CLINICAL CONFERENCE
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Presentation of a Case for Diagnosis
A Clinicopathologic Conference

This conference was presented at the Fourth Annual Heart Symposium sponsored by the Indiana Heart Foundation at the Indiana University School of Medicine, Indianapolis, Ind. on Jan. 20, 1955. Participating were members of the Research Committee of the American Heart Association and members of the Faculty of the School of Medicine. Participants from the Research Committee were: Dr. Robert H. Bayley of the University of Oklahoma College of Medicine, Oklahoma City; Dr. Howard Burchell, Mayo Clinic, Rochester; Dr. Howard Sprague, Harvard Medical School, Boston. Participants from the Indiana University School of Medicine Faculty were: Dr. James O. Ritchey, Department of Medicine, Moderator; Dr. Warren Coggeshall, Department of Medicine; Dr. John A. Campbell, Department of Radiology; Dr. Edward Smith, Department of Pathology; Edited by: Dr. Roy H. Behnke, Department of Medicine.

CASE HISTORY

THE PATIENT, a 45 year-old white female, was admitted to the Robert W. Long Hospital of Indiana University Medical Center on March 29, 1954 with the complaints of fatigue and shortness of breath. Past history, as related by the patient, revealed that she had experienced exertional dyspnea and cyanosis after exercise for an indefinite period antedating her enrollment in grade school. She denied squatting to relieve the dyspnea. At the age of 11 she was told that she had heart disease and during the succeeding years she led what she described as a "restricted" existence.

For seven years prior to her admission the patient had worked in a factory where she was able to perform her duties which required that she stand eight hours each day, but did not necessitate heavy lifting or walking. During the last two years she had been aware of a definite progressive increase in exertional dyspnea and the degree of apparent cyanosis. Symptoms of frank congestive heart failure, cough, paroxysmal nocturnal dyspnea and ankle edema, developed in December of 1953. She was then placed upon digitalis, mercurial diuretics and a salt restricted diet.

Physical Examination: The patient was a well developed, well nourished woman with moderate generalized cyanosis. The neck vessels were not distended. Blood pressure was 122/88, pulse 72 and regular. The point of maximal impulse was not located. One observer felt a faint systolic thrill in the pulmonic area. No precordial shock was felt. Auscultation disclosed an accentuated second sound at the cardiac apex. A grade II blowing systolic murmur was heard along the entire left sternal border, but with greatest intensity in the third left interspace. The second pulmonic sound was accentuated and louder than the aortic second sound. No murmurs were heard at the base.

Laboratory Findings: Urine analysis and blood serologic tests for syphilis were negative. Hemoglobin was 16.4 Gm. Red blood cell count was 5,640,000; white blood cell count was 8,500 with a normal differential.

Hospital Course: On April 8, 1954, the patient was taken to surgery. Her postoperative course was stormy and death occurred on April 10, 1955. An autopsy was then performed.

DR. J. A. CAMPBELL: On cardiac fluoroscopy generalized enlargement was seen in the frontal plane which, upon rotation, was found to be chiefly due to the dilatation in the area of the left ventricle. The right atrium and other border-forming cardiac structures were not remarkable. No calcification was seen in the pericardium, in the musculature, or in the region of the valves or larger peripheral vascular structures. The lungs were clear and the pulmonary
cause it was felt that she had a considerable right-to-left intracardiac shunt because of her cyanosis. The catheter entered the superior vena cava where saturation was 38 per cent and the pressure was found to be 3.2 mm. Hg, which is a mean pressure. The right atrial pressure was 7.5 mm. Hg, somewhat higher than is normally found. The right atrial saturation was 46 per cent. In the right ventricle we found pressure of 14/3 mm. Hg and a saturation of 47 per cent. In the right pulmonary artery, the pres-

vasculature was felt to be more difficult to define than normal.

In the frontal view (fig. 1) the left inferior cardiac segment is somewhat elevated and high with respect to the diaphragm, and this was taken to reflect left ventricular change in the way of dilatation. The aortic knuckle was normal. The aorta descended on the left in normal relationship to the barium filled esophagus. The vena caval borders were straight and full, but not excessively so. The right atrial border was satisfactory. The pulmonary arteries were somewhat smaller than normally expected, and the peripheral lung fields were considerably more clear than usual in a 45 year-old woman with cardiac dimensions of this magnitude. On the lateral projection (fig. 2) there is an increase in the anteroposterior dimension of the heart. It assumes a somewhat spherical globular configuration with the left ventricular contour making sharp impression on the esophagus but not excessively penetrating the posterior cardiac space. The frontal heart is tight against the sternum so that one might conceive of some right ventricular dilatation as well. There is no left atrial impression of significance and the aorta is within normal limits with no apparent calcification.

In the right anterior oblique projection (fig. 3) there is no suggestion of left atrial dilatation. Again the overall increase is primarily attributable to right ventricular enlargement. In the left anterior oblique (fig. 4) there is considerable prominence of the left ventricular segment and the right ventricular border also is prominent. The pulmonary artery is small.

Dr. W. E. Coggeshall: This patient was sent for catheterization on March 31, 1954 (fig. 5) be-

Fig. 1. Posteranterior chest film.

Fig. 2. Left lateral chest film.

Fig. 3. Right anterior oblique chest film.
sure was found to be 17/4 with 44 per cent blood oxygen saturation.

Arterial study showed the following: blood pressure was 91/62 and the blood oxygen saturation was 68 per cent without oxygen. With five minutes of 100 per cent oxygen, arterial blood oxygen saturation rose to 78 per cent, still markedly unsaturated. At that time we felt confident that the shunt was not from the pulmonary system into the aorta because of the marked difference in pressure between the lesser circuit and systemic circuit. The pressure relationships were such that a shunt at the ventricular level was thought improbable, so we felt that if a shunt were present it was at the atrial level. We did not traverse a defect in the atrium during this procedure and we could not enter the inferior vena cava.

Following that catheterization we felt that the inferior vena cava might be displaced and/or we might have a large common atrium. It was decided that the patient should have an angiocardiogram by means of a catheter in the inferior vena cava. Prior to doing the angiocardiogram, we decided to investigate the inferior vena cava and make sure that it emptied into the right atrium. The following findings were noted as we continued with the catheterization (fig. 6). Again, we found the right atrial mean pressure to be elevated—the saturations remained approximately the same. At that time we were able to enter the left atrium and a pulmonary vein where we found normal saturations. We found the right atrial pressure to be higher than the left, a relationship which is the reverse of normal (fig. 7). At that time the catheter also went into the left ventricle where we found a mild increase in saturation when compared to the saturation in the left atrium. This would be compatible with a venous shunt at the atrial level combining with the oxygenated blood coming from the pulmonary veins. At that time we felt that we had established that there was a shunt through the atrial septum. We were not positive why the pressure was elevated in the right atrium. The right atrial pressure was ex-

Fig. 4. Left anterior oblique chest film.

Fig. 5. Cardiac catheterization data from first procedure done via the superior vena cava, March 31, 1954.

Fig. 6. Cardiac catheterization data from the second procedure done via the inferior vena cava, April 4, 1954. The catheter did pass into the left heart through the atrial wall.
amined closely for a pressure pattern suggestive of tricuspid insufficiency, but none was found.

Discussion

Dr. H. Sprague: May I ask one question even though the answer seems to be implicit in this history? Was the patient operated upon for her heart abnormality?

Dr. J. O. Ritchey: You have the answer to this, Dr. Close?

Dr. W. D. Close: Yes.

Dr. H. Sprague: The reason I asked this of my friend is that it permits me to offer a few insulting remarks. If I were not unhappy about this case, you would not have any fun at all. I assure you I was very disturbed about this situation. I must say at once that I had recourse to our competent catheter team and especially Dr. Allan Friedlich to see what they thought might be the possible interpretation of these figures. As I intend to go ahead and stick my neck out and make a diagnosis I will have to assume a few things and issue the following insults. (1) If my diagnosis is correct, the patient probably should not have been catheterized. (2) Since the patient was catheterized, I must say that either the catheter figures are wrong or they were misinterpreted; otherwise the patient would not have been operated upon for the lesion, which I think this patient had, has been considered inoperable.

Now you see these are all assumptions. Let us review the history a moment. The patient had fatigue, dyspnea and cyanosis for as long as she could remember, especially exertional dyspnea and cyanosis even before she went to school. There is no statement as to whether she was a “blue baby” or not. She denies squatting. That is included because, if the patient did not squat, one may assume that this is probably not a case of Fallot’s tetralogy. That is not completely accurate, of course. At the age of 11 she was told she had heart disease; therefore, we know that something went wrong a long way back and we assume that the cardiac condition was something with which this child was born.
She led a restricted existence but did rather well; she was working and she was 45 years old. For seven years prior to her admission she had worked in a factory and was able to keep on with her job and stand for eight hours a day. There was no heavy lifting or walking connected with her work, but she became more and more short of breath when she exerted herself; the cyanosis got worse, and finally she went into congestive failure.

I would judge that the physical examination did show moderate cyanosis. I understand that there was no clubbing of the fingers or toes; neck vessels were not enlarged and not dilated or pulsating. The blood pressure was normal. There might have been a faint thrill in the pulmonic area where she had an accentuated second sound louder than the aortic second sound. We frequently find difficulty in correlating accentuation of pulmonic second sound with catheter evidence of pulmonary hypertension, which this person did not have. There was a grade II systolic murmur along the left sternal border and no murmurs at the base. She did have a high hemoglobin and a red blood cell count of 5.6 million and right bundle branch block in the electrocardiogram.

The cardiac fluoroscopy, which you have described, may I say very cautiously—showed enlargement in the region of the left ventricle, right ventricle, and left atrium. For example, when you get right bundle branch block and a heart which, when looked at sideways is tight against the anterior chest wall, you can be sure that there is a big right ventricle; and as you know the radiologists often sneak into the electrocardiographic laboratory and look at the electrocardiogram before they say whether the right or left ventricle is enlarged. It is conceivable that we are dealing with a big right-sided heart here that is just pushing the left side of the heart back and to the left. The striking thing is that the arterial trunks are small, as evidence here of decreased pulmonary flow rather than increased pulmonary flow.

The catheter data will, no doubt, be further discussed later. As the catheter passes into the right atrium from the vena cava, we apparently find more oxygen in the blood which must come through the interatrial septum, or by anomalous position of the pulmonary venous drainage into the right atrium, or conceivably retrograde in some particular fashion; but it would have to be a rather peculiar fashion. We find, as we go along into the right ventricle, no hypertension of that chamber and further no hypertension in the pulmonary artery. When we get the figures for oxygen saturation, we wonder where in the right side of the heart the end of this catheter actually was. Well, as our catheter friend has said, there seems to be an interatrial septal defect. If that is so, why isn’t there evidence of an increase in pulmonary circulation? There doesn’t seem to be any evidence of an interventricular communication for the only increase in oxygen of significance occurs there in the right atrium. It doesn’t seem to make sense for a pulmonary stenosis. There is certainly no right ventricular hypertension; there is no poststenotic dilatation of pulmonary artery; for example, and no great difference in pressure gradient across the pulmonary valve. It doesn’t seem to make very good sense for a tetralogy of Fallot. Sticking my neck out, as I say, it does make sense for one condition—a situation which I myself have never diagnosed during life; which indeed, was said to be impossible of diagnosis some years ago. However, when Dr. Paul Wood came over from London two or three years ago, he said it is an easy bedside diagnosis in children, and the only patients I have seen with it were children. As all of you now know, since you probably have made the diagnosis already, this is presumably "Ebstein’s disease" or "anomaly" of the tricuspid valve.

There is a big right-sided heart consistent with Ebstein’s disease. In this condition the tricuspid valve is displaced downward into the right ventricle with a very small functioning right ventricle beyond the tricuspid valve and a great big dilated thin-walled part of the right ventricle proximally, communicating directly with the right atrium. It is characterized by cyanosis which usually appears at birth and then disappears, only to reappear later as the prolonged malfunction of this abnormal tricuspid valve results in increased back pressure blowing open the foramen ovale. The foramen may increase in size over the years to the point
where it becomes a large defect. The decrease in the flow to the lungs is explained by poor forward output of the right ventricle. These catheter findings seem to me, at least, to be consistent with what is going on in this big chamber composed of the right atrium and the upper part of the right ventricle.

The reason that I had for some of these insults I mentioned was that if it was assumed that this was a case of Ebstein's disease, catheteterization might be considered to be dangerous because arrhythmias can be initiated easily by catheterization in this condition and the catheter might get tangled in this rather complicated, deformed, tricuspid valve. Certainly there is supposedly some danger of perforation of the thin chamber.

According to Taussig's group (I have not had a chance to look through the literature) one patient has been known to live to the age of 61 and one to 60 years, but the average age at death is 24 years. So far as I know, this entity has not been diagnosed definitely during life, although one case, three years ago and two cases two years ago, were supposed to have been diagnosed, two by catheter and one by angiocardiogram. I can not offer a diagnosis that seems to me better than Ebstein's anomaly and if I am wrong, I will go down smiling; but if I am wrong I really think I should turn in my badge and gun.

Dr. R. H. Bayley: I would like to take a peek at that electrocardiogram (fig. 8). Right bundle branch block in congenital heart disease often offers us additional information, over and above what we see in a more normal electrocardiogram. In uncomplicated right bundle branch block we look for a double R wave, two R deflections on the right side of the precordium. The first of these two R deflections is written by the electromotive forces passing through the septum from the left side only. Following this is a negative deflection which is written by the electromotive forces in the free wall of the left ventricle. For example, when there is a left ventricular hypertrophy, we expect this cleft to be rather prominent. And finally, the right ventricle which is activated last in right bundle

![Fig. 8. Complete right bundle branch block: QRS = 0.15 second](image-url)
branch block is responsible for the final R deflection. So in a sense, we have a separation of the deflections written by the two ventricles in right bundle branch block. If there is a large hole in the septum, or if that septum has been infarcted in the presence of right bundle branch block, the first R deflection disappears. The first R deflection in the first two precordial leads of this patient’s curve is missing. However, the second R deflection here is quite broad and prominent; we might surmise that this could be an instance of right ventricular hypertrophy. I would be much less sure of the septal defect than of right ventricular hypertrophy.

On the left side of the precordium we can interpret the initial ventricular deflections equally satisfactorily when right bundle branch block is present or when it is absent. In other words, if left ventricular hypertrophy is present here, we would expect those deflections to be rather prominent and actually they are quite small. So, I think the electrocardiogram is very much in favor of right but not left ventricular hypertrophy and we still have to explain the absence of the initial R deflection which is ordinarily written by the septum.

We have seen a case in which there was a marked hypertrophy of the right ventricle but the right ventricle was not very large. It was a very thick-walled, small chamber, and the tricuspid valve was in essentially normal position. There was, in effect, a very small cavity in the right ventricle and we obtained pressure distributions in that particular case which were entirely similar to the pressure findings presented today. An auricular septal defect was also present. There was this difference clinically; our patient was in his thirties, he was considerably more cyanotic than this patient and he had been cyanotic from birth. Except for the grade of cyanosis, this picture would fit with that of a markedly hypertrophied small right ventricle with atrial septal defect, not necessarily Ebstein’s disease.

DR. H. BURCHELL: I think that this patient does have Ebstein’s disease. I am going to borrow a sentence from my friend, DR. Frances Wood. If this patient does not have Ebstein’s disease, then DR. Ritchey did the post-mortem on the wrong patient. I think that these particular findings present a clinical picture which allows a specific clinical diagnosis: These findings are an atypical type of right bundle branch block, a peculiar shaped heart with very poor vascular markings in the lungs and then a picture of heart failure and/or cyanosis in the second or third decade of life. Some patients have a characteristic double murmur in the pulmonary area which may be timed by the phonocardiogram as being related to auricular and ventricular systole. DR. Paul Wood has said that it is dangerous to catheterize these patients, but I do not believe this is so. In some patients we have not catheterized, the clinical diagnosis was enough, but we have felt in the other cases that it would be well to know the hemodynamic alterations that were present, because there is considerable variation in Ebstein’s deformity from heart to heart.

The deformity mainly relates to one part of the tricuspid valve which may be a very long veil-like arrangement which may be displaced markedly toward the outlet side of the ventricle. The right ventricle may actually be a pouch, so if you cut across it you might see two chambers separated by this veil-like structure somewhat similar to the tricuspid valve arrangement in the heart of birds. Some of these valves are competent and some are incompetent. I think DR. Taussig has pointed out that the right ventricular failure is related to the inadequate size of the right ventricle; however, I would like to point out that there is more of a variation than that. Sometimes there is perhaps a true right ventricular stenosis from the point of view of the inadequate size of the right ventricular chamber. Sometimes the chamber is inadequate and sometimes the tricuspid valve is incompetent and sometimes it is not. In this case, the mean pressure in the right atrium could be apparently increased over the inferior and superior vena cava for two possible reasons: you might have a tricuspid pulse in the right atrium which caused an error in the calculation of the mean pressure or the catheter might have been a little bit further along and part way into this unusual, right-sided chamber.

We have seen one patient who had postural dyspnea, marked effort dyspnea and cyanosis. We studied him over a period of 18 months and
this symptom-pattern was constant and his arterial saturation on exercise, or sometimes just standing, dropped to levels below 50 per cent. After considerable thought we said this man would be better off with his foramen ovale closed. One can predict that the interatrial communication is likely to be the result of a patent foramen ovale, rather than a large atrial septal defect. This patient was operated upon, had an uneventful recovery and has been normally active for two years. He was a rabbit hunter and he is now able to go back doing that. However, this is regarded as a palliative procedure. We have restored this man to well being for a period of two years, but it is expected that eventually the heart will fail. In another individual, a 35 year old sheet metal worker who has been getting along quite well, we have not recommended any surgical procedure. One patient who came to us in congestive heart failure was observed intermittently for two years before his death. Our total experience would indicate that perhaps in certain individuals without heart failure, where hypoxia plays a significant role in symptoms, one might advise surgical closure of the atrial septum. We operated upon one child two months ago who had fainting attacks. The operation was successful in that the arterial oxygen saturation came up to normal with closure of the atrial defect. The child died, not recovering consciousness after surgery; there was cerebral infarction probably from an embolus. Children or adults with this disease must be regarded as very high surgical risks, but there is a possibility that there may be a small number of them who present primarily a hypoxic picture who could be given surgical palliation.

Fig. 9. (Autopsy No. 7057).: Ebstein's malformation of the tricuspid valve and a patent foramen ovale. An interior view of the right atrium and ventricle is shown. The black line indicates the expected normal line of attachment of the tricuspid valve. The white line indicates the actual line of attachment of the tricuspid valve in this case. The size of the atrium above the white line is increased at the expense of the size of the ventricle below. Note also the partial attachment and irregularity of the posterior leaflet (P) and the anterior leaflet (A), both of which are fenestrated. The medial leaflet (M) is short enough to suggest that an insufficiency of that leaflet may have existed. The foramen ovale (FO) is patent and measures 15 by 20 mm. The pulmonary conus, valve and artery (PA) are essentially normal. The left atrium, left ventricle and associated structures are normal except for the patent foramen ovale.
Dr. E. B. Smith: May I summarize the clinical problem as it pertains to the pathologic findings? The patient was cyanotic, had an increased pressure in the right atrium and a route for the shunt of blood from the right to the left side of the heart. Ebstein's malformation has been correctly proposed to explain the clinical and laboratory findings. The lesion of the heart is described in the legend to figure 9.

At the time of the postmortem examination the foramen ovale was closed by silk sutures which had been placed there two days before death. Dr. Harris B. Shumacker had invaginated the anterolateral wall of the right atrium across the base of the right auricular appendage. He had then neatly sutured the invaginated wall to the margins of the foramen ovale to cover it completely. The sutures were in place and the foramen was still occluded at death.

In order to prepare an informative photograph, the sutures were removed from the specimen postmortem. The heart weighed 455 Gm. The right ventricular wall was three mm. in thickness and the wall of the left ventricle was 12 mm. in thickness. The circumference of each of the valvular rings was as follows: tricuspid, 18 cm.; pulmonic, 8.5 cm.; mitral, 11.5 cm.; aortic, 8 cm. The weight of the lungs totaled 720 Gm. The postmortem examination was limited to dissection of the thorax. According to our records there was a “tendency to clubbing” of the fingers of this woman. Except for the lesion of the heart, no congenital abnormalities were observed.

This is a rare congenital anomaly of the tricuspid valve, first described by Ebstein in 1866. Less than 30 cases have been reported. In most instances the foramen ovale is patent. The clinical diagnosis is difficult and has rarely been made. I congratulate Dr. Sprague upon his correct diagnosis.
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