Possibly Unrecognized Forms of Heart Disease

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The title of this presentation might imply that many forms of heart disease were possibly escaping recognition. Perhaps a more hackneyed title, such as "Pitfalls in Diagnosis," "Missed Diagnoses," or "Reflections on Cardiac Diagnosis," would be more appropriate.

At the end of the month or semester, one may reflect on his practice and figuratively calculate his diagnostic batting average; the analysis of his errors, minor or major as they may be, can be a humbling, yet rewarding task. Obviously, all personally unrecognized, or erroneously labeled, forms of heart disease cannot be evaluated during such reflection. It is necessary to have a final diagnosis, obtained either on a subsequent occasion or by a colleague in either a clinical or a pathologic, anatomic setting.

General categories of missed diagnoses are: (1) rarities, (2) simulators, (3) complicators, and (4) the emotionally obscured.

Rarities

These may be unknown to the physician or even yet to be described. Each specialist undoubtedly sees a patient, each week or so, who presents a picture so puzzling that he may wonder if some new syndrome is teasingly waiting to be identified. Are there disease states equivalent in rarity to the carcinoid syndrome, primary aldosteronism, renal artery fibromuscular disease with hypertension, and pheochromocytoma that are escaping our understanding? Rarities seem to become commonplace once accurately described.

One must also remember disease states that one may produce ("diseases of medical progress") of which a neat cardiovascular example is that of the unique type of anemia related to fragmentation of red cells,1,2 assumedly related to their impact on Teflon prostheses surgically placed in the heart.

Rarities when recognized can be fun, as illustrated by two recent cases: Hypoparathyroid hypocaleemic laryngospasm was mistaken for paroxysmal left heart failure, and liquorice-induced hypokalemia was initially thought to be aldosteronism.3 Pheochromocytoma having been mentioned, it is pertinent to remark that the clinical diagnosis may sometimes be most difficult to make, for a patient’s presenting complaints may be bizarre. For instance, nervousness, sweating spells, or transient shock may occur in the absence of hypertension or, at least, when hypertension has been overlooked.

The cardiologist, to justify his appellation, must be abreast of the times in regard to eponyms and syndromes that abut his specialty. He must know about such syndromes as pseudoaxanthoma elasticum4-7 and angio-keratoma corporis diffusum universale, where vascular disease may be rampant. However, as Hutchison8 said, we must not mistake a label for a diagnosis and be content with descriptive rather than etiologic classifications. The example of pseudoaxanthoma elasticum was chosen not because of its exotic features and rarity but to introduce the increasing importance of the genetic background of patients. The science of genetics looms ever

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larger on the horizon of medical practice; a practitioner cannot expect to master its intricacies, but he can expect to do yeoman service in the collection of data on the forebears and relatives of his patients.

It is trite, but necessary to say, rarities are rare—common things most commonly occur. At times, because of enthusiasm and perhaps of snobbishness, physicians in a specialty practice may have a tendency to overdiagnose rarities. However, the missing or nonrecognition of a common condition in unusual guise is more humiliating than the missing of a rarity—even for a specialist.

Schultz, Brown, and I recently reviewed a series of records of aged people who died of heart disease—the overwhelming majority died of coronary disease and a large number of acute infarctions. There was an almost monotonous repetition of descriptions of coronary sclerosis in the autopsy protocols pointing out that coronary atherosclerosis is a killing disease in the aged as well as the middle-aged. Nevertheless, its clinical expressions may not be wrapped in a neat package, and it may not be readily diagnosed. Age per se should be no barrier to the diagnosis of conditions more readily associated with the young, for example, myocarditis, tuberculosis, and idiopathic pericarditis.

Simulators

In this category of diagnostic problems, data from the history, physical examination, roentgenologic examination, electrocardiogram, or special laboratory procedures may indicate the wrong disease. I intend to dwell at some length on this type of potential error in relation to valvular defects, but a few examples of simulators follow.

In respect to history, pain suggestive of myocardial infarction may be due to acute pericarditis. As yet it is not generally known, I believe, how suddenly the pain of pericarditis occasionally may begin and that it may be associated with a syncopal reaction or transient shock, presumably neurogenic and related to the vasodepressive receptors in the epicardium.

In respect to general aspects of physical examination, increased venous pressure might result from obstruction of the superior vena cava in a patient with a normal heart. A clue which may be helpful in this condition is the bluish suffusion of the face when the patient is recumbent; this may be mimicked, in the ambulant patient, by only a few other conditions, as constrictive pericarditis and tricuspid stenosis. This brings to mind constrictive pericarditis manifested by icterus, which may so mimic portal cirrhosis that the physician may treat the patient for cirrhosis for considerable periods—the descended veins in the neck cry for recognition, but they are out of the physician’s focus as he directs his attention to the abdomen. In a rare instance, constrictive pericarditis may simulate another condition (really associated), namely, nephrosis with gross albuminuria and hypoproteinemia.

In respect to auscultation, examples of confusing findings may be illustrated by two conditions which have attracted a great deal of interest in recent years. These conditions are aortic subvalvular muscular stenosis, which may be mistaken for mitral incompetence, and mitral incompetence related to rupture of chordae to the posterior mitral eaflet, which may readily be mistaken for aortic stenosis.

Intraaortic Muscular Stenosis

This disease, the dynamic aspects of which were pointed out by Broek and which was, within a few years, the subject of many papers and an editorial, deserves discussion in some detail. It is probably the most intriguing condition whose hemodynamic aberration has recently been identified. I emphasize the hemodynamic identification, not clarification, as in many aspects the nature of the fault is still mystifying and tantalizingly obscure. A number of features are generally agreed upon: 1. There is a trend toward familial incidence—remarkable family histories have been published. 2. There is rapid progression of...
hemodynamic obstruction in some cases and absence of progression in others. 4. There is a tendency for clinical expression to occur at different ages. 5. There is a possibility that hypertension, or more particularly ‘‘myocardopathy,’’ may initiate or accelerate the obstructive features. (At present, confusion exists regarding how obstructive primary myocardial disease may be, and familial cardiopathies may not be, associated with obstruction.) 6. There is a false paradox. Angiocardiographically, relative to sites of obstruction. 7. There are phantom qualities of the pressure gradient. (This last characteristic was forcibly brought to my attention years ago when a patient with a measured gradient was operated on—on the operating table, the surgeon could demonstrate no gradient.)

The condition gains interest because of many paradoxes. Here is a disease which is often lethal, sometimes suddenly so, but in many instances without, in my opinion, evidence of cellular disease. One could refer to the condition as ‘‘the heart against itself,’’ for, according to present concepts, the more healthy and vigorous the myocardium, the more obstruction is created to the outflow of blood. Gross hypertrophy of the ventricle, often particularly and selectively of the septum, gives rise to a progressive distortion of the outflow tract of the left ventricle. Theoretically, the fault could lie in the relationship of the aortic root to the deep myocardial bands. Could the muscle bundles have lost their proper moorings and the septum act functionally, if not pathologically, as a tumor? This hamartoma simile was suggested by Teare. The greatly hypertrophied septum may encroach on the right ventricular outflow tract as well as on that of the left, so that an associated mild-to-moderate pulmonary subvalvular obstruction may be present. I have not seen evidence of an inflow obstruction as has been suggested as a possible concomitant by Goodwin and co-workers. In the fully developed condition, the aortic root appears to be unduly eccentric to the outlet of the left ventricle; perhaps the offset is accentuated by systole. The boundaries of the outlet of the left ventricle are, bilaterally, the shoulders of myocardium contiguous with the left and right aortic cusps and, posteriorly, the base of the anterior mitral leaflet which may be ‘‘pulled’’ obliquely over the top of the ventricular septum. The latter may give the impression of a congenitally deformed mitral leaflet. Also, the anterior mitral leaflet is vulnerable to damage at the time of surgical repair. In this distortion of the outflow tract, there is the anatomic basis of another superficial paradox, namely, lateral angiocardiograms may be thought to show an immediately subvalvular ‘‘stenosis’’ most clearly in diastole—when the anterior mitral leaflet is pushed against the massive septum by the blood entering the ventricle from the atrium. This deformity produces a characteristic triangle of opaque material below the aortic cusps. Again, in the lateral view, angiocardiograms might be misinterpreted to show a widely open tract during early systole, when the anterior mitral leaflet is pushed away from the septum toward the atrium. In the posterior-anterior view (or variations thereof), one can observe systolic narrowing, and often the apical portion of the left ventricle appears as a very small chamber with tremendously thickened walls. Indeed, in some cases the apical portion of the ventricle may be likened to an inadequately functioning self-obstructing diverticulum, and stroke volume is only from ‘‘the top of the ventricle.’’ Angiocardiograms also will demonstrate mitral regurgitation when it is present. It is clearly evident that not only does the hemodynamic state vary from time to time in the same individual and in different individuals but also the locus of obstruction may be at different ventricular levels in different individuals.

The patient with subaortic muscular stenosis, either child or adult, may present himself for the evaluation of a murmur or for the elucidation of syncope, anginal distress, or effort intolerance related to fatigue. The
symptoms may have had an accelerated aggravation during a period of a few months or years or have been unchanged, or even periodically absent, for several decades. An illustration of the latter, benign phenomenon is sometimes clearly portrayed by a relative, who happens to accompany a symptomatic patient scheduled for surgical treatment and who is found to have the disease also, but without significant symptoms.

On physical examination, auscultatory findings are usually the focus of interest. A precordial systolic murmur is heard best to the left of the midsternum, but is often heard also at the apex. It may or may not be heard well to the right of the upper part of the sternum, but it is rarely heard over the vessels in the neck. Usually it clearly starts after the first sound and stops before the second sound; thus it has the characteristics of an ejection murmur. However, the murmur may be holosystolic and louder at the apex than at the base, in which instance one suspects a concomitant mitral regurgitation. The murmur may have musical qualities and rarely, when severe obstruction is present, a systolic thrill may be elicited. A loud fourth heart sound is frequently present, and, indeed, sometimes one can feel (and record) a presystolic (atrial) ventricular-apical outthrust (fig. 1). In most cases, the diagnosis of aortic stenosis is suggested; however, it is interesting that in others the diagnosis of mitral regurgitation may have been entertained for years. After I had told him my diagnosis, one patient, a world traveler, mentioned that he had visited outstanding cardiologists throughout the world over the previous year and that now the tie was broken—it had been two for aortic stenosis and two for mitral insufficiency. (Undoubtedly, at the present time, it would be unanimous.)

When the diagnosis of "just aortic stenosis" is made in the case of a young person, the finding of a pulse having an initial forcible thrust, the presence of an aortic second sound, and the absence of an ejection click should give one reasons for excluding aortic valvular obstruction. There is frequently a pulsus bisferiens, and the initial rapid rise in the blood pressure differentiates the condition from valvular aortic stenosis. A pulsus bisferiens may also be present in mitral or aortic regurgitation, but the contour usually takes the form of two spikes rather than of a spike and hum ("percussion" and "flow" waves) of subvalvular functional stenosis. The differential diagnosis is not easy. (Studies of reference value are those of Hancock and of Braunwald and co-workers.)

A characteristic sign of subvalvular muscular stenosis is the increase in the obstruction (or gradient) in the cycle after a premature ventricular contraction, as has been pointed out by Brockenbrough, Braunwald, and Morrow. This may be clinically recognized, by noting that the pulse of the subsequent beat after the compensatory pause does not have the expected accentuation. It is presumed that increased filling of the ventricle and increased diastolic fiber length, resulting in increased vigor of contraction, also increases the outflow-throttling effect. A few persons with this disease have reported a decrease in well-being with digitalis therapy, suggesting that the inotropic effect of this drug, the associated decrease in chamber size, or both had caused an increased obstruction to flow. Braunwald and co-workers have published hemodynamic data which support this concept.

The reasons for the lack of consistency of data relating to the obstructive phenomena require further study. It was natural to consider such possibilities as a different end-systolic volume of the heart, a different contractility related to neurogenic or hormonal factors, and an aberrant type of ventricular excitation. Braunwald and Ebelt have demonstrated that different pressor amines vary in regard to their effect, either increasing, or virtually abolishing, the gradient, presumably depending on the presence or absence of an inotropic effect. The possibility of aberrant excitation of the ventricle has attractive features, but the lack of evidence of early
Figure 1

a. Records taken on a man aged 30 who had clinical and roentgenologic pictures of "functional" muscular stenosis and successful surgical treatment. Note fourth heart sound, unusually prominent "a" wave in apex tracing, and midsystolic ejection murmur.

b. Records from same patient illustrating increase in fourth heart sound and murmur, together with change in carotid pulse having a larger "tidal wave," after three breaths over broken amyl nitrite pearl. Ischemic type ST depression is noteworthy. Patient was not taking digitalis. Coronary arteries did not show any evidence of occlusive disease, when opacified at time of angiocardiography designed primarily to outline the left ventricle.
systolic obstruction and the variation in the obstructive site seem to deny its likelihood.

**Mitral Regurgitation Simulating Aortic Stenosis**

It was some years ago that I had my first encounter with a patient with a systolic thrill in the aortic area and a loud, harsh murmur extending from the apex to the aortic area, whose heart showed no evidence of aortic stenosis at necropsy. The posterior mitral leaflet in this heart was flail-like and unsupported, because of rupture of the major chordae tendineae. Gross mitral regurgitation was the obvious defect. Doctor Edwards and I\(^29\) adduced evidence from the endocardial lesions of the left atrium that the regurgitant jet was directed forward and would impinge on the atrial wall directly contiguous with the aortic root. Further support for this explanation has come from another case wherein the surgeon (Doctor Kirklin) felt a thrill at the aortic root during operation. The thrill completely disappeared after repair of the mitral leaflet. The knowledgeable amongst my readers might say 'forewarned is forearmed,' and, knowing of the possible confusion, argue that, if auscultation revealed the holosystolic nature of the murmur, its lack of propagation to the vessels in the neck, and the presence of an aortic second sound, if there were an enlarged left atrium, and if an

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**Figure 2**

Phonocardiograms of man of 52 years whose cardiac disability began with attack of pulmonary edema. Loud systolic murmur was heard at apex and "aortic area," and some trained observers believed aortic stenosis was present. Hemodynamic studies confirmed alternate opinion of mitral incompetence, and surgical exploration revealed rupture of chordae to posterior leaflet. Simultaneous phonocardiograms taken at apex and aortic area show rectangular holosystolic murmur in former and triangular aortic-stenosis-type murmur in latter.

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\(^{29}\) Edwards, Doctor
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electrocardiogram showed normal axis deviation with evidence of left ventricular hypertrophy with upright T waves in the left precordial leads, the physician would not fall into such a trap. Even when these important features are considered, I can assure you that the trap should not be disdained as so obvious as not to deserve respect. The systolic murmur at the “aortic area” may have a “triangular” shape and one could be led astray rather than enlightened by the usual type of phonocardiogram. The murmur may have two components, the apical one of holosystolic regurgitation and the aortic one related to the period of maximal regurgitation (fig. 2).

The hypothesis that the aortic bruit originates from resonance of the aortic wall secondary to the jet impact remains attractive to me, although the nature of the energy released is obscure. Competing ideas include vortex shedding and variations in values of instantaneous flow.

A “functional center” of the heart might be placed at the aortic valve-mitral valve region, including the membranous ventricular septum which lies usually in the angle between the bases of the posterior and right aortic cusps. This anatomic region (fig. 3) can be the center for “black magic” and for the focus of one’s attention in the case of a number of clinically diagnosable complexes; such lesions may test one’s skill to the utmost. In this area also, just under the membranous septum (or posterior and inferior to the usual type of ventricular septal defect), lies the atrioventricular conduction system, wherein a lesion a millimeter or so in diameter may be lethal. Parenthetically, in early childhood, it indeed may not always be easy to decide clinically between the diagnoses of mitral insufficiency and of ventricular septal defect.

Further illustrations of defects in this crucial center of the heart and the attendant clinical syndromes follow: (1) ventricular septal defect with aortic valvular incompetence, the latter being the prominent clinical fault, (2) ventricular septal defect and a ruptured right anterior aortic sinus aneurysm, simulating aortic incompetence, (3) ventricular septal defect of the type of an atrioventricular cushion defect (with the outstanding and presenting problem being mitral regurgitation), (4) posterior aortic sinus aneurysm with rupture to the right atrium, (5) an anteriorly located ventricular septal defect simulating pulmonary stenosis (the defect lies just below the pulmonary valve and anterior to the crista supraventricularis), and (6) ventricular septal defect, or acquired aortic stenosis, with complete heart block. (The occurrence of heart block with “corrected transposition” is also noteworthy.)

It may be recognized that not a few of these conditions may give rise to continuous bruits at the base of the heart. Other conditions that may give a continuous bruit are coronary arteriovenous (or arteriovenocameral) fistulae, constrictive lesions in the large pulmonary arteries, aortieopulmonary arterial

Figure 3
Coronal section through heart shows relationship of aortic root to mitral valve, membranous septum, and tricuspid valve. This preparation made by Dr. Jesse Edwards shows relationship superbly. (Reproduced with permission from Edwards, J. E., and Burchell, H. B.: The Pathological Anatomy of Deficiencies Between the Aortic Root and the Heart, Including Aortic Sinus Aneurysms. Thorax 12: 125-139, 1957.)
(supravalvular) communicating defects, valve incompetent foramen ovale in the presence of a tight mitral stenosis, certain aortic arch syndromes, and the well-known patent ductus arteriosus. One may ponder how the self-confident, remarkably able Doctor Gibson, who died just a half century ago, would have fared amongst these diagnostic traps.

In one instance of mitral stenosis and valve incompetent foramen ovale, the surgeon (Doctor Kirklin) could feel a nearly continuous thrill over the right atrium. He estimated the orifice of the foramen ovale to be 10 by 7 mm. and said the mitral orifice was typically severely stenotic (3 by 6 mm.). Preoperatively, calculation of the mitral flow was 4.3 liters per minute and of the foramen-ovale flow (from left to right), 5.7 liters per minute. It was of interest that this patient had occasional blackouts, perhaps related to transient increases in the diversionary left-to-right shunt. In another patient with a valve incompetent foramen ovale and with gross mitral incompetence related to ruptured chordae tendineae to the posterior leaflet, the diagnosis of ventricular septal defect was suspected and the "wedge" contour, or right atrial trace, showed no significant V wave; however, gross mitral insufficiency is not always associated with a high V wave when the atrial septum is intact.

Some discussion of the present status of the Graham Steell murmur is warranted. Pulmonary incompetence is uncommon and most blowing diastolic murmurs heard along the left side of the sternum are best ascribed to aortic incompetence. It is fair and valuable to quote from Steell’s original work:

"I am prepared for the objection that the murmur under consideration is only the murmur of a slight amount of aortic regurgitation. . . How difficult it is to distinguish between the murmurs of aortic and pulmonary regurgitation, by means of auscultation alone, will be admitted. . ." To make the diagnosis of pulmonary valvular incompetence, one should be certain that pulmonary hypertension has long been present and that the pulmonary artery is dilated. Even when these circumstances are present, one must not expect the pulmonary valvular incompetence to be readily revealed by the usual hemodynamic studies. Despite the relative loudness of some Graham Steell murmurs, the amount of regurgitation revealed by dye indicator methods in our laboratories has been small. An exception to this axiom of being chary of the diagnosis of pulmonary valvular regurgitation is the rare syndrome of congenital dysplasia of the pulmonary valves with pulmonary regurgitation.

Potential Roentgenologic Error

The roentgenogram of the chest is an integral part of the cardiac examination, and it sometimes may be practically pathognomonic, when the physical examination has not revealed the nature of the cardiac problem. A few examples are total anomalous pulmonary venous connection to the superior vena cava system (the "figure-of-eight" or "snowman" silhouette), Ebstein’s malformation, marked "flasklike" cardiac enlargement, small vascular shadow and oligemic lungs, and, in some instances, transposed great vessels with levoposition of the aorta and with "physiologic correction" to the ventricles.

One of the difficult diagnostic problems not infrequently encountered by the cardiologist concerns the patient without complaints or abnormal physical findings, who is referred for consultation because of an enlarged heart on the roentgenogram. In such instances one must consider the possibility that the shadow might be noncardiac, for instance, a cyst, a lipoma, or a hernia through the foramen of Morgagni. A pericardial effusion should also be in the list of suspected conditions, and this suspicion would lead to the necessary exclusion of covert myxedema—a condition all too easily overlooked unless one is alert to its protean nature and its mild manifestations in some patients. In problems such as these, special roentgenologic techniques, in particular angiocardiology and scanning after injection of a radioisotope, may be needed for elucidation.
Possible Electrocardiographic "Red Herrings"

An electrocardiogram should rarely lead one astray in a diagnostic problem. On occasion, one overhears electrocardiographic analysis being damned and belittled as an approach to cardiac diagnosis—perhaps because of its shortcomings in the diagnosis of ventricular hypertrophy or coronary sclerosis, a criticism which does not have a rational basis. In general, the electrocardiographic picture, whether it be within the range of normal or shows gross aberrations therefrom, contributes neatly to the final diagnosis—if it does not fit, one searches for the reason why, as, for instance, for complications of the primary condition. The value of the electrocardiogram in the recognition of electrolyte disturbances, particularly of potassium, is well established. It must be remembered that, as hypokalemia generally is associated with a trend toward alkalosis and hyperkalemia with acidosis, there may be a confusion electrocardiographic picture when the associations are reversed.

One example of an electrocardiographic-simulating condition concerns elderly persons—an abnormal electrocardiogram suggesting the scar of an infarct could be related to amyloid deposition or to the degenerative changes of hemochromatosis. These conditions also may give rise to intraventricular conduction defects, including gross left axis deviation, as may coronary disease. One may claim a renaissance for left axis deviation\(^{32}\) manifest either in the standard leads or in the vectorecardiogram.

Complicators

Having mentioned rarities and simulators (or imitators), what are some pitfalls in the diagnosis of complicating or associated defects?

The manifestations of aortic regurgitation may be so dominant that an associated coarctation is overlooked.

In primary coarctation of the aorta, with a clinically classic enough picture, subaortic fibrous stenosis may be overlooked. The additional aortic systolic murmur and greater-than-usual evidence of left ventricular hypertrophy may not "register" on the examiner. There is an increasing number of syndromes which include cardiac anomalies, one of the latest being mental deficiency, unusual facial features, and supravalvular aortic stenosis.\(^{33}\)

Associated small ventricular septal defects or associated subaortic stenosis may not be suspected in cases of primary pulmonary stenosis. An error may be made readily in some cases of infundibular (muscular) pulmonary stenosis associated with a small ventricular septal defect, when the latter gives rise to a prominent murmur. In such instances, a phonocardiogram may tip one off to the proper situation. Closure of the pulmonary valve may be delayed, although the sound may be so muted as not to be clinically recognized. In cases of infundibular stenosis in which the infundibular chamber is thin, the electrocardiogram may suggest the diagnosis, if the vectoral loop is anterior and to the right in the fashion of right ventricular hypertrophy, but not superior (resulting in an absent R wave in aVR).

In the case of a primarily ventricular septal defect one may overlook a corrected type of transposition of the great vessels, or a double outlet from the right ventricle, aorta, and pulmonary artery.

A mitral lesion associated with an incompetent foramen ovale may be overlooked and the lesion thought to be primarily an atrial septal defect.

Primary aortic stenosis is perhaps the most difficult lesion of all to assess in a person with angina, for how much coronary disease is present? In this instance, the assessments of the value and risk of coronary arteriography are current problems without well-established answers. It may be remembered that if one can diagnose a gross infarct by the electrocardiogram, severe coronary sclerosis may be assumed to be associated with the aortic stenosis.

The Emotionally Obscured Diagnosis

The fourth category of missed diagnoses...
has been called the emotionally obscured and there are three subtypes. In the first, the doctor either rides a hobby, is afraid to make a certain diagnosis because of a previous error, or has a mania for disagreeing with the general practitioner's diagnosis; in the second, anxiety or depressive symptoms in the patient thwart a methodical search for associated disease; and in the third, the patient may mislead the physician for reasons related to insurance or occupational gains.

Miscellaneous

Some other conditions are readily unrecognized. The examples are such that, from the armchair, we would think the diagnosis, theoretically, should not be difficult; but, at the bedside, for most of us, they often are. Diagnosing the following lesions may test one's perseverance: (1) atrial tumors, (2) atrial septal defect in the middle-aged and elderly, (3) acute and chronic myocarditis, (4) recurrent pulmonary emboli with pulmonary hypertension, and (5) pheochromocytoma.

I must emphasize that most patients with atrial tumors have clinical pictures sufficiently different from patients with other types of heart disease that the diagnosis should be at least tentatively made. One hesitates to mention that such has been accomplished in all recent cases, for one might fail to recognize the next one. The last case of right atrial tumor I saw presented the picture of right inflow obstruction, but it was unique in that there was polycythemia, suggesting the possibility of renal tumor (secreting erythropoietin?) growing upward from the kidney through the inferior vena cava to the right atrium. However, the tumor turned out to be "native" to the right atrium. There are many symptoms and signs of left atrial tumors that may be clues to the proper diagnosis—besides the usual absence of a history of rheumatic fever or of any murmur heard in early life—namely, rheumatic pains, low-grade fever, emboli, and variable murmurs. Interestingly, there may be an early diastolic snapping sound, supporting, for the unwary, the diagnosis of mitral stenosis. This sound is separated from the aortic second sound by a greater period of time than would be expected in mitral stenosis, if the patient's symptoms were significant. That is, with high atrial pressures and mitral stenosis, the opening snap is usually close (0.06 second) to the aortic second sound. It is important to emphasize that a left atrial tumor may simulate mitral regurgitation as well as mitral stenosis. In a few cases, observed at the Mayo Clinic, a combination of two findings on cardiac catheterization have been characteristic of left atrial tumor: namely, a high V (or regurgitant) pressure wave from wedged pulmonary catheter and a normal dye dilution curve. The latter would ordinarily exclude gross mitral incompetence. In the instance of the tumor, it reflects the small left atrial volume. It seems possible also that tumors may move retrogradely during systole, simulating a plunger forcing a pressure (V) wave back into the pulmonary veins. In the category of lesions simulating mitral valve disease one should be aware of the rare conditions of cor triatriatum and stenoses of the pulmonary veins.

The diagnosis of atrial septal defect in the elderly may be unexpectedly difficult, particularly in the presence of a loud apical systolic murmur and atrial fibrillation. In the latter condition, the apparent splitting of the second sound may not be "fixed" but may vary inversely with the duration of the preceding diastole, and it may be difficult to differentiate from the opening snap of mitral stenosis. It is easy to confuse such cases with disease of the mitral valve, with or without suspecting a concomitant valve incompetent foramen ovale.

Primary myocardial disease whether it be initially inflammatory or degenerative, as in amyloidosis and hemochromatosis, frequently presents difficulty in recognition. Differential diagnoses are usually pericardial disease, endocardial sclerosis, diffuse systemic sclerosis, and, in older persons, coronary disease. The marked cardiac enlargement with little or any response to treatment, indolent course,
persistent gallop rhythm, pulsus alternans, and arrhythmias are clues to the proper diagnosis of myocarditis. In young persons, an anomalous coronary artery from the pulmonary artery may result in left ventricular enlargement, and, in rare cases, mitral regurgitation may be an outstanding feature.

Comment

An attempt has been made to discuss some cardiac conditions that have been unrecognized on initial cardiac surveys or have given trouble in exact diagnosis. The conditions that are prone to escape recognition have been categorized primarily as the rare, the simulacarians of other conditions, the associated or complicating defects, and the emotionally obscured. In the challenge presented by such cases, lies much of the joy of a consulting cardiologic practice. It is acknowledged that a properly programmed computer could do an acceptable or even better job in the diagnosis of the known, but one hopes temporarily to surpass the machine in being able to probe into the nature of the puzzling cardiac syndromes which presently escape our understanding. In essence, the computer is very superior as an integrator but we may compete successfully as discriminators or as “thinking machines.”

References

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Mitral Stenosis

Mitral stenosis, though noted in 1669 by John Mayou (1640-79), was more fully described pathologically in 1715 by Raymond de Vieussens (1641-1716), Professor of Medicine at Montpellier, who also noticed the characters of the pulse; Giambattista (1682-1771) also described it. Its clinical recognition naturally waited on the discovery of auscultation, though in 1806 Corvisart (1755-1821) insisted on the diagnostic value of the thrill. The presystolic murmur was apparently heard by Bertin in 1824, but it was not until 1843 that its importance was fully recognized by Fauvel, who must be given due credit for this correlation.—SIR HUMPHRY DAVY ROLLESTON. The Harveian Oration. Great Britain, Cambridge University Press, 1928, p. 41.
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