Unusual Forms of Heart Disease

By Howard B. Burchell, M.D.

I AM in general accord with Hutchison’s “Don’ts for Diagnosticians,” the first two of which read, “Don’t be too clever” and “Don’t diagnose rarities.” Hutchison¹ himself quoted Samuel Gee, who said, “Common things most commonly occur,” and it may be emphasized that many rare cardiac conditions perhaps could be all but forgotten, without loss, in day-to-day cardiologic practice. Unusual manifestations of common diseases probably mislead the physician more often than do unusual diseases. Rare conditions tend to be the spice of practice, however, and constitute the substratum whereon one’s probing wits are tested. To a certain extent, also, missing of the diagnosis of a rarity is the bête noir of the specialist.

Moreover, in this era of rapid transportation, the patient who has sought diagnosis without success often travels far looking for special help. Thus, by force of numbers, a rarity becomes less an event in a large institution than in general practice. This fact is well known and is well exemplified in the discussions of consultants who participate in the “Weekly Clinicopathological Conferences” based on “Case Records of the Massachusetts General Hospital” and reported in The New England Journal of Medicine. Some readers of these reports, whose native language is other than English, must be perplexed by the high incidence of symptoms and signs which are designated as possible “red herrings.” And the mystification of such foreign visitors would be fully as deep if they were present when one or another of us, under the duress of a similar conference, became, and was called, “as independent as a hog on ice.”²

In cardiac clinicopathologic conferences which I attend, of the difficult cases chosen for presentation, some are selected because the diagnosis originally was missed, partly or entirely. We believe there is no “news leak”; nevertheless, by the time the clinical part of the conference is over, at least three-fourths of the cases are accurately assessed. Thus, in our clinic at least, if patients who constitute diagnostic enigmas could be tagged at the time of registration, “Potential Conference Material,” and if they could appear at one of the clinicopathologic conferences, the diagnostic accuracy of the consultants would increase several fold.

The unusual cases wherein the cardiologist might be at a loss for a diagnosis may be classified as follows: (a) rare anatomic conditions, for example, Ebstein’s malformation of the tricuspid valve, traumatic rupture of a valve, primary neoplasm; (b) heart affected as an integral part of a general disease, for example, amyloidosis, lupus erythematosus, scleroderma, hemochromatosis; (c) cardiac complications of other disease, for example, diphtheria, purpuric pericarditis, sickle cell and other anemias, thyrotoxicosis both overt and occult, arteriovenous fistula, myocardial metastasis; (d) conditions precipitating or accelerating the development of more usual forms of acquired heart disease, for example, xanthomatosis, possibly obesity, myxedema; (e) conditions mimicking primary heart disease, for example, thrombosis or involvement of the inferior vena cava by tumor.

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⁠² This phrase is attributed to William Osler.
The foregoing classification could be followed in this presentation although there are many other approaches. A somewhat frantic person, searching for novelty, might write “A Cardiologist’s ABC of Rarities,” thus: A is for amyloid, B for beriberi; C is for contusion, D for diphtheria; E is for Ebstein’s, F for Fiedler’s; G is for glycogen storage, H for hemochromatosis, and so on. For the privilege of being represented by certain of the letters, there would be competition among several conditions. Rare conditions would come to mind to correspond with every letter except J and Z. That approach admittedly is childish but, since it has been made, it could be followed ‘out, spottily, in rhyme:

Don’t fail to note, in amyloid
Abundant syndromes are deployed;
And always with a heart contusion
Arise both doubt and much confusion.

The palsy of diphtheria
Must not be thought hysteria.

In hemochromatosis we
Demand hepatic biopsy.

That, I hope, Dr. Bett3 would admit as “judicious levity.” At all events it is not entire nonsense and what follows is serious. In any complete listing of abnormal cardiac conditions, many would fall into the category of controversial states. The question whether some of them actually exist often would have to be answered: “Perhaps,” or “Perhaps; probably.” To many other questions positive answers cannot be given; here are some of the questions: Can a thyroid nodule be responsible for auricular fibrillation in the absence of signs of hyperthyroidism? Can auricular fibrillation, which is present for many years, of itself result in heart failure or cardiac enlargement? What are the basic faults in conditions called presbycardia or kyphoscoliotic heart disease (without pulmonary disease)? Could there be a basic physiologic and anatomic defect in the form of anxiety neurosis, still frequently termed “neurocirculatory asthenia”?

The first condition on the alphabetical list is amyloidosis. This is not a particularly rare condition; I see several cases a year. It appears in either a primary or a secondary form. In the former the heart is often seriously affected and heart failure is the predominant feature. Primary amyloidosis may be chiefly manifest in any of many organs or tissues, particularly in the heart, liver, kidneys and peripheral nerves. The similarity of the picture to that of chronic constrictive pericarditis has been emphasized and, in the two conditions, the same type of curve of abnormal filling of the ventricle may be seen.4 In the presence of disease of many organs the diagnosis of primary amyloidosis should be particularly considered. The diagnosis is best confirmed by needle biopsy of the liver. Some of the problems encountered when a patient presents himself with the disease were represented in a clinical conference recently arranged by Pruitt, Daugherty and Edwards.4

The term “amyloid,” from the Greek word for “starch,” was coined because of the avidity with which the abnormal substance takes up iodine, as does starch. This property aroused in Hanson and McConahey,6 of our clinic, the hope that a tracer study with radiiodine would be diagnostically useful. Their hope has been substantiated in an occasional case but not in others. In two of four cases the extrathyroidal disposal rate was high, and in one of these radioactivity over the liver and spleen was increased.

Even secondary amyloidosis, although sparing the heart, may not necessarily spare the heart specialist of a consultation. A case in point (and it may be used as the basis of a plea against too early and too rigid subspecialization) is the following:

A middle-aged man complained of abdominal pain and diarrhea of two years’ duration. In addition, he had lost weight, was weak and his legs were edematous. Because of the last, a cardiologist was asked to see the patient. The primary diagnoses were regional ileitis, ileocecal fistula and possibly pelvic abscess. Surgical operation was necessary in any event. In spite of appropriate treatment, however, the patient died on the second day after operation, with azotemia and in shock. On postmortem examination, amyloidosis, mainly of the kidneys and adrenal glands, was found. It is of interest that these diagnoses were tentatively made in the clinicopathologic conference, before the postmortem diagnosis was disclosed.
In my “ABC of Rarities,” under B is found beriberi. In the northern Mississippi Valley, this condition is rare indeed. In spite of our suspicions when individuals, particularly alcoholics, present unusual types of heart failure, I believe there has not been a single established case of beriberi heart failure in a Rochester hospital in the past 10 years.

Another disease in association with which heart failure may be the presenting symptom is hemochromatosis. In about a fifth of the 13 cases in which the diagnosis has been made in our clinic, heart failure has been present. The quantity of pigment in the myocardium at times has seemed sufficient to be responsible for the heart failure. However, other contributing factors, such as coronary disease accelerated by the diabetic state, or possibly anemia from bleeding esophageal varices, must be suspected.

In an early paragraph of this article unusual cases in which the cardiologist might be at a loss for a diagnosis were classified. The conditions just discussed were in the second category of that classification; that is, the heart was affected as part of the general disease. The first category of the classification included rare anatomic conditions and here, as an example, I have chosen Ebstein’s malformation of the tricuspid valve. At the clinic we now have recognized nine cases. In this condition, the ability of the right ventricle to impel an adequate quantity of blood into the lungs is impaired by reason of a misplaced, deformed tricuspid valve. Sometimes there is tricuspid insufficiency, sometimes not. The characteristics of the syndrome are varying cardiac enlargement, clear lung fields and low pressure in the right side of the heart as determined by cardiac catheterization. The electrocardiogram not infrequently gives evidence of right bundle-branch block. In the majority of the cases there is an associated atrial septal defect, which allows a right-to-left shunt, and resulting mild to moderate cyanosis. If the patient is seen before heart failure has entered the situation and the shunt is copious, closure of the atrial septal defect may be considered. In Rochester this was done in one case and the patient, who previously had been nearly an invalid, has been able to lead a normal, active life. After the operation, arterial oxygen saturation became normal. Sometime in the future heart failure may develop, but, from the point of view of amelioration of the condition, the operation has been an outstanding success.

In passing, mention may be made of one congenital condition which, previously thought to be rare, is now recognized as relatively common. This is isolated pulmonary stenosis, perhaps more properly called “pulmonary stenosis with intact ventricular septum.” In some cases there is obvious, or gross, shunting of venous blood through atrial septal defects; in others, the shunt is minimal and is demonstrable only by special technics, such as central injection of dye; in some, the shunt may be considered only potential; in still others, the shunt is left-to-right through an atrial septal defect; finally, in some cases the atrial septum is intact and no shunting of blood is possible.

Among anatomic rarities, the cardiac and vascular defects associated with Marfan’s syndrome and the syndrome associated with anomalous origin of the left coronary artery from the pulmonary artery should be remembered. Another anomaly, one in which surgical measures may be lifesaving, is a form of obstruction to venous outflow from the lungs; it thus simulates mitral stenosis, and is called “cor triatriatum.” Edwards and I believe the condition is related to congenital stenosis of a common pulmonary vein. Our patient was an infant, but in Borst’s case the patient lived to adulthood and, incidentally, the auscultatory findings were those of mitral stenosis.

To depart from the order of the classification, other conditions that have simulated mitral stenosis are worthy of mention. Such conditions are acquired stenosis of the pulmonary veins and left atrial tumor. The case of the former condition with which I am familiar is believed to be particularly pertinent to the pathogenesis of arterial lesions of the lung, because the arterial lesions were predominantly in the lobes which corresponded with the more marked venous obstructions. The left atrial tumors which I have in mind are polypoid masses which usually are designated as myxomas. But some of these masses might be considered, by some pathologists, not to be
true tumors but organized thrombi. That the one can mimic the other is unquestionable. Particularly in the presence of a normal mitral valve, however, a cellular mass with a stalk, accompanied by deep disruption of the atrial wall, is a tumor, both in my opinion and in that of Pritchard,\textsuperscript{11} who studied a large series.

In a number of cardiac states a patient’s condition may suddenly change. Among these states are spontaneous rupture of valves, of chordae tendinae, or of aortic sinus aneurysms. It is considered best not to use the word “spontaneous” in relation to that type of rupture of chordae tendinae which occurs after myocardial infarction. In one case of mitral insufficiency reported by Becker, Edwards and me,\textsuperscript{12} the date of rupture seemed established and the patient lived approximately a year. Within this type pulmonary arterial changes developed.

Edwards\textsuperscript{13} mentioned another most unusual case of corrected complete transposition of the great vessels in which the left atrioventricular valve simulated the tricuspid valve and was insufficient.

Aortic sinus aneurysms frequently rupture into the right side of the heart and a continuous bruit, like that of patent ductus arteriosus, results. The sudden onset of weakness or dyspnea coincident with the appearance of such a bruit is clinically diagnostic. Substitution may be had from cardiac catheterization, whereby arterial blood is obtained from the right atrium or ventricle. Aneurysms usually are in either the right anterior or the posterior aortic sinus. Those in the former situation not infrequently are associated with a ventricular septal defect. When rupture in this area occurs, it is into the outflow tract of the right ventricle, whereas lesions in the posterior aortic cusp rupture into the right atrium. As a corollary, when “arterialization” is found in the right atrium, the lesion, as compared with an anterior one, should be more amenable to surgical treatment, either from the atrial or the aortic side, because it could be more easily located and its origin would not be associated with the origin of a coronary artery; the posterior aortic cusp is the “noncoronary” one. It is well to emphasize that a rupture with a remarkably small orifice (2 to 3 mm.) may give rise to heart failure in this condition. Kirklin\textsuperscript{14} has unsuccessfully attempted to close one such rupture of a posterior aortic cusp aneurysm through a right atrial approach.

Idiopathic pulmonary hypertension remains an enigma from the etiologic viewpoint. At present, its rapid course makes it seem unlikely that it bears any direct etiologic relationship to systemic primary hypertension. If primary pulmonary hypertension ever is akin to the systemic type, it very rarely is so. When severe pulmonary hypertension develops, whether from pulmonary arterial lesion including emboli, or perchance from primary pulmonary hypertension, and right heart failure ensues, there may develop a cyanotic condition related to functional opening of a foramen ovale. This condition has been seen at the clinic on a number of occasions and, by the dye indicator method, the site of a right-to-left atrial shunt clearly has been demonstrated.\textsuperscript{15}

The term, “idiopathic acute pericarditis” and the condition it denotes may be regarded as relatively commonplace. It is important to emphasize, however, that diagnostically, the term is indefinite. It is well, always, to consider etiologic possibilities and, if reversion to health or of the cardiac silhouette to normal size is delayed, pericardial biopsy should be considered. By such a measure granulomatous or neoplastic disease may be discovered in time to allow appropriate management to be instituted.

Occasionally heart failure is related to systemic arteriovenous fistula. The last case of this sort encountered in Rochester was that of a young man who was referred there because he was in desperate straits from the effects of a fistula involving the aorta and inferior vena cava, which had been recognized following operation for a protruded lumbar disk. Kirklin\textsuperscript{14} successfully performed aortic graft and the patient has returned to normal health. Since the report of Edholm and associates\textsuperscript{18} on heart failure in association with Paget’s disease, cases of this disease have been observed at the clinic in which there was a high normal cardiac output, but in none of the cases was the heart failing. In one case in
A rare condition superficially suggesting heart disease is that related to a renal tumor, with invasion and obstruction of the inferior vena cava. One of these cases is found each year or so at the clinic. It is possible for death to occur from a huge pulmonary embolus consisting of tumor tissue. Occasionally cases of phlebothrombosis of the lower extremities may be transiently suspected of being cases of heart failure. In this connection it is worth re-emphasizing that apparently idiopathic venous thrombosis carries a high correlation with neoplastic disease, particularly with that of the pancreas. Absence of cervical venous engorgement practically should exclude heart disease as the underlying cause of edema of the leg related to obstruction of the inferior vena; yet it is remarkable how often this simple sign is not sought. Even in the reverse situation—that is, when cervical venous engorgement is present and crying for recognition—it may be disregarded for a long time. Consequently, chronic constrictive pericarditis may be labeled, erroneously, portal cirrhosis.

In spite of many advances in knowledge of heart disease, patients who come to the physician with symptoms of circulatory deficiency and enlarged hearts, may improve, depart, and diagnosis never may be established in their cases, not even by the regrettable method of postmortem examination. It is easy to label such cases rheumatic or viral myocarditis, but there is an intellectual dissatisfaction in complete acceptance of such diagnoses. Viral myocarditis as an entity is well established but certainly is a rarity, difficult of exact etiologic substantiation and difficult of assessment as regards the amount of histologic change to correlate with the clinical picture.

Much is left untouched in this survey of the cardiologist's role in recognition of the unusual syndromes with which he may be confronted. For instance, the whole field of unusual electrocardiograms has not been entered because, although they are important, it is only occasionally that they are related directly to the patient's presenting symptoms. Clinical practice often gives evidence of the truth of the old adage that to think of a rarity is practically to exclude it. Particularly in teaching institutions, and especially in the early years of teaching, an unusual problem offers a temptation to adopt desperate measures in the attempt to obtain a diagnosis. It is best sometimes to leave the diagnosis indeterminate, particularly when tests that might be employed entail danger or worrisome expense and cannot lead to definitive treatment. As Jeans has been quoted as saying: "A living problem is better than a dead certainty."

An ABCD of rarities ought to end with W, X, Y, Z. This one can so end if W is allowed to stand for work hypertrophy, X for xanthomatosis, Y for yellow fever heart, and Z for zootomic hearts. Then, with some license as to stressed syllables, the following so-called couplets emerge:

Work hypertrophy may have been found
In coolies pulling jinrickshas round.
A precocious status anginosis
Is one that's called xanthomatosis.
An evil virus, at least in part,
Is the cause of yellow fever heart.
Last come the strange zootomic hearts,
Not known or seen within these parts.

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