mercurial injections. He denied dyspnea and orthopnea, and his lung fields remained clear of rales.

In March 1960, intractable ascites and peripheral edema developed. Also, he noted weakness, myalgia, and blurred vision. He was readmitted to the hospital with the diagnoses of digitalis toxicity and hypopotassemia.

Physical examination was little changed from the previous admission except for slow atrial fibrillation (40 per minute), left pleural effusion, ascites, and more intense cyanosis.

The blood urea nitrogen was 119 mg., the serum creatinine 3.8 Gm., and the uric acid greater than 10 mg. per 100 ml. Serum potassium was 2.5 mEq. per liter.

The electrocardiogram showed atrial fibrillation, multifocal premature ventricular contractions, and short runs of ventricular tachycardia. The withdrawal of digitalis and intravenous administration of potassium chloride improved the arrhythmia. Despite thoracentesis and vigorous attempts at diuresis, the patient remained in severe rightsided congestive heart failure. Three weeks after admission, he complained of a suffocating feeling and became noticeably dyspneic for the first time. He was found dead in bed the next day.

The clinical diagnosis was atrial septal defect and idiopathic enlargement of the left ventricle. The necropsy diagnoses were isolated hypoplasia of the right ventricle, atrial septal defect, mild aortic stenosis due to fibrosis and calcification, left ventricular hypertrophy, left ventricular endocardial fibroelastosis, congestive heart failure, and bronchopneumonia.

The aorta, venae cavae, pulmonary veins, and coronary vessels were normal. The pulmonary artery was slightly narrowed.

The heart weighed 650 Gm. (fig. 5). The right ventricle was small, its lowermost portion reached less than one half the distance from the base to the true cardiac apex. The chamber proportions are depicted schematically in figure 6. The tricuspid valve was normal in shape and position; there was functional dilatation of the ring. The aortic valve was distorted by fusion of all the commissures, fibrous thickening of the free and closing margins of the cusps, and irregular calcified nodules between the cusps; the valve area was 1.9 cm.² The endocardial surface of the left ventricular wall was dull gray, and histologic examination.

### Table 1

<table>
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<tr>
<th>Case</th>
<th>Age (Years)</th>
<th>Sex</th>
<th>Race</th>
<th>Pressure (mm, Hg)</th>
<th>RV, PA</th>
<th>SVC</th>
<th>IVC</th>
<th>Oxygen content (%)</th>
<th>LA</th>
<th>RA</th>
<th>PA</th>
<th>SVC</th>
<th>IVC</th>
<th>Arterial oxygen saturation</th>
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<td>M</td>
<td>White</td>
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<td>50 (18)</td>
<td>10</td>
<td>72</td>
<td>114 (82)</td>
<td>70</td>
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<td>13.88</td>
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<td>114 (82)</td>
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*Mean of multiple samples.

Abbreviations: LA, right atrium; RV, right ventricle; PA, pulmonary artery; PCV, pulmonary capillary venous; LA, left atrium; RA, brachial artery; IVC, inferior vena cava; SVC, superior vena cava; PV, pulmonary vein.
showed proliferation of fibroelastic tissue. The right ventricular endocardium was normal. The left ventricular wall measured up to 19 mm. in width; the right, 4 mm. Glycogen, amyloid, tri-chrome, mucopolysaccharide, fat, and elastic tissue stains of the myocardium were unremarkable.

Both atria were markedly dilated as were the atrial appendages, which were filled with adherent, partially organized thrombus. There was an oval, smooth-walled atrial septal defect, which measured 4.5 cm. in diameter.

All viscera showed marked passive congestion.

**Case II-4**

C.E., was a 22-year-old janitor who had cyanosis and easy fatigability dating from childhood. Nevertheless, he enjoyed fair health and always held a job. In July 1956, on a routine physical examination, a hemoglobin of 22 Gm. per 100 ml. was discovered. He was treated with monthly phlebotomies and was referred to the Charleston General Hospital in September 1956.

Physical examination revealed a cyanotic but otherwise healthy appearing man. The blood pressure was 130/100 and the pulse 96. The weight was 76 Kg., the height was 185 cm. The fingers were long and tapered; clubbing was present. The lungs were clear. The heart was enlarged to the left. The rhythm was regular. There were a moderately loud, holosystolic murmur in the third interspace parasternally, a short basal diastolic murmur, and an apical presystolic murmur or gallop. The liver was nontender and palpable just below the costal margin. There was no peripheral edema.

The hemoglobin was 22 Gm. per 100 ml., the hematocrit level 73 per cent, and the white cell count 7,000. The blood urea nitrogen was 17 mg. per 100 ml. There was persistent proteinuria, ranging from 1+ to 3+.

The electrocardiogram showed right atrial enlargement and left ventricular hypertrophy. The mean QRS axis was -45° (fig. 3). The chest roentgenogram showed moderate cardiac enlargement and a slight decrease in vascular markings (fig. 7). Cardiac catheterization (table 1) and venous angiocardiography (fig. 8) were carried out. The diagnosis of an atypical tricuspid atresia was made and the patient was discharged to await surgery.

In August 1957, the patient underwent thoracotomy. The right ventricle and pulmonary artery

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**Figure 2**

Pedigree of family E: I-1, T.E., age 60 years, worked all his life as a coal miner, never hospitalized; I-2, M.E., died at age 56 years, cause of death was uremia, had diabetes mellitus; II-1, propositus, F.E., died at age 39 years, right ventricular hypoplasia; II-2, G.E., age 35 years, employed as coal miner, served in Army; II-3, S.E., age 31 years, employed as coal miner, served in Army; II-4, C.E., died at age 22 years, right ventricular hypoplasia; II-5, E.E., age 23 years, good health; III-1, C.E., age 13 years, right ventricular hypoplasia; III-2, H.E., age 11 years, good health; III-3, age 5 years, good health; III-4, age 3 years, good health; III-5, age 1 year, good health.
Electrocardiograms of Family E: A-II-1, B-II-4 (the precordial leads are one-half standard), and C-III-1. There is a remarkable similarity: all show left axis deviation, atrial enlargement, left ventricular hypertrophy with “strain” pattern, and top normal or prolonged P-R interval.

were markedly underdeveloped, the atria and left ventricle were enlarged. The tricuspid and pulmonic valves were hypoplastic but not atretic, insufficient, or stenotic. A fenestrated atrial septal defect about 3 cm. in diameter was present. No corrective procedure was attempted. Postopera-
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tively, the patient did poorly. He had frequent episodes of pleuritis and bronchitis. He died at home in congestive heart failure in 3 months. No necropsy was obtained.

Case III-1

Ca. E. is a 13-year-old schoolgirl. At the age of 5 years, during examination for a foreign body in the auditory canal, a harsh holosystolic murmur was heard parasternally. Except for frequent attacks of tonsillitis and pharyngitis, the patient had been asymptomatic. No cyanosis, clubbing, or hepatomegaly was noted. The hemoglobin was 14 Gm. per 100 ml. The electrocardiogram and chest roentgenogram were suggestive of early left ventricular enlargement. The lung fields showed a slight increase in vascular markings. Cardiac catheterization (table 1) was performed, and a diagnosis of ventricular septal defect was made.

After a tonsillectomy at age 8 years, she had no further attacks of frequent respiratory infections. Her lips became blue on occasion and she had been on a program of limited physical activity. In September 1959, she began to participate in school gymnastics and noted easy fatigue compared to her classmates. In January 1960, she had a cold that was followed by ankle edema and moderate dyspnea, but no orthopnea. The patient was then admitted to the Hospital of the University of Pennsylvania.

Physical examination revealed an acutely ill girl who lay in bed with minimal discomfort. She was cyanotic and had moderate pretibial and pedal edema. The blood pressure was 125/70, the pulse 90. The cervical veins were distended at 90°, and filled from below. There were fine rales and rhonchi in both lung fields. The heart was enlarged to the left. The rhythm was regular. There was a loud holosystolic, harsh murmur parasternally, which radiated to the apex. A protodiastolic gallop was present. The liver was palpable 5 cm. below the costal margin and was nontender.

The patient was treated with bed rest, diuretic agents, and digitalis. During the first 72 hours she lost 5 Kg. and during her hospitalization 9 Kg. of edema. After the diuresis, the gallop rhythm disappeared though the systolic murmur persisted.

The hemoglobin ranged from 13.5 to 15.9 Gm. per 100 ml. and there was persistent 2+ proteinuria. The total serum protein was 6.4 Gm. albumin 3.0 and globulin 3.4 Gm. per 100 ml. The electrocardiogram showed right and left atrial enlargement and left ventricular hypertrophy. The mean QRS axis was -30° (fig. 3). The chest roentgenogram showed nonspecific cardiac enlargement and a slight decrease in pulmonary vascularity (fig. 9). Cardiac catheterization (table 1) and venous angiocardiography (fig. 10) were performed.

The patient is permitted limited activity and is fairly well compensated except for paroxysmal supraventricular and ventricular tachycardia.

Cardiac Catheterization

These data are summarized in table 1 and figure 11. None of the cases showed clinical evidence of congestive heart failure at the time of their study; cases II-1 and III-1 had been digitalized. The pulmonary arterial pressures were normal. There was significant elevation in right ventricular end-diastolic pressures (except in the first catheterization in case III-1). No pressure gradient was found across the pulmonic or tricuspid valve. Right atrial pressure equaled left atrial or pulmonary capillary venous pressure in individual cases. A step-up in oxygen content at the atrial level was consistent with atrial septal defect in cases II-1 and III-1; the data in case II-4 were incomplete. Higher concentrations of oxygen in the right ventricle or pulmonary artery were compatible with incomplete mixing of the shunted blood when

*Details of this case were furnished through the courtesy of Dr. John H. Helwig, Jr., Associate in Medicine, University of Pennsylvania School of Medicine.

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other data were taken into account. For example, in case II-1, the data also suggested ventricular septal defect. This was excluded by the failure to record the characteristic murmur in the right ventricle at the time of cardiac catheterization. Although the findings in case III-1 were consistent with ventricular septal defect at the time of the first catheterization, the data indicated atrial septal defect at the second catheterization. Nevertheless, it appears from a further step-up in oxygen content that a ventricular septal defect is present as well. Oxygen samples low in the atrium and near the valve were not significantly different from the ventricular samples. Finally, all patients showed arterial oxygen desaturation from right-to-left shunting through the atrial septal defect.

Angiocardiography

These were difficult to interpret in the anteroposterior projection (figs. 8 and 10) and reference should be made to the pathologic specimen (fig. 5) for orientation. The right atrium was dilated and there was early appearance of dye in the left atrium signifying right-to-left shunt. Because of superimposition of the right ventricle on the left atrium, the full extent of the right ventricle was difficult to distinguish with certainty.

Discussion

Anatomy

The defect was characterized by a small right ventricular chamber with normally separated and positioned tricuspid and pulmonic valve leaflets. The tricuspid valvular ring was small or functionally dilated by heart failure. The myocardium was thin or of normal thickness and showed normal histologic features. Its vascular supply was normal. The atria were dilated and hypertrophied because of the
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Figure 6
Schematic view of heart of case II-1. The relative dimensions obtained at necropsy are depicted (for abbreviations see figure 5A and B).

interatrial shunt and the restriction to filling offered by the hypoplastic right ventricle. In the 2-month-old baby in the present paper and the two infants reported by Medd et al., the foramen ovale was patent but competent. In the older patients, the 4-year-old child reported by Gasul et al. and cases II-1 and II-4, 39 and 22 years old, respectively, an atrial septal defect was present. The left ventricle was dilated and hypertrophied presumably because it bore the brunt of the circulation. In case II-1, a mild aortic stenosis and left ventricular fibroelastosis were found but were not considered to play a significant part in the clinical picture.

This disorder must be differentiated from right ventricular hypoplasia associated with other malformations. By far, the most common of these is tricuspid atresia. Pulmonary atresia with closed ventricular septum, overriding of the tricuspid valve as in some cases of complete transposition of the great vessels, and tricuspid stenosis may also be responsible. In Ebstein’s malformation, the right ventricle is of thin or normal thickness but its cavity is always diminished by a downward displacement of the tricuspid valve. Cooley et al. mentioned a case of right ventricular hypoplasia secondary to an absence of the right coronary artery. Finally, Uhl reported a 7-month-old child in whom the right ventricular myocardium was replaced by fibrous tissue (“parchment heart”). In this anomaly the right ventricular endocardium and epicardium were contiguous while the left heart and coronary vessels were normal. In contrast to the diminutive chamber in isolated right ventricular hypoplasia, the right ventricular chamber was dilated and filled with a large laminated thrombus.

Etiology
The etiology of this anomaly is unknown. Medd et al. pointed out that the hypoplasia of the right ventricle may be a primary developmental anomaly or be secondary to a reduction of tricuspid flow during fetal life. It is of interest that five of the eight reported cases are familial. Moreover, it is the only type of congenital heart disease in the affected fami-
ties. This is in agreement with the observations of Wood, who stated that congenital heart disease occurring in more than one member of a family is nearly always of the same type.

**Hemodynamics**

The case of Gasul et al. and the three cases in the present report constitute the basis for this discussion; all had atrial septal defects. No data are available for those cases with patent foramen ovale who died during infancy.

The basic hemodynamic disturbance is the obstruction to inflow of blood into the right ventricle due to the small size of the chamber. Before puberty, the right ventricle end-diastolic pressure may be normal (Gasul et al. and case III-1). There is a left-to-right atrial shunt but, in addition, a right-to-left shunt is present because the diminutive right ventricle cannot accept both the systemic return and the blood shunted from the left atrium. At this stage, the only manifestation of the restriction to inflow is a prominent “a” wave in the atrial pulse and on the right ventricular pressure tracing. With time, the small right ventricle dilates and the end-diastolic pressure rises but this still is inadequate. The mixed atrial shunt persists and the arterial desaturation provokes secondary polycythemia. Thus, the picture in the adult is that of a mixed atrial shunt, an elevated end-diastolic pressure in the right ventricle with a prominent “a” wave, and the absence of pulmonary hypertension, tricuspid stenosis, or pulmonary stenosis.

This is a unique situation previously described only in certain cases of Ebstein’s malformation. The recording of intracardiac pressure and intracardiac electrical potential simultaneously serves to differentiate this anomaly from isolated right ventricular hypoplasia. In Ebstein’s malformation, right ventricular electrical potential is recorded with right atrial pressure pulse. This denotes that the right ventricle forms part of the right atrial cavity.

Though the end-diastolic right ventricular pressure is approximately one third the systolic, a situation akin to chronic constrictive pericarditis, the pressure pulse contour is not the same. The striking feature in constrictive...
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Chest roentgenograms of case III-1. At the age of 10 years, the heart is top normal in size and the pulmonary vascular markings are slightly increased. Three years later on March 7, 1960, the heart has undergone considerable enlargement and the pulmonary vascular markings are less prominent.

Pericarditis is an early diastolic dip followed by a rapid rise of the diastolic pressure to form a plateau. In contrast to the prominent "a" waves in the right atrial and ventricular pulse pressures in isolated right ventricular hypoplasia, giant "a" waves have not been noted. Furthermore, arterial oxygen desaturation has not been observed in cases of atrial septal defect complicated by constrictive pericarditis.

In case II-1, aortic stenosis had reduced the valve area to 1.9 cm². Unfortunately, left heart catheterization was not carried out but on the basis of a theoretical formula relating pressure, flow and valve area, it appears that the lesion was not hemodynamically significant.

Clinical Features

Habitus. If this anomaly is symptomatic at birth or an early age, development may be retarded. On the other hand, if symptoms develop after puberty, as in Family E, the patients are tall and slender, with normal or slightly decreased weight. The physiognomy is not consistent with that of the Marfan syndrome.

Cyanosis. This appeared at birth or in childhood and was a feature common to all the cases. Secondary polycythemia was present in the adults. Clubbing of the fingers was noted in two of the eight cases (Gasul et al. and case II-4).

Dyspnea. In general, this was not a major complaint and was brought out usually with exercise. Nevertheless, the adults with the disorder were able to carry out moderately strenuous activity without discomfort (case II-1 was a coal miner).

Signs of Venous Congestion. All the adults showed elevated cervical venous pressure with prominent "a" waves, hepatomegaly, proteinuria, and peripheral edema at some stage.
of their disease. The elevated cervical venous pressure persisted even after diuresis to a dry weight. However, in Gasul's 4-year-old child, after clearing of the presenting signs of ascites, edema, and increased cervical venous pressure, the right atrial pressure became normal. Finally, the two infants who died during the first week of life, both had pulsating livers.4

Signs of Pulmonary Congestion. Orthopnea was not recorded in any of the cases. Moreover, fine basilar rales were heard infrequently even in patients with marked right-sided failure.

Cardiac Impulse. The impulse was displaced to the left and was forceful though diffuse. In case II-1, an apex cardiogram revealed that the presystolic outward thrust was as prominent as the ventricular systole. This probably was due to the large amount of blood filling the left ventricle from a left atrium distended by the right-to-left shunt.

Cardiac Auscultation. There are no specific murmurs in this disorder though gallop rhythms are common. Thus, a presystolic gallop was heard in case 2 of Medd et al.,4 in Gasul et al.,3 and cases II-1 and II-4; a protodiastolic gallop was heard in III-1. The two infants and the baby in the present report had no murmurs. In cases II-4 and III-1, rather harsh pansystolic murmurs were heard parasternally. Their significance is questionable; perhaps they were due to a functional tricuspid insufficiency, though the contour of the pressure pulses from the right atrium did not lend support to this hypothesis. A regurgitant jet probably would not produce a large "v" wave, for the right atrial dilatation was extreme, and there was free communication with the left atrium. In case II-1 an attempt
Pressure pulse tracings of the right ventricle (RV) and the right atrium (RA). From the top downward are cases II-1, II-4, and III-1. The curves are remarkably similar in contour (the tracings are damped in II-4). The right ventricular systolic pressures are normal or slightly increased; the end-diastolic pressures are increased. The “a” waves are prominent and superimposed on the ventricular pressure tracings.
was made to localize the source of the auscultatory findings by recording the intracardiac sounds. The presystolic gallop, which was transmitted widely over the precordium, originated wholly or in part from the right side of the heart (the left atrium and ventricle were not examined). The soft, blowing, ejection murmur at the aortic region originated in the pulmonary artery and was probably due to increased flow from the left-to-right interatrial shunt. Finally, the systolic click was not heard in the right side of the heart nor ascending aorta, and its site of origin is unknown.

**Electrocardiography**

The electrocardiographic features of isolated right ventricular hypoplasia are quite similar to those found in tricuspid atresia. In six cases of right ventricular hypoplasia (Gasul et al., Medd et al., Family E), all showed signs of left ventricular hypertrophy or absence of right ventricular potential in the precordial leads. There was left axis deviation in five cases, right axis in one case. Combined atrial hypertrophy was found in four cases, right atrial hypertrophy was found in two cases. In tricuspid atresia, a condition also associated with defective right ventricular development, practically all cases show left ventricular hypertrophy in the precordial leads, about 90 per cent have left axis deviation, and approximately 70 per cent show "P" wave abnormalities. Therefore, on electrocardiographic grounds, isolated hypoplasia of the right ventricle and tricuspid atresia cannot be differentiated. In Ebstein's malformation, however, which may present with similar clinical findings, left ventricular hypertrophy is quite rare and approximately 75 per cent show right bundle-branch block.

**Roentgenography**

*Teleroentgenograms.* The cardiac silhouette is normal or shows moderate enlargement with no specific contour. Frequently left ventricular hypertrophy is present. In contrast to tricuspid atresia of which approximately 80 per cent have scanty pulmonary vascular markings, the markings were normal or slightly increased in 80 per cent of the five cases of isolated right ventricular hypoplasia (the present series, Gasul et al. and Medd et al.).

**Angiocardiography.** This shows an enlarged right atrium with early opacification of the left atrium due to the right-to-left shunt. The right ventricle is diminutive and may be difficult to define because of its superimposition on the early opacified left atrium. The left ventricle is hypertrophied and dilated.

**Diagnosis**

The diagnosis of this disorder should be suspected when cyanosis and signs of peripheral venous congestion predominate in a patient with left ventricular hypertrophy of uncertain etiology. Nonspecific systolic murmurs and gallop rhythms are frequent. The electrocardiograms show left axis deviation, left ventricular hypertrophy, and right atrial or combined atrial enlargement. The clinical diagnosis is confirmed by cardiac catheterization and venous angiocardiography. The presence of a mixed shunt at the atrial level in the absence of pulmonary hypertension, tricuspid or pulmonic valvar disease demonstrates the restriction to inflow caused by the hypoplastic right ventricle. This restriction is further suggested by the prominent "a" waves in the right atrial and ventricular pressure pulses.

Pulmonary atresia, Ebstein’s malformation, tricuspid atresia, and Bernheim’s syndrome must be considered in the differential diagnosis because of the findings of cyanosis, right-sided failure, left ventricular enlargement, and absence of distinctive murmurs. Pulmonary atresia can be excluded because a split second sound is present. As contrasted to the left ventricular hypertrophy in isolated right ventricular hypoplasia, the electrocardiogram in Ebstein’s malformation shows right bundle-branch block in 75 per cent of cases. A definite differentiating point can be obtained only by recording intracardiac pressures and electrocardiograms simultaneously. In Ebstein’s malformation right atrial pressure is recorded with right ventricular electrical potential. The clinical electrocardiographic and roentgenographic features of tricuspid atresia.

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are quite similar to isolated right ventricular hypoplasia. Cardiac catheterization readily differentiates the two, since the catheter cannot be passed across the atretic tricuspid valve. In Bernheim’s syndrome signs of isolated right-sided heart failure are produced by stenosis of the cavity of the right ventricle by displacement of the interventricular septum. Cyanosis may be present due to peripheral stasis. A history of preceding left ventricular disease is usually obtained.

Prognosis

It is noteworthy that the reported patients who died in infancy and the 2-month-old baby in this paper had patent but valvular competent foramen ovale. However, the cases of 4 to 39 years of age had atrial septal defect. Whether this is a fortuitous finding or is significant cannot be decided on the limited number of cases. The cause of death is severe intractable heart failure, which may be complicated by serious cardiac arrhythmias.

Treatment

Gasul and associates reported the only case in which surgical palliation of the defect has been attempted. They anastomosed the superior vena cava to the right pulmonary artery in an effort to lower the load to the small right ventricle. Symptomatic improvement was evident, the peripheral arterial oxygen saturation rose from 80.6 to 89.4 per cent, the mean right atrial pressure dropped from 7 to 4 mm Hg, and a left-to-right atrial shunt then predominated. However, no electrocardiographic or roentgenographic changes resulted, and the patient showed signs of congestive heart failure 9 months postoperatively. The authors stated that they are considering closure of the atrial septal defect.

Summary

With the rare exception of connective-tissue replacement of the right ventricular myocardium (“parchment heart”), underdevelopment of the right ventricle, as a clinical entity, has been related to tricuspid atresia or stenosis, pulmonary atresia and certain cases of transposition of the great vessels. This report is concerned with four cases, aged 2 months to 39 years, in which a diminutive right ventricle was not dependent on these factors. Three cases occurred in the same family and, in association with atrial septal defect, produced distinctive clinical and hemodynamic features. Right-sided heart failure predominated, dyspnea was mild to moderate, and cyanosis was present in association with clinical, electrocardiographic, and roentgenographic evidence of left ventricular enlargement. Cardiac catheterization and venous angiography showed a bidirectional interatrial shunt, normal pulmonary arterial pressure, no pressure gradient across the pulmonic and tricuspid valves, and a diminutive right ventricle. Anastomosis of the superior vena cava to the right pulmonary artery and closure of the atrial septal defect may ameliorate the hemodynamic disturbance.

References


Religio Medici

Think not thy time short in this World since the World it self is not long. The created World is but a small Parenthesis in Eternity and a short interposition for a time between such a state of duration, as was before it and may be after it.—Sir Thomas Browne. Religio Medici, 1642. Edited by W. A. Greenhill, M.D., Oxon., London, MacMillan and Co., Limited, 1950, p. 230.
Isolated Right Ventricular Hypoplasia with Atrial Septal Defect or Patent Foramen Ovale

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