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THE DIAGNOSIS OF CONGENITAL HEART DISEASE

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Congenital defects of the heart and great vessels are not uncommon, and about half of the abnormalities encountered might be considered to be clinically unimportant, were it not for their liability to involvement in an infective endocarditis. Space does not permit the delineation of every type of congenital abnormality, and this article deals only with those lesions which can be recognised with reasonable certainty when the usually available methods of investigation are employed.

Until comparatively recent times congenital heart disease was considered to be an adequate diagnosis, and to venture upon a more detailed anatomical diagnosis was exceptional. The advances of the last decade have altered this view, and there are now a number of anomalies which present a sufficiently characteristic clinical and radiological picture to permit accurate diagnosis. None the less, the conception still lingers that congenital heart disease implies a puny, cyanotic, and otherwise ailing child. This is far from the truth, for in practice the cyanotic cases are fewer than the acyanotic, but they attract more attention. The acyanotic case is frequently missed until some routine examination, and then because of their often marked and noisy physical signs, a quite unnecessary gravity may be attributed to their condition. The congenital heart case, particularly if acyanotic, presents few symptoms largely because he has adapted himself to his abnormality, and is quite different in this respect to the subject of acquired heart disease. It is a matter of first-rate importance to differentiate these cases so as to avoid undue restriction of activity and cardiac invalidism. Likewise, now that ligation of the ductus arteriosus has become an accepted, and often beneficial surgical procedure, the selection of an appropriate case demands an accurate diagnosis.

In the general investigation of a case of suspected congenital heart disease a careful history is essential. Special reference should be made to symptoms and the order of their appearance, and as to the date when a lesion was first suspected or diagnosed. A murmur persisting from early infancy is presumptive evidence of a congenital etiology. Enquiry should also be made with regard to rheumatic incidents.

Inspection will reveal the presence or absence of cyanosis and clubbing, and will disclose any associated somatic abnormalities. Cyanosis may not be obvious in early infancy despite the presence of a gross abnormality and shunt. Consequently it is not wise to make a firm diagnosis in the first two years of life unless it is well supported by laboratory data. Cyanosis may become established in later childhood and then justify the often applied term of the *maladie bleue*. In other cases cyanosis is episodal with pulmonary infection, or terminal, with or without congestive heart failure. The significance of these types of cyanosis is discussed below. Clubbing, which is rare before the age of two, is generally proportional to the cyanosis and may reach an extreme degree. A bulge of the left chest suggests cardiac enlargement from an early age. Marked pulsation in the neck is visible in the patent ductus arteriosus when the arteriovenous shunt is large, and in coarctation of the aorta.

Palpation enables an estimation of the size of the heart. A thrill should be carefully sought, and its site of maximum intensity noted. The presence of a thrill is not necessarily an indication of the severity of a lesion. The smaller the lesion or aperture concerned in its production, the more intense the thrill. Thrills are generally systolic in time, and a thrill over the base of the heart suggests stenotic lesions in the aorta or pulmonary artery. If in the second left space a patent ductus or pulmonary stenosis may be present. In the former the thrill may be continuous. In the second right space a thrill is due to aortic or subaortic stenosis. In the mesocardial region, and between the sternal border and apex in the fourth left space the systolic thrill of a localised interventricular septal defect may be felt. A palpable diastolic shock in the pulmonary area suggests pulmonary artery dilatation. A thrill may be felt, together with pulsation, in the vessels of the collateral circulation in coarctation of the aorta. The pulse is small in aortic stenosis. The femoral artery should be routinely palpated if coarctation is not to be missed.

Percussion adds very little to the facts gained by palpation. A dilated pulmonary artery may be accompanied by "ribbon dullness" in the second and third left spaces adjacent to the sternum, and may be anticipated in a patent ductus or interauricular septal defect.

Auscultation is concerned with the presence or absence of murmurs. A gross cardiac abnormality may be unaccompanied by any murmur, or thrill, if there is a large septal defect, or if there

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is atresia of a valvular orifice. Commonly murmurs are systolic in time, and their site of maximum intensity and propagation must be most carefully noted. Systolic murmurs in situations other than those of acquired heart disease are very suggestive of a congenital lesion. Systolic murmurs at the base of the heart may be caused by aortic or pulmonary valvular lesions and are conducted along the subclavian artery towards the right axilla in aortic stenosis, or to the left clavicle in pulmonary stenosis. A continuous or "machinery" murmur in the first and second left spaces is the hall mark of a patent ductus arteriosus. Loud systolic murmurs in the third and fourth left spaces, heard with diminishing intensity as the periphery of the heart is approached, are found in isolated interventricular septal defects. Systolic murmurs may be heard over the vessels of the neck in aortic stenosis, or over the vessels of the collateral circulation in coarctation. The aortic or pulmonary second sound may be reduced or absent in stenotic lesions. On the other hand, in cases with pulmonary artery dilatation the second sound is accentuated, and accompanied by visible pulsation in the upper left intercostal spaces.

The blood pressure should always be taken. This may be difficult in small children, but a folded brassard will remove some of the difficulty and leave room for the stethoscope. The blood pressure is low in aortic and subaortic stenosis. It tends to be low in interauricular septal defects. A low diastolic pressure is present in the patent ductus and it becomes lower still with exercise if the shunt is at all large. Hypertension suggests a coarctation of the aorta, and is pathognomonic if accompanied by a low pressure in the legs.

Finally, the special methods of investigation, the radiological picture and the electrocardiogram may be employed. Radiology may be very helpful, but gross abnormalities of the heart can exist without any special alteration in the X-Ray picture. In some lesions there may be a rather characteristic cardiac silhouette. Radiology gives information as to the size and shape of the heart as a whole, the relative sizes of its chambers, and allows visualisation of the pulmonary artery and its branches. Rounding of the left ventricle is found in aortic stenosis and coarctation. A "coeur en sabot" with its upturned apex due to right ventricular hypertrophy is found in the tetralogy of Fallot. A dilated pulmonary artery is found in interauricular septal defects, patent ductus arteriosus, pulmonary stenosis with closed septum and in rarer conditions. Dilatation extends to the branches of the pulmonary artery in interauricular septal defects, and is especially visible on the right side where the vessel is not covered by the heart. On the screen a hilar dance may be witnessed in

both interauricular septal defects and in the patent ductus. In pulmonary stenosis complicated by an interventricular septal defect (Fallot) and overriding aorta, there is a concavity at the site of the pulmonary arc. The aortic knuckle is small in conditions tending to depletion of the systemic circulation such as interauricular septal defects, in aortic hypoplasia, and coarctation. Notching of the undersurfaces of the ribs is pathognomonic of coarctation.

With the exception of mirror image dextrocardia, the electrocardiogram does not provide any pathognomonic picture. The much quoted tall ventricular complexes are uncommon, but diphasic ventricular complexes in two or more leads are suggestive. The electrocardiogram does furnish important confirmatory evidence by indicating left or right ventricular strain by left or right axis deviation. A large P wave may be present in conditions involving auricular hypertrophy, and conduction defects are readily recognisable.

A blood count will help in the recognition of the early stages of polycythaemia, and in established cyanosis may help in prognosis. A blood culture is essential in the diagnosis of infective endocarditis.

Identification of Individual Abnormalities

Certain abnormalities are of small clinical importance. Mirror image dextrocardia is readily diagnosed by its apex beat in the fifth right space and inversion of the P, QRS, and T waves in the 1st lead of the electrocardiogram. A right aortic arch, a rare cause of dysphagia, is a purely radiological diagnosis. Ectopia cordis requires no further description.

Idiopathic congenital hypertrophy of the heart may result from so many conditions that to speak of idiopathic is a misnomer. Many cases are examples of von Gierke's disease; others result from various little understood forms of myocarditis, in some cases due to an abnormal origin of a coronary artery from the pulmonary artery; yet others may be accompanied by hyperplasia of the genital organs in the female, and occur in the children of diabetic mothers. The heart is grossly enlarged. There are commonly respiratory difficulties, and the prognosis is poor. More often than not exact diagnosis is a post-mortem event.

Aortic and Subaortic Stenosis

Most cases of aortic stenosis arise from a foetal endocarditis, and the distinction from a subaortic stenosis, the result of persistence of the bulbus cordis in the left ventricle, may be difficult. The

common feature is a loud systolic murmur in the second right space, often with accompanying systolic thrill. The murmur is conducted up into the carotids, and along the subclavian vessels to the axilla. The pulse is small and the blood pressure low. The aortic second sound is normally unchanged in a subaortic stenosis, but may be diminished, or followed by a diastolic bruit in aortic stenosis. The apex beat is generally forcible but clinical enlargement of the heart is rarely apparent in early years. The X-ray picture shows occasionally some dilatation of the ascending aorta and slight hypertrophy of the left ventricle. The electrocardiogram shows a normal or left axis. Clinically the cases may be quite symptomless and latent. Infective endocarditis may involve either lesion, pulmonary tuberculosis is not uncommon, and valvular aortic stenosis may be a cause of fatal syncope and sudden death.

Coarctation of the Aorta

Coarctation refers to a narrowing of, or obliteration of the aorta at the site of insertion of the ductus arteriosus. Hypertension in a young subject should always suggest coarctation until some other explanation has been found. Apart from hypertension the other important physical signs are diminished or absent pulsation in the femoral arteries, with corresponding low blood pressure in the legs, and a collateral circulation evidenced by visible and palpable vessels around the scapulae, in the root of the neck, intercostals, and not infrequently the abdominal wall. The auscultatory signs are a systolic bruit, often quite inconspicuous, and at times best heard posteriorly along the vertebral column, and characteristically over the enlarged vessels of the collateral circulation.

Thus an abnormally sited systolic bruit should lead to a search for other signs. The heart is not enlarged until relatively late. After the age of 12 the radiogram may show notching of the under-surfaces of the third to the tenth ribs, especially on the right side, due to pressure atrophy induced by the hypertrophied and tortuous intercostals of the collateral circulation. Notching of the ribs is pathognomonic. The electrocardiogram shows a normal or left axis. Death may result from infective endocarditis of the commonly associated bicuspid aortic valve, or of the site of coarctation. It may also result from rupture of an associated congenital cerebral aneurysm. The condition may remain latent and symptomless until middle life.

Patent Ductus Arteriosus

The pathognomonic sign is a continuous "machinery" murmur in the first and second left spaces close to the sternum. The murmur commences with the first sound and reaching its maximum intensity with the second sound, continues at this pitch through diastole. The pulmonary second sound is accentuated because of pulmonary artery dilatation. Unfortunately the machinery bruit is not always present, and may be represented by a systolic bruit or by both systolic and diastolic murmurs. It is rare before the age of 2, and clinical experience suggests that it may not be present in its typical form until the third year. The absence of a machinery bruit raises diagnostic difficulties, for now that there is a distinct trend towards surgical treatment, there must be certainty that a patent ductus is actually present. Further the technical difficulties of operation increase as age advances, for the ductus tends to become shortened and less easily accessible. The operation

Coarctation of Aorta. Small aortic knuckle, slight left ventricular enlargement, and notching of the under-surfaces of the ribs.

Patent Ductus Arteriosus. Prominence of the pulmonary arc.

is best done before the age of 15, unless infective endocarditis is present when it may be attempted at any age. If the shunt is at all large there is vigorous pulsation in the neck. The diastolic pressure is low, and falls lower still with exercise. Enlargement of the heart indicates a large shunt, such cases often being small and under-developed. The electrocardiogram nearly always shows a normal axis in the young, and a left axis may be present in older cases. The finding of an alteration in electrical axis in a young subject must lead to a careful scrutiny of all the evidence, as it may indicate the presence of some associated cardiac lesion, and is a contraindication to operation. The X-ray picture shows prominence of the pulmonary arc, and on the screen a hilar dance. Symptoms may be entirely absent and depend upon the size of the shunt from aorta to pulmonary artery. Death usually results from infective endocarditis, to which the ductus is peculiarly vulnerable, or congestive heart failure.

Isolated Interventricular Septal Defect (Maladie de Roger)

The maladie de Roger is characterised by its marked physical signs and absence of symptoms. There is a loud harsh systolic murmur in the fourth left space, heard with diminishing intensity as the periphery of the heart is approached. It is often accompanied by a systolic thrill. The site of the defect, just anterior to the membranous septum, brings it into proximity with the bundle of His, so that congenital heart block or other less pronounced disturbance of conduction may very

a. Congenital complete heart block in child aged 3.

b. A typical bundle branch block in child with inter-ventricular septal defect.

occasionally be present. The rate of a congenital heart block is usually more rapid than that of an acquired block, and rates of 50-60 are not unusual. The electrocardiogram is physiological, apart from those rarer cases where a block or other conduction disturbance is present. There is no abnormality in the X-Ray picture other than fullness of the pulmonary arc giving the cardiac silhouette a straight left border.

Interauricular Septal Defects

An interauricular septal defect of 1 cm. diameter is usually considered to be the starting place of pathology, and must not be confused with the common slit-like patency of the foramen ovale which is accepted as a normal anatomical variant. Such a lesion may remain entirely latent and symptomless until the third decade, or later. There is a constant gravitational shunt through the defect from left to right and gradually enlargement of the right auricle and then right ventricle becomes apparent, so that the patient complains

dilated and this is shown clinically by an accentuated pulmonary second sound, and occasionally a diastolic murmur of pulmonary incompetence. A systolic murmur may also be present over the dilated pulmonary artery. Late in the course of the disease auricular fibrillation may be present, and this defect is the only congenital lesion where it is likely to appear with any frequency. When mitral stenosis is associated there is very little alteration in the physical signs, but the course may be more severe and rapid. Such a large number of cases have mitral stenosis at autopsy, without recognised signs during life, that it is unwise to assume that mitral stenosis is not there. The radiological picture is characteristic and shows a small aorta with a very large, often aneurysmal pulmonary arc, and a large right branch of the pulmonary artery. The heart is enlarged. The right auricle is conspicuous on the right border, and even if mitral stenosis is present there is no enlargement of the left auricle and no displacement of the barium filled oesophagus. The electrocardiogram shows a right axis in the bulk of cases

Interauricular septal defect. Note general cardiac enlargement with prominent pulmonary arc and large right pulmonary artery, and small aorta.

of increasing breathlessness, and possibly some cyanosis on exertion. The breathlessness may be so habitual that it is not always complained of. There are no characteristic murmurs because there are no vigorous shunts or obstructive lesions likely to cause them. There may be the murmur of a mitral stenosis which is very commonly associated, even in the absence of any rheumatic history. The heart is large, and this enlargement alone, in the absence of any recognisable factor such as hypertension or aortic incompetence should at once raise suspicion and is a strong diagnostic point. The pulmonary artery is always

and the ventricular complexes may be widened with the occurrence of atypical forms of bundle branch block. Death generally results from congestive heart failure after a long period of increasing dyspnoea and cyanosis. The lesion is almost exempt from infective endocarditis, and the occurrence of the latter is a pathological curiosity.

Cyanotic Group

In any series of congenital heart cases the cyanotic case figures prominently and gives a false idea of its frequency, for the reason that cyanosis

commands more attention and interest, perhaps because there is something so obviously wrong. Cyanosis depends upon a variety of factors and, these are, in congenital heart disease, a venous-arterial shunt, stasis in dilated capillaries at the periphery, and in a few cases where there is engorgement of the pulmonary circuit, as in the interauricular septal defect, deficient oxygenation in the lungs. A venous-arterial shunt can arise not only through septal defects, but also in those cases where there is dextroposition of the aorta or a transposition of the great vessels, so that the aorta may either over-ride the septal defect, or else arise wholly from the right ventricle.

From a clinical point of view cyanosis may be apparent at birth, and if severe denotes some grave anomaly such as transposition of the great vessels without an interventricular septal defect, a condition only compatible with a few days of life, and possibly recognisable by absence of murmurs and other signs. In other cases cyanosis may only be slight at birth but subsequently progressive in intensity. Such cases are numerous, and the largest proportion of them fall into the category of the tetralogy of Fallot. Cyanosis in an infant, provoked by crying or suckling is often due to a patent ductus arteriosus. Cyanosis appearing in episodes during pulmonary infection, or as a terminal event, or in congestive heart failure, constitutes the cyanose tardive, and is due to reversal of an arteriovenous shunt in habitually acyanotic lesions such as the *maladie de Roger*. Lastly cyanosis appearing for the first time in early middle life suggests an inter-auricular septal defect. Corresponding to the degree of cyanosis all grades of clubbing of the fingers may be present. Clubbing is rare in infancy.

Polycythaemia is present and counts of 7-9 millions are frequent. A high red cell count carries a poor prognosis.

The identification of individual lesions in a cyanotic case taxes all the resources of a clinician. It is unwise to venture on accurate diagnosis in early childhood when signs are minimal, and a guarded prognosis should accordingly be given.

The Tetralogy of Fallot

This combination of defects, the most frequently encountered in the cyanotic group, consists of an interventricular septal defect, dextroposition (over-riding) of the aorta, pulmonary stenosis (or atresia), and right ventricular hypertrophy. About 85 per cent of cyanotic cases fall into this category, and the clinician confronted with a cyanotic congenital heart case in late childhood has considerable justification in diagnosing this combination. Marked cyanosis and clubbing are present. The heart is usually not enlarged. There is a basal systolic murmur, best heard in the second left space, and often conducted up into the vessels of the neck, and towards the left clavicle. There may be an accompanying systolic thrill in the same situation. The pulmonary second sound may be diminished or absent. The X-ray picture tends to show a *coeur en sabot* cardiac silhouette, and there is a concavity at the site of the pulmonary arc. The aorta is more to the right than normally. The electrocardiogram invariably shows a marked degree of right axis deviation. These cases may live a surprising length of time despite a very great handicap. The largest proportion die in the first two decades, but the oldest recorded case lived to 49 years.

Eisenmenger Complex

This is a combination of defects similar to the tetralogy with the exception that the pulmonary artery is dilated. Cyanosis and clubbing are

Tetralogy of Fallot. Concavity at site of pulmonary arc, and *coeur en sabot*.

FIG. 1.—Patent ductus arteriosus in female child, aged 6.

FIG. 2.—Coarctation of aorta.
Pregnant woman, aged 27.
Marked notching of ribs.

FIG. 3.—Tetralogy of Fallot with right aortic arch. Child, aged 8.

FIG. 4.—Tetralogy of Fallot. Female, aged 12.

FIG. 5.—Interauricular septal defect. Female, aged 24.

FIG. 6.—Pulmonary stenosis with closed ventricular septum.

present. There are no characteristic auscultatory signs. The X-ray picture shows a dilated pulmonary artery in contradistinction to the concavity of the Fallot. Right axis is present in the electrocardiogram.

Pulmonary Stenosis

Pulmonary stenosis may be either valvular or infundibular, and the greater number of cases are complicated by septal defects, and form part of the tetralogy of Fallot. There are however a relatively few cases which arise either as a foetal endocarditis, or as a developmental abnormality. The foramen ovale is usually open. The striking physical sign is a loud, rasping, systolic murmur

Dextrocardia. Inversion of P and T waves in leads 1 and 2.

in the second left space with corresponding systolic thrill. The bruit is conducted towards the left clavicle. The X-ray picture shows dilatation of the pulmonary artery or conus, and the electrocardiogram a right axis. Cyanosis is absent or late in appearance, and rarely reaches a high degree.

Tricuspid Atresia

This lesion may be a cause of very severe cyanosis. There are usually no murmurs, and the X-ray picture shows a small pulmonary artery and right ventricle. It has the distinction of being the only congenital cyanotic lesion with a left axis in the electrocardiogram.

STERILITY IN PRACTICE

An Editorial Commentary

The advent of clinics and specialisation has done much to rob the general practitioner of his interest in this most fascinating of subjects. It is suggested that he should still occupy an important position in the team necessary for the proper investigation and treatment of infertility. There is none better fitted than himself for tackling a condition in which the knowledge of the home life and of the habits of the couple concerned, and frequently those of the previous generation, are of paramount importance in the disentanglement of the many and varied causes of the infertile marriage. He is all the more necessary since many women still feel that there is some sort of stigma attached to this state of barrenness. This stigma may in part be associated with the teachings of the Bible "God has taken away my reproach," cried Rachel after pregnancy had succeeded her infertility. The modern Rachel prefers to approach the friendly atmosphere of the family physician rather than that of the somewhat austere surroundings of a clinic.

However, those associated with a sterility clinic cannot fail but to notice that many practitioners are still confused regarding their method of approach to the subject. . . . How often has the patient merely been told to "Take a good holiday . . . or has been handed some form of glandular tablet without any previous examination or investigation? Much worse still, she may have been subjected to curettage and treatment in the absence of any proper tests regarding her husband's potency. This latter procedure should be condemned as a grave professional misdemeanour.

This short article will endeavour to help the practitioner as to how far he should go with his own investigations before calling in other help. The scope of these few words will not allow of more than a few pointers for the doctor and of a few "Do's and Don'ts": Discussion of the multitudinous fertility factors and their respective treatments would call for many hundreds of pages. The result of condensation may lay the author open to the criticism of dogmatism, to which this is his only defence. That the matter under discussion is an important one and that it has been rendered even more important owing to the ravages of war is something about which we shall all agree. In America, for example, it is stated that there are about five million infertile marriages (Popenhoe, 1943). Other statisticians claim that sterile unions form as many as 15 per cent of the total marriages. Other experts such as Titus and Hamblen (1935) state that from 25 to 50 per cent