Ebstein’s Anomaly of the Tricuspid Valve
Report of Three Cases and Analysis of Clinical Syndrome

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In Ebstein’s anomaly the tricuspid valve is displaced downward so that the upper portion of the right ventricle is incorporated in the right auricle. This impairs the efficiency of the right side of the heart and produces a distinctive syndrome, which is described here for the first time. Diagnosis is important because this malformation, which is not amenable to surgery, may be confused with the tetralogy of Fallot.

Ebstein in 1866 first described a congenital malformation of the tricuspid valve in which the leaflets are fused into a membranous structure, which extends like a basket down into the cavity of the right ventricle and separates the ventricle into a proximal and a distal chamber. The proximal portion is continuous with the right auricle, and the distal portion, which functions as the right ventricle, includes the outflow tract of the right ventricle.

Often the valve leaflets are completely fused with the ventricular endocardium over large areas so that it is difficult to identify the individual cusps with certainty. The anterior leaf is usually the largest, the posterior leaf usually the most malformed. Sometimes the origin of the valve leaflets posteriorly appears displaced downward from the normal site at the annulus fibrosus toward the apex of the right ventricle. Medial to the anterior leaflet there is an opening into the functional portion of the right ventricle. The right ventricle above the valve is exceedingly thin-walled. The foramen ovale in most of the cases is anatomically and functionally patent.

Since Ebstein’s original case there have been 22 such malformations reported. Including the 3 patients herein described, the total number of recorded instances of this malformation is 26.

In all of the reports in the literature the diagnosis of Ebstein’s anomaly has been made only at postmortem examination. Yater and Shapiro in their review in 1937 stated that “it would appear impossible to make the diagnosis during life.” We have recently studied three patients with Ebstein’s malformation of the heart and have re-examined the case previously reported by Taussig. Although the correct diagnosis was not made during life, an analysis of these cases has revealed several features of this malformation which are sufficiently characteristic to permit clinical diagnosis.

The diagnosis is of more than academic interest, because when there is cyanosis and a diminished pulmonary blood flow, this malformation may resemble the tetralogy of Fallot. Indeed, in two of our cases (Cases 2 and 3) operation to increase the blood supply to the lungs was undertaken. Both patients died following the operative procedure.

Case Reports

Case 1.—G. C. (HLII No. A 61737), a 10 year old white girl, was examined in the Cardiac Clinic in March, 1948.

Past History and Present Illness: A heart murmur was noted at birth, and the family was informed that she had congenital heart disease. Nothing is known concerning her color at birth, but she was kept in oxygen for the first three days. Throughout early childhood she had infrequent attacks of “violent pains” in her abdomen and seemed tired for the rest of the day. During the attacks her color was “peculiar.” In retrospect the mother thought that the patient had been cyanotic during these episodes. The child was never robust, but she played with the other children and started school at the normal time.
When she was 7 years old, she was taken to the family physician for a routine examination. He found that her heart was enlarged (cardiothoracic ratio of 68 per cent) and, although she was asymptomatic, thought that she had acute rheumatic fever. She was put to bed and given salicylates for three weeks; after three months she was allowed to get up.

At the age of 8 years cyanosis occurred after exertion, and she felt tired and weak. She was examined by one of us (H. B. T.), who found a greatly enlarged heart with a rough systolic and diastolic murmur which simulated a pericardial friction rub. Teleorontgenogram of the chest showed an increase in the cardiothoracic ratio to 75 per cent. The diagnosis was made of pericarditis superimposed on a congenital malformation of the heart. A period of bed rest with salicylates and sodium bicarbonate medication was advised.

Thereafter the patient's cyanosis increased progressively, and by the age of 9 years cyanosis was constantly present. Dyspnea was never a striking feature. She could walk four blocks very slowly before becoming tired, but she did not squat to rest. She seemed exhausted much of the time. She was given digitalis for a month without improvement; therefore, digitalis was discontinued. Four weeks before her visit to the clinic, she had a brief "fainting spell" during which she became unconscious, intensely cyanotic, and had an involuntary bowel movement. She experienced a similar episode on admission to the Harriet Lane Home.

Physical Examination. Temperature 37 C.; pulse 104 per minute; respirations 26 per minute; blood pressure 110/86 mm. of Hg. She was a tall, thin, 10 year old girl who had slight cyanosis of the mucous membranes and nailbeds at rest. The cyanosis was of equal distribution over the body. There was no clubbing of the fingers or toes. The left chest anteriorly was more prominent than the right. There was no Broadbent's sign. Her heart was greatly enlarged to the right and to the left. The point of maximal impulse was in the anterior axillary line in the fifth left interspace. No thrill was palpable. The heart sounds were almost inaudible; there was a gallop rhythm. There was a rough, soft, to and fro systolic and diastolic murmur heard all over the precordium and over the lung fields anteriorly. The murmur sounded scratchy in the fourth interspace over the sternum. Over the left chest posteriorly and maximal at the left base, there was a very loud, rasping, systolic murmur; this murmur was less well heard over the right chest posteriorly. The liver was palpable 1 cm. below the right costal margin. It was firm but was neither tender nor pulsatile. The pulses in the extremities were equal and of small volume. There was no venous distention nor peripheral edema.

Laboratory Findings: Fluoroscopy revealed a tremendously enlarged heart. It almost filled the left chest and extended far into the right chest. The base of the cardiac shadow appeared narrow in comparison with the bulk of the body of the heart. The cardiac pulsations were of exceedingly small amplitude. Because of these findings the possibility of a pericarditis and a pericardial effusion were considered, but nothing was found to substantiate the diagnosis. In the right and left anterior-oblique positions, the right ventricle extended to the left anterior chest wall and was flattened against it for a distance of several centimeters. This was interpreted as indicating a huge right ventricle. In the left anterior-oblique position, the heart posteriorly completely overlapped the spine even at 80 degrees of rotation. For this reason the left ventricle was considered enlarged. There was no prominence of the pulmonic conus region. The pulmonary arteries were obscured by the heart anteriorly but with rotation were seen to be of normal size without pulsations. The lung fields were unusually clear. Barium swallow showed evidence of a left aortic arch but no left auricular enlargement. The telesoentgenogram (figs. 1 and 2) showed the cardiothoracic ratio to be 77 per cent.

The standard limb leads and the unipolar precordial leads of the electrocardiogram showed prolonged A-V conduction time and right bundle branch block. The P waves were high and peaked. The hemoglobin level was 19.8 Gm. (137 per cent). The red blood cell count was 7.2 million per cu. mm.; the hematocrit reading was 61. The white blood cell count and the differential count were normal. The sedimentation rate was 2 mm. in one hour uncorrected. Studies of the arterial blood revealed an oxygen content of 19.7 volumes per cent with a
capacity of 26.4 volumes per cent, giving an arterial oxygen saturation of 74 per cent. The carbon dioxide content was 39.4 volumes per cent. The arm-to-tongue circulation time determined with Deeholin was prolonged to 20 seconds. (In our experience the upper limits of normal for a circulation time so determined is 12 seconds for a patient of this size.) Tuberculin tests were negative up to 1.0 mg. of old tuberculin.

Clinical Impression: This patient presented a diagnostic and therapeutic problem. Because of the large heart, the poor heart sounds, the scratchy to and fro murmur which simulated a friction rub, the gallop rhythm, and the decreased amplitude of pulsations under the fluoroscope, it was postulated that she had chronic pericarditis, most likely rheumatic in origin, superimposed on a congenital heart defect. The nature of the congenital malformation was puzzling.

Course: After a ten day stay in the hospital, she was transferred to a convalescent home for a period of bed rest. There her heart continued to enlarge. On the morning of her twentieth day in the convalescent home, while lying in bed, she suddenly died.

Postmortem Examination: Autopsy No. 21188, performed by Dr. Payne. On opening the thoracic cavity, the heart was seen to occupy over three-fourths of the transverse diameter of the chest. Its great size was due entirely to the tremendous dilatation of the right auricle and the right ventricle. The left ventricle, which was essentially normal in size, was completely concealed behind the huge right ventricle.

The heart weighed 260 grams (normal approximately 150 grams). The pericardium was normal, and there was no fluid in the pericardial cavity. The superior and inferior vena cavae entered the right auricle, and the pulmonary veins entered the left auricle in the normal fashion. The orifice of the coronary sinus was wide. The right auricle was hypertrophied as well as dilated. The foramen ovale was patent, measuring 1.5 cm. in greatest diameter. Although it contained a valve, in the dilated state of the right auricle this valve was incompetent.

The tricuspid valve was grossly malformed (fig. 3). It was ballooned downward into the right ventricle and in places fused with the ventricular endocardium and in other places closely bound to it by short chordae tendineae and small papillary muscles. In the region of the infundibulum of the right ventricle, it formed a redundant membrane which divided the right ventricle into two parts: a small, distal outflow chamber which emptied into the pulmonary artery, and a much larger proximal chamber continuous with the right auricle through the unprotected tricuspid valve ring. This ring measured 12 cm. in circumference.

The leaflets of the tricuspid valve were so malformed that they could not be individually identified. On the septal portion of the auriculoventricular ring, there was no free valve leaflet distinguishable from the smooth endocardial surface. About 1 cm. distal to the ring, however, there was on the septal endocardium a small, irregular, raised area which measured 1 cm. in diameter and probably represented a malformed portion of the tricuspid valve. From the remainder of the auriculoventricular ring, there arose a large leaflet which extended into the ventricular cavity closely bound to its walls. This leaflet blended distally with the septal endocardium except in the region of the infundibulum of the right ventricle, where it presented a free margin for a distance of 5 cm. There were two fenestrations in this leaflet; one measured 1 cm. and the other 1.5 cm. in diameter. Both of these openings were held close to the ventricular wall by short chordae tendineae and papillary muscles so that they were functionless. The main communication between the two chambers of the right ventricle was through an opening formed by the redundant free margin of the valve and the prominent moderator band. This opening would have closed during ventricular systole; therefore, the valve was competent. The moderator band was enlarged; it measured 3 cm. in length and 1 cm. in diameter. It stretched across the ventricular cavity from the right ventricular wall to the septum.

The wall of the right ventricle proximal to the tricuspid valve was very thin (fig. 4); in some places it measured not more than 2 mm. in thickness. Microscopically the muscle fibers were normal in appearance but were reduced in number. Irregular
Fig. 3.—Ebstein's anomaly of the heart. Case 1. Upper drawings show the great enlargement of the right auricle and the right ventricle, the patent foramen ovale, and the anomalous position of the tricuspid valve. Lower drawing shows the interior of the right auricle and the manner in which the tricuspid valve is fused with the endocardium of the thin-walled right ventricle. The opening from the large proximal chamber into the functional portion of the right ventricle is illustrated.

Fig. 4.—Section through tricuspid valve ring. Case 1. A indicates right auricular wall, V the right ventricular wall, and TV the tricuspid valve leaflet. The thinness of the right ventricular wall as compared with the right auricular wall is apparent. The portion of the valve leaflet shown in this section is not fused with the ventricular endocardium.
muscle trabeculae arose from the right ventricle in the region where the valve leaflet blended with the septal endocardium. Below the displaced valve the myocardium of the right ventricle was 5 mm. in thickness and microscopically was normal.

The pulmonary valve was normal; the valve ring measured 4.5 cm. in circumference. The pulmonary artery was normal in size. The ductus arteriosus was closed. The ventricular septum was intact. The left auricle, the mitral and aortic valves, and the aorta were normal. The left ventricle was slightly hypertrophied; its wall measured 9 mm. in thickness. The coronary arteries were grossly normal, but microscopically moderate sclerosis was seen. There was moderate chronic passive congestion of the liver and pancreas.


Case 2.—J. T. H. (H.L.H No. A 44703), a 5 year old white boy, was first examined in the Cardiac Clinic at the age of 3 years in December, 1945.

Past History and Present Illness: At the age of 2 days a heart murmur was heard, and he was diagnosed as a "blue baby." Dyspnea was never as apparent as the cyanosis. He tired easily and rested often, but he never squatted to rest. At the time of his first visit, he could walk only one hundred feet before becoming tired.

Physical Examination: Temperature 37 C.; pulse 112 per minute; respirations 30 per minute. The blood pressure could not be obtained by auscultation, but by palpation the systolic pressure was 90 mm. of Hg. He was a moderately well developed and well nourished boy who was quite cyanotic when crying. His fingers and toes were clubbed. There was a precordial bulge. The heart was enlarged to percussion. The heart sounds were so distant that they were heard only with difficulty. There was a gallop rhythm. There was a soft, blowing, systolic murmur over the entire precordium. The liver was palpable 2 1/2 fingers breadth below the right costal margin; it was not pulsating. There was no peripheral edema.

Laboratory Findings: Fluoroscopy showed the heart to be enlarged to the right and to the left. There was enlargement of the right auricle and the right ventricle. In the left anterior-oblique position the left ventricle overlapped the spine at 60 degrees of rotation. There was no fullness in the region of the pulmonary conus. The lung fields were remarkably clear. A barium swallow demonstrated a left aortic arch and no evidence of left auricular enlargement.

The teleorontgenogram showed the cardiothoracic ratio to be 63 per cent (fig. 5). The electrocardiogram showed first degree heart block and right bundle branch block. The P waves were high and peaked. The hemoglobin level was 27.5 Gm., and the red blood cell count was 8.55 million per cu. mm. An arterial blood sample obtained when the patient was crying revealed an oxygen content of 8.75 volumes per cent, an oxygen capacity of 28.2 volumes per cent, and an arterial oxygen saturation of 31 per cent. The carbon dioxide content was 34.8 volumes per cent.

Clinical Impression: The patient had a malformation which caused inadequate pulmonary blood flow, but the nature of this malformation was not clear.

![Fig. 5.—Anterior-posterior view of the chest. Case 2.](http://circ.ahajournals.org/)

Course: Because of the enlarged heart and liver, the patient was given digitalis. While on this medication, his liver decreased slightly in size, his cyanosis lessened, and his exercise tolerance increased so that by the age of 5 years he could walk four blocks slowly and climb one flight of stairs. His heart, however, continued to enlarge. The cardiothoracic ratio in January, 1947, measured 68 per cent. The heart sounds remained faint; the gallop rhythm and systolic murmur persisted. His blood pressure was 98/84 mm. of Hg. The arm-to-lips circulation time determined with fluorescein was 12 seconds. (The upper limits of normal for his size is 9 seconds.) In January, 1948, at the age of 5 years, he returned for additional diagnostic studies.

Angiocardiogram in the anteroposterior view showed that the contrast medium entered through the superior vena cava into the large right auricle and then dispersed throughout the entire heart. The contrast medium extended almost to the margins of the cardiac silhouette, indicating that the
walls were thin. The Diodrast lingered for an abnormally long time in the right side of the heart. The pulmonary vascular bed opacified slowly and in
drast passed from the large right auricle directly into the left auricle, indicating a defect in the auricular septum. The left auricle emptied the dye

![Angiocardiogram, lateral position. Case 2.](image)

**Fig. 6.**—Angiocardiogram, lateral position. Case 2. *a.* Film taken 1 sec. after injection shows Diodrast entering right auricle through superior vena cava. *b.* Two seconds. Dye has filled the enlarged right auricle and is visible in the left auricle. *c.* Four seconds. The contrast medium is dispersed throughout the right auricle and right ventricle and is visible in the pulmonary arteries. The aorta is opacified. *d.* Seven seconds. Dye has disappeared from the left side of the heart and the aorta but still lingers in the right side of the heart.

a subnormal amount. The aorta was not visualized in the anteroposterior view. In the films taken with the patient lying on his left side, some of the Diodrast promptly into the left ventricle; then the aorta filled. (See fig. 6.)

*Cardiac Catheterization:* The oxygen content of
EBSTEIN'S ANOMALY OF TRICUSPID VALVE

the superior vena cava was 16.1 volumes per cent; of the right auricle 17.5 volumes per cent; and of what was thought to be the right ventricle, 17.8 volumes per cent. The pressure in the right auricle was 8/1 mm. of Hg. and in the latter chamber, 21/7 mm. of Hg. The pulmonary artery could not be entered. The catheter tip passed into two chambers, the oxygen contents of which were higher than those of the right side of the heart. These chambers were interpreted as the left auricle (oxygen content 21.8 volumes per cent; pressure 8/2 mm. of Hg.) and the left ventricle (oxygen content 21.5 volumes per cent; pressure 48/12 mm. of Hg.). The oxygen content of the femoral artery was 20.2 volumes per cent, and the oxygen capacity 30.6 volumes per cent, giving an arterial oxygen saturation of 66 per cent. The pulmonary blood flow was markedly reduced; it was only 1790 cc. per square meter of body surface per minute as compared with a systemic blood flow of 4350 cc./sq. M./min. The effective pulmonary flow was calculated to be 1320 cc./sq. M./min. Although there was some shunting of oxygenated blood into the right auricle, the overall intracardiac shunt was from right to left and was of the magnitude of 2560 cc./sq. M./min.

On the basis of these tests it was believed that the patient had a tetralogy of Fallot and an auricular septal defect, and probably could be benefited by a Blalock-Taussig operation. On January 12, 1948 an anastomosis was performed between the end of the right subclavian artery and the side of the right pulmonary artery. Three times during the operation the heart stopped beating; shortly after the chest was closed, the heart again started and could not be revived.

Postmortem Examination: Autopsy No. 21012, performed by Dr. Payne. On opening the thoracic cavity, the right auricle and right ventricle were seen to be so dilated that the heart filled most of the left chest and extended far into the right chest. As in the first case the left ventricle was normal in size and was entirely concealed behind the huge right ventricle.

The heart weighed 100 grams (normal approximately 95 grams); there was little or no hypertrophy. The pericardial sac was normal. The venous return was normal. There was a poorly developed eustachian valve, and the orifice of the coronary sinus was atretic. The right auricle was hypertrophied as well as dilated. The foramen ovale was patent, measuring 1 cm. in diameter. Although the foramen ovale was guarded by a valve, the tremendous dilatation of the right auricle caused the valve to be incompetent.

The tricuspid valve was greatly malformed (fig. 7). It extended down into the right ventricle and was fused in places with the endocardium and in other places closely bound to it by short chordae tendineae and small papillary muscles. In the region of the infundibulum of the right ventricle, it formed a redundant membrane which divided the right ventricle into two parts: a small outflow chamber which emptied into the pulmonary artery, and a much larger chamber continuous with the right auricle through the unprotected tricuspid valve ring. The latter measured 9 cm. in circumference.

The individual leaflets of the tricuspid valve were difficult to identify. On the septal portion of the auriculoventricular ring, there was no valve leaflet distinguishable from the smooth endocardial surface. From the remainder of the auriculoventricular ring, there arose a large leaflet which extended into the ventricular cavity and was closely attached to its wall. The leaflet blended distally with the septal endocardium except in the region of the infundibulum of the right ventricle, where it presented a partially free margin for a distance of 6 cm. In this portion of the valve there was an opening which measured 1.5 cm. in diameter and was situated 1 cm. from the free margin. The edges of this opening were attached to the ventricle wall by short chordae tendineae and tended to overlap. Communication between the two chambers of the right ventricle was through the opening in the valve leaflet itself or through the orifice formed by the free margin of the valve and the ventricular septum. Both of these openings would have closed during ventricular systole. The wall of the right ventricle above the abnormal valve was exceedingly thin; in some places it measured not more than 1 mm. in thickness (fig. 8). Microscopically the muscle fibers were normal in appearance but reduced in number. In the region of the infundibulum of the right ventricle, however, the myocardium was 3 mm. in thickness and microscopically was normal.

The pulmonary valve was entirely normal; the valve ring measured 2.5 cm. in circumference. The pulmonary trunk was normal. The right subclavian artery was anastomosed surgically to the right pulmonary artery. The ductus arteriosus was obliterated. The ventricular septum, the left auricle and left ventricle, the mitral and aortic valves, and the aorta were normal. The coronary arteries were normal. The thebesian veins were prominent. There was no passive congestion of the viscerum.


Case 3.—C. T. (H.1.11 No. A 46196), a 16 year old white girl, was seen in the Cardiac Clinic in February, 1946, at the age of 14 years.
**Past History and Present Illness:** She was born with scoliosis and was cyanotic at birth. After the first few days the cyanosis disappeared. She was limited. Until the age of 11 dyspnea was very slight; thereafter, it steadily increased although it never became as marked as the cyanosis. As her dyspnea increased, she developed the habit of occasionally squatting to rest.

**Physical Examination:** Temperature 37 C.; pulse 100 per minute; respirations 24 per minute. She was a moderately well developed and well nourished

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**Fig. 7.** Ebstein's anomaly of the heart. Case 2. Note similarity to figure 3.

**Fig. 8.**—Section through tricuspid valve ring. Case 2. Note extreme thinness of right ventricular wall, V, as compared with right auricular wall, A. The valve leaflet is not present in this section.
EBSTEIN'S ANOMALY OF TRICUSPID VALVE

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A girl with a severe S-shaped dorso-lumbar scoliosis and marked chest deformity. Cyanosis was moderately intense. There was slight clubbing of the fingers and toes. The heart was enlarged. The point of maximal impulse was beyond the posterior axillary line. The first heart sound was obliterated by a loud, rasping, systolic murmur which was audible all over the precordium. It was especially well heard in the second intercostal space at the left sternal border, in the fourth and fifth intercostal spaces, and in the posterior axillary line. The liver was palpable at the costal margin but was not pulsating. There was no edema. The remainder of the examination was negative.

Fig. 9.—Anterior-posterior view of chest. Case 3.

Laboratory Findings: Both teleoroentgenogram and fluoroscopy revealed a tremendously enlarged heart which filled the left chest (fig. 9). There was an abrupt angulation at the base and an absence of fullness in the pulmonary conus region. The pulmonary arteries were not enlarged, and there were no expansile pulsations visible in them. The lung fields were exceptionally clear. With barium swallow there was seen a left aortic arch and no evidence of left auricular enlargement. The cardiothoracic ratio was 66 per cent.

The electrocardiogram showed right axis deviation, first degree heart block, and prolongation of intraventricular conduction time suggesting right bundle branch block. The P waves were high and peaked. T2 and T3 were inverted. Unipolar precordial leads were not obtained. The red blood cell count was 7.31 million per cu. mm.; the hemoglobin level was 20 Gm.; the hematocrit reading was 75. Analysis of the arterial blood sample revealed an oxygen content of 21.7 volumes per cent, an oxygen capacity of 34.1 volumes per cent, and an arterial oxygen saturation of 64 per cent. The carbon dioxide content of the arterial blood was 31.4 volumes per cent.

Clinical Impression: It was believed that the primary difficulty was an inadequate pulmonary blood flow, but the great cardiac enlargement and the scoliosis were considered contraindications to a Blalock-Taussig operation.

Course: In 1948, when seen elsewhere, she was thought to have a tetralogy of Fallot. At that time a gallop rhythm was heard, and her blood pressure was found to be 130/110 mm. of Hg. Because she was extremely anxious for operation, it was attempted. Due to technical difficulties it was impossible to complete the anastomosis. Two weeks later, following a stormy postoperative course complicated by thrombophlebitis and cardiac failure, she died.

Postmortem Examination*: Autopsy showed that the right auricle and the right ventricle were extremely dilated. The anomalous tricuspid valve formed an apron-like structure, which extended so far down into the cavity of the right ventricle that the functional portion of the right ventricle was very small, and only the outflow tract of the ventricle was relatively normal in size. The posterior leaflet of the tricuspid valve was fused to a considerable extent with the endocardium. The anterior leaflet in its inferior margin was fixed to the endocardium. The medial leaflet was small and could not be separated from the posterior leaflet. The actual ostium of the distorted tricuspid valve measured 8 cm. in circumference. The myocardium of the right ventricle proximal to the valve was exceedingly thin; it measured between 1 mm. and 2 mm. in thickness. The myocardium distal to the valve was of normal thickness. The foramen ovale was widely patent. The ventricular septum was intact. The pulmonary valve and pulmonary artery, the left auricle and left ventricle, the mitral and aortic valves, and the aorta were normal. There was passive hyperemia of the abdominal viscera. An adherent thrombus filled the main iliac veins and extended into the inferior vena cava. There was marked scoliosis.


Analysis of the Pathologic, Clinical, and Laboratory Findings

The anomaly of the tricuspid valve described in these 3 cases is similar to that recorded by Ebstein. In each instance the foramen ovale was patent, as it was in Ebstein's original case and in two-thirds of those subsequently reported. In association with the

* We are indebted to Dr. K. Terplan for this report.
malformation of the tricuspid valve, in all 3 hearts there was a congenital hypoplasia of the right ventricular myocardium above the malformed valve. The myocardium of the chamber of the right ventricle against which the tricuspid valve was plastered was extremely thin, whereas the ventricular wall of the outflow chamber was of normal thickness. It seems quite unlikely that dilatation alone could have caused this unusual thinness of the myocardium. Thinness of the right ventricular wall proximal to the valve has been noted in nearly all the recorded cases.1–2, 7–10, 18 The case previously reported by Taussig19 was re-examined and in this instance, too, the myocardium in the upper portion of the right ventricle was extremely thin. In 2 cases8, 15 there was an area of localized out-pouching where the wall was exceedingly thin. These areas were considered to be developmental defects. It seems probable, therefore, that hypoplasia of the myocardium of the right ventricle proximal to the malformed tricuspid valve is an integral part of the malformation.

This malformation of both the tricuspid valve and the wall of the right ventricle appears to be due to a developmental defect in the formation of the tricuspid valve and of the myocardium of the right ventricle. Streeper20 has shown that embryologically the myocardium arises from a specialized part of the visceral coelomic wall and is separated from the partially distended endocardium of the primitive heart by the myoendocardial space, which extends throughout the length of the heart tube. This space is filled with a homogeneous transparent jelly, the cardiac jelly. As the myocardium develops, the myoendocardial space gradually disappears except at strategic sites, where it persists as the so-called endocardial cushions from which the tricuspid and other valves are formed. It is thus possible that any defect in the visceral coelomic wall in the region where the right ventricle is to develop could not only cause defective development of the right ventricular myocardium but, by distortion of the position of the persisting primitive myoendocardial space, could cause a malformation of the tricuspid valve.

In these hearts the tricuspid ring is unprotected by a valve, and at first sight it appears that there must have been extreme tricuspid insufficiency. It has frequently been stated in the literature that the valve is insufficient; indeed, “congenital tricuspid insufficiency” has been used as a synonym for Ebstein’s anomaly. Nevertheless, it is a striking fact that in our Case 2 and also in the case reported by Taussig19 there was no chronic passive congestion of the viscera at autopsy. Moreover, in the first case there was only moderate chronic passive congestion of the liver and pancreas; there was not the intense damage that would have been expected had a tricuspid insufficiency existed over a period of years. Only in Case 3 was there passive hyperemia of the abdominal viscera; this was in all probability a terminal event.

The lack of chronic passive congestion and the absence of clinical evidence of tricuspid insufficiency in the 3 other cases were due primarily to the fact that, although the tricuspid ring was unprotected by a valve, the anomalous valve was so arranged that it was competent. The opening between the two chambers at the margin of the redundant valve was closed during ventricular systole. Although the upper chamber may have been unable to empty itself completely with each contraction, the thinness of the wall of the ventricular portion permitted it to serve as a distensible reservoir and lessened the manifestations of right heart failure. Furthermore, the patency of the foramen ovale acted as an “escape valve” and enabled blood to be shunted into the left auricle.

It seems probable that there were two factors involved in the dilatation and hypertrophy of the right auricle proper: first, the opening from the proximal chamber into the outflow chamber was considerably smaller than that of a normal tricuspid valve and thus constituted a functional tricuspid stenosis; second, the outflow chamber, which was smaller than a normal ventricle, was too small to receive all the blood contained in the upper chamber, so that the right auricle was unable to empty itself completely.

Because of the anomalous position of the tricuspid valve, this malformation primarily alters the efficiency of the right heart. The
inefficiency is more readily apparent when one considers how the heart functions when part of the right ventricle is included in the right auricle.

Venous blood is returned in the normal fashion to the right auricle. Auricular systole directs the blood through the proximal portion of the right ventricle, which is in free communication with the right auricle. The direction of the blood through the malformed tricuspid valve into the distal portion of the right ventricle is, however, difficult. The effectiveness of the auricular contraction is lessened by the dilated upper part of the right ventricle, which during auricular systole is in diastole. The tricuspid orifice is relatively small and, furthermore, the distal chamber is too small to receive all the blood from the large proximal chamber. Consequently, the upper chamber is unable to empty itself completely. Although the expulsion of blood may at first be relatively adequate, gradually the volume of blood remaining in this chamber increases. It follows that the ability of the chamber to empty itself progressively lessens. This chamber gradually enlarges, and the pressure increases. The greater the proportion of the right ventricle above the tricuspid valve, the smaller is the distal chamber; as a consequence, the greater is the difficulty of the proximal chamber in propelling blood forward, and the greater is its enlargement.

If the foramen ovale is not completely sealed, as the pressure within the right auricle increases, the valve is forced open, and venous blood is shunted into the left auricle. As the pressure continues to rise, the foramen ovale is constantly held open, and the right-to-left shunt becomes persistent. If the right auricle becomes so distended that the foramen ovale is stretched wide open, there is in effect a gross defect in the auricular septum.

During ventricular systole the misplaced tricuspid leaflets close the opening between the lower and upper chambers, and the distal chamber sends the blood to the lungs. Inasmuch as the volume of blood contained in the lower chamber is less than normal, the lungs receive an inadequate supply of blood for oxygenation. The pulmonary circulation is further diminished by the shunt through the foramen ovale. Although the musculature of the "auricularized" right ventricle is thin and cannot exert much force, it seems probable that it too contracts during ventricular systole and sends the blood against the closed tricuspid valve, against the walls of the auricle, and possibly through the foramen ovale to the left auricle.

The venous blood shunted from the right auricle to the left auricle is mixed with the fully oxygenated blood which is returned from the lungs to the left auricle. This admixture of venous and arterial blood reaches the systemic circulation via the left ventricle and the aorta, and when the venous-arterial shunt is of sufficiently large volume, cyanosis results. If the foramen ovale is closed and there is no defect of the auricular septum, there is no right-to-left shunt; consequently, there is no cyanosis. Under such circumstances the course of the circulation, except for the delay in expulsion of blood from the proximal chamber, is normal.

An analysis of these cases and of those in the literature bears out the theory that the presence or absence of cyanosis is related to the structure of the auricular septum. The foramen ovale was patent in fifteen1, 3–5, 7, 11–15, 18 of the 22 cases reported. Cyanosis was present in all 11 of these 15 cases in which clinical information was given. Another patient who was cyanotic had a gross defect in the auricular septum; this would similarly permit a right-to-left shunt.19 One patient had only probe patency of the foramen ovale and was not cyanotic.16 In 3 patients the foramen ovale was closed. In one of these9 there was no cyanosis. In the second9 cyanosis was noted only "at times" on the third day before death and was associated with terminal heart failure. In the third7 no clinical history was given. In the remaining 2 cases6, 17 there was no information given concerning cyanosis or the structure of the foramen ovale. In each of our cases the foramen ovale was patent and all 3 patients showed persistent cyanosis. In the case reported by Tausig,19 although the foramen ovale was patent, the patient became cyanotic only during the periods of paroxysmal tachycardia and terminally when in heart failure. In this case it is noteworthy that there was less disproportion
between the sizes of the chambers proximal and distal to the malformed tricuspid valve than in the preceding three cases, and consequently the pressure in the right auricle was in all probability but slightly increased.

The malformation may be compatible with life for varying lengths of time. Marxsen's patient lived to be 61 years old, and Malan's lived to the age of 60 years. On the other hand, one child lived for only eight months, and several others died in early childhood. The average age at death was 24 years. The variation in longevity and also in symptomatology is in all probability due to the relative proportions of the right ventricle above and below the anomalous tricuspid valve. If the distal chamber is approximately of normal size, the alteration of the course of circulation is slight, and the symptoms are correspondingly few. On the contrary, when the tricuspid valve is displaced so far downward into the cavity of the right ventricle that the distal chamber is much reduced in size and the greater portion of the right ventricle is proximal to the valve, then the right heart becomes extremely inefficient. Under such conditions the cardiac enlargement is great and progressive, symptoms appear at an early age, and the duration of life is relatively short.

The clinical and laboratory findings in patients with Ebstein's anomaly of the tricuspid valve are explicable on the basis of the altered function of the right side of the heart. The delay in the onset of the cyanosis is dependent on the physiologic closure of the foramen ovale shortly after birth. A right-to-left shunt is thus prevented until the pressure in the right auricle has increased to the level where the foramen ovale is forced open. Thereafter, when a sufficient volume of unoxgenated blood is shunted into the systemic circulation, cyanosis becomes apparent. The tremendous cardiac enlargement is the result of the difficulty in the expulsion of blood from the right auricle. The muffled quality of the heart sounds and the gallop rhythm doubtless reflect the poor functioning of the dilated right side of the heart. The origin of the murmurs, however, is not clear. There are a number of possible factors. The systolic murmur may have been caused by the passage of blood from the right to the left auricle and perhaps also by the regurgitation of a small amount of blood from the lower to the upper chamber through the fenestrations in the malformed valve. The loud systolic murmur heard posteriorly in Case 1 was possibly caused by blood coursing over the enlarged moderator band. The diastolic murmur noted in addition to the systolic murmur in Case 1 and in some of the previously reported cases may have been associated with the abnormal currents of blood within the chamber proximal to the malformed valve as with each cardiac cycle the auricular and the ventricular portions contracted independently.

There was electrocardiographic evidence of prolonged auriculoventricular conduction time in all 3 patients, and in 2 there was a right bundle branch block. In Case 3 there was right axis deviation and evidence of delayed intraventricular conduction suggesting a right bundle branch block. Unipolar precordial leads were not obtained on this patient; hence, no definite statement can be made as to the presence or absence of a bundle branch block. In each of the 3 cases recorded in the literature in which electrocardiograms were illustrated, there appeared to be prolongation of auriculoventricular and intraventricular conduction time. Bauer's patient had a right bundle branch block. Conduction defects are not surprising in view of the tremendous dilatation and thinning of the right auricle and proximal portion of the right ventricle.

The abnormally long circulation time is due to the delay in the expulsion of blood from the large upper chamber. This causes the test solution to linger there before it circulates through the lungs and then reaches the systemic circulation. Although the foramen oval be patent, it has been our clinical experience that rarely is sufficient test material shunted from right to left to give a short circulation time.

The fluoroscopic findings of abnormally clear lung fields and absence of pulsations of the pulmonary arteries are caused by the reduced pulmonary blood flow. It seems reasonable to believe that weak pulsations of the right heart are characteristic of this malformation and will be found, if carefully searched for, in all such
patients. In Case 1 of this report and in Bauer's patient, a decreased amplitude of cardiac pulsations was observed.

The condition leads to progressive cardiac enlargement. In our cases prior to death the cardiothoracic ratio ranged from 66 per cent to 77 per cent, and in Bauer's patient it eventually reached 84 per cent. It is worthy of note that in both our first two patients the left ventricle as well as right ventricle was thought to be enlarged because in the left anterior-oblique position the left ventricle overlapped the spine, even upon extreme rotation. Autopsy, however, showed that all the enlargement was right auricle and right ventricle. Thus it is evident that the right side of the heart can enlarge so greatly that it displaces the left ventricle far posteriorly and causes it to overlap the spine even when the patient is rotated almost into the lateral position.

The findings on angiocardiography reflect the inefficient action of the right heart and the patency of the foramen ovale. The Diodrast was pooled for an abnormally long time in the large proximal chamber. Although the dye which reached the functioning portion of the right ventricle was promptly expelled into the lungs, the lungs never opacified normally because only a small amount of contrast medium was delivered by each ventricular contraction. The concentration of Diodrast in the aorta after some of the dye had been shunted through the foramen ovale from the right auricle into the left was much less dense than that seen with early visualization of an overriding aorta such as occurs in the tetralogy of Fallot.

The catheterization findings of a reduced pulmonary blood flow and a right-to-left shunt are due to the shunting of unoxygenated blood away from the lungs through the patent foramen ovale into the left side of the heart. Although safely performed in one of our patients (Case 2) we feel that cardiac catheterization in patients with Ebstein's anomaly is not without danger. Because of the common occurrence of conduction disturbances, there is the possibility of initiating an arrhythmia which might prove fatal. Furthermore, there is the theoretical danger of entangling the catheter in the delicate, basket-like membrane or its fenestra-


tions. Finally, it is conceivable that the catheter might perforate the exceedingly thin-walled ventricular portion of the upper chamber, especially in a patient with a localized aneurysmal dilatation.

The Clinical Syndrome

The correlation of these clinical, laboratory, and pathological findings reveals that a distinct picture is produced when Ebstein's malformation of the tricuspid valve is combined with patency of the foramen ovale or with a gross defect in the auricular septum.

History: The onset of cyanosis is usually delayed. If present at birth, the cyanosis promptly lessens or disappears but returns at a later age. It is transient at first and insidiously becomes persistent. The cyanosis is more marked than the dyspnea, which is quite mild. There is easy fatigability. Although the patients tire quickly and often have to stop to rest, squatting is not a common habit.

Physical Findings: Outstanding features, in addition to the cyanosis and slight clubbing, are the enlarged heart, the left-sided chest deformity, the distant or muffled heart sounds, and often a gallop rhythm. There is a systolic murmur maximal at the left sternal border in the third intercostal space but audible all over the precordium. There may also be a diastolic murmur over the sternum, which may give the impression of a friction rub. The pulse pressure is narrow. The liver is slightly to moderately enlarged, but there are no pulsations palpable at its margin unless with terminal failure, and there are no other signs of tricuspid insufficiency.

Laboratory Findings: There is arterial oxygen unsaturation and compensatory polycythemia. The circulation time is prolonged. The electrocardiogram usually shows right bundle branch block and prolonged A-V conduction time. Fluoroscopy in the anteroposterior view usually shows a greatly enlarged heart with diminished pulsations. The tremendous size of the right auricle and right ventricle causes enlargement both anteriorly and posteriorly in the oblique views. There is no fullness in the the region of the pulmonary conus. A pulmonary artery of normal size is seen bilaterally,
but no expansile pulsations are visible therein. The lung fields are abnormally clear. The esophagogram upon barium swallow is normal.

Angiocardiogram shows a large right auricle and then an early but less dense concentration of the contrast medium in the right ventricle. The entire cardiac shadow visible in the anterior-posterior view appears to be formed by the right auricle and the right ventricle. The contrast medium extends nearly to the margin of the cardiac silhouette, indicating that the chambers are quite thin-walled. The Diodrast lingers for several seconds in the right auricle and the "auricularized" right ventricle, whereas the dye is quickly expelled from the functioning right ventricle into the pulmonary arteries. The opacification of the lungs is less than normal. A small amount of the contrast medium may be seen to pass from the right auricle into the left auricle, the left ventricle, and into the aorta. Cardiac catheterization shows a reduced pulmonary blood flow and an overall right-to-left shunt between the auricles. The pressure in the right ventricle distal to the valve is within normal limits.

Differential Diagnosis: This malformation is to be differentiated from other conditions in which there is cyanosis and an inadequate pulmonary blood flow. The most important malformations from which to differentiate it are the tetralogy of Fallot and valvular pulmonary stenosis.

The chief features which differentiate this malformation from the tetralogy of Fallot are the delayed onset of cyanosis, the absence of paroxysmal dyspnea and of squatting to rest, the cardiac enlargement, the diastolic murmur, the long circulation time, the electrocardiographic evidence of first degree heart block and of bundle branch block, and finally the angiocardiographic evidence of the enormous size and slow emptying of the right auricle.

Ebstein's malformation may even more closely resemble an isolated valvular pulmonic stenosis with patency of the foramen ovale and no defect in the ventricular septum than it does a tetralogy of Fallot. In a subsequent publication\textsuperscript{22} the clinical and laboratory findings of this type of pulmonic stenosis will be presented and the differential diagnosis discussed.

**Summary**

The clinical, laboratory, and pathologic findings in 3 cases of Ebstein's anomaly of the heart have been presented. This brings the total number of cases in the literature to 26. A correlation of the findings in the cases discussed in this paper and of those collected from the literature has demonstrated that this malformation is sufficiently characteristic to constitute a clinical syndrome which may be correctly diagnosed during life.

In this malformation the displaced tricuspid valve divides the right ventricle into two parts and thereby causes the proximal portion to be continuous with the cavity of the right auricle. The anomalous valve is so arranged, however, that it is competent. The myocardium of the right ventricle proximal to the malformed tricuspid valve is congenitally thin. The primary effect of the anomaly is to reduce the efficiency of the right heart. As the upper chamber cannot empty itself completely, it enlarges progressively. If the foramen ovale is incompletely sealed, it is opened, and venous blood is shunted from the right auricle into the left auricle and thence into the systemic circulation. The lower chamber, which receives less than the normal volume of blood, delivers an inadequate amount of blood to the lungs for oxygenation.

The outstanding clinical manifestations are the delayed and insidious onset of cyanosis, which is out of proportion to the mild dyspnea; the easy fatigability, and the infrequency of squatting to rest when tired. Physical examination shows excessive right heart enlargement, poor heart sounds usually associated only with a systolic murmur but sometimes also with a diastolic murmur and often with a gallop rhythm, and absence of signs of tricuspid insufficiency. The chief laboratory findings are the x-ray evidence of progressive cardiac enlargement and a concave pulmonary conus region and abnormally clear lung fields, the fluoroscopic visualization of diminished pulsations of the right side of the heart and absence of pulsations in the pulmonary arteries, the electrocardiographic signs of delayed A-V conduction and of right bundle branch block, the prolonged circulation time, the oxygen unsatu-
ration of the arterial blood, and the compensatory polycythemia. Angiocardiography is helpful in confirming the diagnosis and in this malformation is safer than is cardiac catheterization.

It is important to distinguish this malformation, which cannot be helped by present forms of surgery, from those such as the tetralogy of Fallot which are amenable to operation. The differential diagnosis is discussed.

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