CYANOTIC CONGENITAL HEART DISEASE

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The last 15 years have witnessed a very remarkable change in the attitude of the clinician towards cyanotic congenital heart disease. It is within the memory of many that it was the custom to make a simple diagnosis of congenital heart abnormality with cyanosis, and express the opinion that little or nothing could be done about it. Then there appeared the pioneer work of Abbott and others which produced not only a classification of the various abnormalities, but furnished a mass of clinical and postmortem observations from which

it was possible to build up a clinical picture and so arrive at a reasonably correct anatomical diagnosis. At the same time knowledge of the natural history of congenital heart disease has also increased, and some idea of its prognosis has been ascertained. Lastly, the conception of Taussig that an inadequate pulmonary blood supply was the critical abnormality in many cyanotic cases, and the development of an anastomotic operation between the systemic and pulmonary circulations so as to furnish an artificial ductus arteriosus, by Blalock

FIG. 1.—Tetralogy of Fallot. Female aged 3 years. Right ventricle opened. An arrow marks the subvalvular stenosis of the pulmonary artery. Ventricular septal defect with probe passing into it behind the crista supraventricularis.

FIG. 2.—Tetralogy of Fallot. Female aged 3 years. Left ventricle opened. A large aorta lies astride a high ventricular septal defect.

and Taussig (1945), has entirely revolutionized our outlook on the treatment of the cyanotic case. The assessment by Campbell (1948) that only one in two cyanotic children is likely to reach the age of 7 years, only one in four to reach the age of 14, and less than one in ten to reach the age of 21, certainly justifies an operation even if its risks are considerable. It is all we have to offer to a patient who may exist rather than live. Accordingly, each case that presents itself should be fully investigated with a view to an accurate anatomical diagnosis, so that its suitability for operation may be considered. The investigation demands well-planned team work between the cardiologist, surgeon, radiologist and clinical pathologist. Important contributions are made by all of these towards the elucidation of the case both in regard to diagnosis and treatment. Highly specialized technical work is involved in both cardiac catheterization and angiocardiology. The pioneer work of Cournand (1949) in

cardiac catheterization and its development by others, together with numerous studies in angiocardiology have made it possible to observe physiological alterations in the living patient, and so furnish important diagnostic information.

There was a time when cyanotic congenital heart disease was considered to be uncommon, and it was certainly rare in individual experience. The publicity in connection with operation for blue babies has literally brought to light many cases that have remained sheltered in their homes, incapable of any sustained activity and so incapacitated that some have not even walked. Many, but not all, of the children are underdeveloped. Some are retarded mentally because they have not had the opportunities for education afforded to their normal fellows. All are the subjects of great affection from their parents, and it is peculiar that both parents usually accompany the child to the out-patient department.

FIG. 3.—Pulmonary atresia. Female aged $4\frac{1}{2}$ years. (a) Posteroanterior. (b) Left anterior oblique.

The Approach to the Cyanotic Case

The obvious presenting sign is cyanosis which may be of all degrees. It is most necessary to enquire carefully as to the first appearance of cyanosis. It may have existed since birth or have developed subsequently. Extreme cyanosis at birth usually denotes a severe abnormality, often incompatible with prolonged existence. It is usually in the nature of a transposition of the vessels with a closed septum, a severe pulmonary atresia without a patent ductus arteriosus, or a pulmonary atresia with extreme dextroposition of the aorta. Moderate and obvious cyanosis from birth is the most usual finding, the cyanosis becoming more marked as age advances. Only a small percentage of cases develop cyanosis at the transition between the vegetative stage of infancy and the more active stage that follows it. Cyanosis appearing at puberty or in later childhood suggests the Eisenmenger anomaly, or pulmonary stenosis with a patent foramen ovale. Cyanosis appearing for the first time in the late teens or early 20's brings to mind the auricular septal defect and is rarely very marked. Isolated pulmonary stenosis with a closed septum may also show cyanosis at this time from changes that have occurred in the smaller pulmonary vessels. Lastly, the duration

of cyanosis may have some bearing on the diagnosis. If it has existed in any marked degree since birth, growth and nutrition are likely to be below average. In older children the presence or absence of clubbing furnishes an important clue as to its duration. Recent cyanosis with no clubbing may be due to the simple reversal of a shunt from extracardiac causes such as pulmonary infection, and is unaccompanied by polycythaemia. Cyanosis may develop as a permanency in the late stages of a patent ductus arteriosus, auricular septal defect, or ventricular septal defect in response to pulmonary vascular changes and pulmonary hypertension.

Anatomical Factors in Cyanosis

Most cases of cyanotic congenital heart disease present two basic developmental disturbances to cause cyanosis. Firstly, there is stenosis of some part of the pulmonary tract, in either conus, valve or artery, so that an insufficient volume of blood reaches the lungs for oxygenation. It is this factor which is capable of surgical modification and it is towards its detection, and the estimation of its severity, that our efforts must be directed. Secondly, there may be a shunt of mixed blood into the aorta which commonly overrides a high

FIG. 4.—Tetralogy of Fallot. Right aortic arch. Female aged 21 years recently delivered of a normal child.

ventricular septal defect, or perhaps the ventricular septum is absent in the functional sense. This factor in cyanosis cannot be remedied. It is a variable factor partially depending upon the extent to which the dextroposed aorta overrides the septum and right ventricular cavity, and the severity of the pulmonary stenosis. Severe pulmonary stenosis prevents the rapid passage of blood into the pulmonary artery during systole so that a large volume of venous blood is diverted into the aorta. Slighter degrees of pulmonary stenosis allow the more easy passage of blood into the pulmonary artery and a corresponding diminution in the volume shunted into the aorta. Exercise, which increases the cardiac output, at the same time must increase this shunt for the narrowed pulmonary artery cannot utilize this augmented blood flow. Thus cyanosis may be minimal at rest and increases with exertion. In some cases the only indication that exists of such a shunt is a reduction in the oxygen saturation of arterial blood, insufficient to cause clinical cyanosis. There are other shunts and other factors which may contribute to, or emphasize already existing cyanosis, but they will not be considered in any detail here. Mention should be made that in transposition of the vessels the pulmonary blood supply is adequate and the main difficulty is in securing a return of the oxygenated blood to the systemic circulation. In certain abnormalities and

in polycythaemia there may be important changes in the pulmonary capillaries and endothelium, which prevent the blood from being properly oxygenated.

Physiological Factors

A detailed discussion of the physiological mechanisms involved in cyanosis will not be attempted here. Only its general effects and complications will be mentioned. Cyanosis may be due to anoxaemia from any cause and implies the presence of at least 5 gm. reduced haemoglobin in the peripheral blood. Persistent cyanosis over a period of time results in polycythaemia and clubbing, the former being an effort by the body to increase its oxygen carrying capacity and adapt itself to the anoxaemia. Polycythaemia has important implications, and although accompanied by clubbing is unaccompanied by enlargement of the spleen. Red cell counts of seven or eight million are common and the level of haemoglobin is correspondingly raised above 100 per cent. A rising red cell count is of bad prognostic significance. A falling count, due to anaemia, is also a bad sign because it means a lower oxygen capacity of the blood, although the patient may look much better and less cyanosed as a result of this. Polycythaemia has the attendant dangers of thrombosis, cerebral thrombosis with hemiplegia occurring at times. Severe polycythaemia may also induce changes in the pulmonary capillaries and endothelium, or cause areas of infarction in the lung. All these conditions interfere with adequate oxygenation. There is also the risk of cerebral

FIG. 5.—Tetralogy of Fallot. Female aged 6 years. Marked right axis with 2 to 1 block.

abscess in the cyanotic case where a large volume of blood does not reach the lung and so escapes the filter of the pulmonary endothelium. The position of the aorta over a septal defect is a favourable circumstance for this to occur.

Laboratory Examinations

The recent work in cardiac catheterization and blood analysis has materially increased our knowledge and has greatly assisted diagnosis. Estimation of the arterial oxygen saturation of the blood is an important and informative examination. It should be done with the subject at rest and after exercise. In severe pulmonary stenosis or atresia the oxygen saturation may be as low as 20 per cent. In cases with a higher oxygen saturation there may be a marked and rapid fall on slight exertion, and if this occurs it means a severe stenosis with poor pulmonary circulation. In less severe cases there may be a level of 70 per cent. oxygen saturation which hardly falls on exercise, confirming that the pulmonary stenosis is not of marked degree. Lastly, an estimation of arterial oxygen saturation reveals the presence of reduced

FIG. 7.—The Eisenmenger complex. Pulmonary artery enlarged and the aorta lying astride a high ventricular septal defect.

FIG. 6.—Eisenmenger complex. Female aged 24 years. Infective arteritis of pulmonary artery.

haemoglobin in the circulating blood in an amount insufficient to produce clinical cyanosis in some abnormalities where a venous arterial shunt is of importance.

The measurement of pressures within the cardiac cavities and pulmonary artery, and the movements of the catheter itself within the heart have also furnished much valuable information. These techniques are discussed in another paper and will not be entered into here. Although these methods have the greatest possible value, the impression should not be formed that it is impossible to make a diagnosis without them. The pioneer work of Blalock and Taussig was largely done without these highly specialized aids to diagnosis, their reliance being placed on clinical observation and radiological examination.

Types of Congenital Heart Disease with Cyanosis

It is impossible to describe every type of congenital heart disease with cyanosis within the compass of this paper. Only certain anomalies will be described and these are mainly those that may derive benefit from surgery. They are not arranged in order of severity of cyanosis, as every

FIG. 8.—The Eisenmenger complex. Male aged 12 years. Incipient cyanosis. Marked right axis in electrocardiogram.

FIG. 9—Pulmonary stenosis with patent foramen ovale. Male aged 15 years.

case within a group may vary, but are arranged in the order of their frequency from the author's experience.

(1) Tetralogy of Fallot (pulmonary stenosis, ventricular septal defect, overriding aorta and right ventricular hypertrophy).

(2) Eisenmenger complex (ventricular septal defect with overriding aorta and normal or dilated pulmonary artery).

(3) Isolated pulmonary stenosis (pulmonary stenosis with or without a patent foramen ovale).

(4) Tricuspid atresia.

(5) Transposition of the great vessels.

(6) Persistent truncus arteriosus.

(7) Single ventricle with diminutive outlet chamber.

The Tetralogy of Fallot

The tetralogy of Fallot is by far the commonest type of congenital heart disease and accounts for about 75 per cent. of cyanotic cases surviving infancy. The anatomical findings, which are variable from case to case within considerable but defined limits, are stenosis or atresia of the pulmonary

artery, a high ventricular septal defect, dextro-position of the aorta so that it overrides the septal defect, and hypertrophy of the right ventricle, a logical sequence of these structural abnormalities (Figs. 1 and 2). The pulmonary stenosis may be at almost any point in the pulmonary tract and may be valvular, infundibular or both, the artery distal to the stenosis being generally hypoplastic. In severe atresias the trunk of the pulmonary artery may be reduced to an impermeable fibrous cord ending blindly in the ventricle. Sometimes there may be just sufficient blood flow through a stenotic vessel to maintain life, and the ductus and foramen ovale may be closed. Where there is anatomical or functional atresia a pulmonary circulation is maintained through a patent ductus arteriosus and also by means of a collateral circulation developed through the bronchial and other arteries. These two different types of pulmonary circulation have led some to classify pulmonary atresia as a distinct syndrome from a functional point of view. At times the dextroposition of the aorta is so marked that the aorta may arise wholly from the right ventricle. Occasionally, transposition of the vessels also occurs and the stenosed pulmonary artery arises from the left ventricle. In at least a fifth of the cases there is a right aortic arch with the aorta crossing the right bronchus.

The physiological results of this combination of structural abnormalities is that, firstly, a large volume of blood from both ventricles passes directly into the aorta, the proportions of venous and arterial blood being to some extent determined by the position of the dextroposed aorta vis-a-vis the higher septal defect and right ventricular cavity; secondly, an inadequate volume of blood reaches the lungs for oxygenation through the stenosed pulmonary tract, or through a patent ductus arteriosus. The severity of pulmonary stenosis naturally influences the volume of blood that can reach the lungs for oxygenation and consequently there are variations from case to case in regard to the onset and degree of cyanosis. Slight stenosis of the pulmonary artery may well be compatible with life with relatively small handicap, and cyanosis appearing first in response to exertion, often is not marked until late childhood. In severe pulmonary stenosis amounting to functional atresia the main blood supply to the lungs may be through a patent ductus arteriosus and correspondingly early and marked cyanosis is likely. Clearly the severity of the pulmonary stenosis and its consequent effects upon the pulmonary circulation determines the survival of the patient and it becomes necessary to distinguish, as far as possible, between those cases with pulmonary atresia, actual or functional, and those with pulmonary stenosis of lesser degree. The patient with pulmonary atresia is at the mercy of his patent ductus arteriosus. Should this close, and it may close spontaneously early or late, there is a marked deterioration in his condition and death may result.

Clinical Picture

Marked cyanosis from birth and clubbing of the fingers in a child or young adult at once suggests the tetralogy as a probable diagnosis on the basis of survival alone. Cyanosis may not necessarily be clinically apparent at birth and may only appear when the child becomes active, or when the ductus closes. Once it appears it becomes permanent and progressive and it is accompanied by dyspnoea on the least exertion so that activity is strictly limited. Clubbing is proportional to the degree of cyanosis. It is a curious fact that most cyanotic children when suffering from dyspnoea assume a squatting position which enables them to breathe more easily. The child sits on his heels with the knees drawn up to his chest and may even sleep in this attitude. Spontaneous attacks of paroxysmal dyspnoea with intense cyanosis and difficulty in expiration sometimes occur in young children and are a potential cause of sudden death.

The heart is not enlarged. Some deformity of the left chest from right ventricular hypertrophy occurring at an early age may be present and there

is frequently a basal systolic thrill and accompanying systolic murmur. The murmur is generally not heard in the neck but is heard along the left sternal margin. The systolic murmur is caused by the pulmonary stenosis and is often absent in pulmonary atresia. The pulmonary second sound is usually unaltered.

Radiology

The heart is not enlarged and there is a concavity at the site of the pulmonary arc (Figs. 3 and 4). In the left anterior oblique view there is enlargement of the right ventricle which projects

FIG. 10.—Pulmonary atresia with closed ventricular septum and patent foramen ovale. Collateral bronchial artery circulation. Marked right ventricular hypertrophy. Tall P waves.

FIG. 11.—Tricuspid atresia. Female child aged 7 months markedly cyanotic with a normal axis in the electrocardiogram. (a) The aplastic right ventricle has been opened and an arrow points to a ventricular septal defect. (b) Interior of the right auricle. Depression at site of the tricuspid valve. Foramen ovale open.

FIG. 12.—Tricuspid and pulmonary atresia. Female aged 8 months. Marked cyanosis and left axis in the electrocardiogram. (a) External view of heart. The atrial pulmonary artery has been opened. It ended blindly in the wall of the ventricle. (b) Right auricle opened. There is a patent foramen ovale. The ductus arteriosus was patent.

FIG. 13a.—Tricuspid atresia. Female aged 2½ years.

towards the anterior wall of the chest. There is an unusually clear pulmonary window. The hilar shadows are smaller than normal owing to the reduced pulmonary circulation, and on the screen pulsation is not evident. Where there is a collateral circulation the hilum has a peppered appearance due to numerous small vessels. The lung fields are abnormally clear. In about a fifth of the cases there is a right aortic arch.

Electrocardiogram

Marked right axis is the rule. The P waves are often tall and peaked in lead 2. Conduction abnormalities are rare (Fig. 5).

Diagnosis

The tetralogy of Fallot is the commonest anomaly found in children who survive infancy. The history of cyanosis appearing at birth or in the neonatal period, together with polycythaemia and clubbing gives a good diagnostic lead. The diagnosis is essentially radiological and the characteristic silhouette shows no enlargement of the heart as a whole, a concave pulmonary arc and a blunt turned up apex. The aortic window is abnormally clear and there are ischaemic lung fields with small hila and absent hilar pulsation. Catheterization shows an increased pressure in the

FIG. 13b.—Tricuspid atresia. Female aged 2 years. Left anterior oblique view shows enlargement of the left ventricle. E.C.G. left axis.

right ventricle and a diminished pressure in the pulmonary artery. Final proof can be obtained from the angiocardigraphic picture. The most difficult differential diagnosis is from transposition of the vessels, or from a persistent truncus arteriosus. In transposition with a ventricular septal defect the heart is enlarged, the aortic window is not clear and there are congestive changes in the lung fields. In addition the vascular pedicle is narrow and widens as the patient is rotated to the oblique view. The persistent truncus shows an abnormally broad aorta and a narrow pedicle in the oblique view.

The Eisenmenger Complex

The Eisenmenger complex consists of an inter-ventricular septal defect with an overriding aorta and a normal or dilated pulmonary artery (Figs. 6 and 7). There is no deficit in the pulmonary circulation. Cyanosis is apt to occur and it is not known whether this is consequent upon secondary changes in the lungs due to the large volume of blood that flows to them, or due to a congenital abnormality of the alveolar endothelium preventing proper oxygenation. There is always a shunt of venous blood into the aorta and its volume varies, depending upon the position of the aorta in regard to the ventricular septal defect. It must

be very rare for this shunt alone to cause visible cyanosis.

Clinical Features

Cyanosis is usually absent until late childhood or adolescence and in childhood the patient is not severely incapacitated. The late development of cyanosis, which increases gradually, with the appearance of symptoms of dyspnoea and tiredness, is quite characteristic of the Eisenmenger anomaly. Clubbing is rarely marked. Haemoptysis occurs occasionally. The heart is slightly enlarged and there is a loud systolic murmur in the pulmonary area and in the third and fourth left intercostal spaces. Quite often there is a diastolic murmur and this may be due to either relative pulmonary incompetence or to aortic incompetence. The latter is due to a frequently associated abnormality of the aortic cusps. The pulmonary second sound is accentuated.

Radiology

The cardiac silhouette is variable in appearance depending upon the size of the pulmonary artery. There is always prominence of the pulmonary arc, and large pulsating pulmonary branches (Fig. 8). The right side of the heart is enlarged and very occasionally there may be the coeur-en-sabot appearance. Where the pulmonary artery is grossly enlarged the cardiac silhouette is identical with that of an auricular septal defect.

Electrocardiogram

The electrocardiogram is almost invariably found to show a right axis and large P waves are usual.

Diagnosis

In no other congenital abnormality is the diagnosis more frequently missed. This is perhaps due to failure in remembering the existence of the anomaly, or perhaps because in acyanotic cases the physical signs, particularly if there is a diastolic murmur, bear a superficial resemblance to those of rheumatic aortic valvular disease. Aortic stenosis can usually be differentiated by its small pulse pressure and the absence of any rheumatic history. The isolated ventricular septal defect may also present somewhat similar physical signs but, in this case, the heart is not enlarged and the pulmonary arc is never as prominent as in the Eisenmenger anomaly. The greatest difficulty is in connection with the auricular septal defect where the radiological picture may be identical. Basal systolic murmurs and thrills occur in both conditions. The circulation time is markedly shortened in the Eisenmenger type owing to the overriding aorta. The oxygen saturation of the arterial blood is always lowered and oxygen inhalation will not raise it to a normal figure owing to the venous arterial shunt. Angiocardiography will demonstrate the presence of an overriding aorta by simultaneous visualization of both aorta and

FIG. 14.—Tricuspid atresia. Cyanotic child aged 2 years. Left axis.

FIG. 15.—Tricuspid atresia. Male aged 12 years. Left axis.

FIG. 16.—Female aged 7 months with tricuspid atresia. Cyanosis and normal axis in electrocardiogram.

pulmonary artery. Measurements of intracardiac pressures are of great value, as also in other cyanotic cases where the differentiation from the tetralogy may be in question. Pulmonary artery pressure is always raised in the Eisenmenger type and abnormally low in the tetralogy. In a markedly cyanotic case there may be difficulty in the distinction between the tetralogy and this anomaly, but such cases are rare. Fluoroscopy shows pulsating pulmonary arteries and the lung fields are not ischaemic as in the tetralogy.

Isolated Pulmonary Stenosis

The term isolated pulmonary stenosis is applied to a group of cases in which there is pulmonary stenosis with or without a patent foramen ovale. It might also be applied to those cases of pulmonary stenosis where there is an interventricular septal defect, but the aorta is normally sited and does not override it. Some hold that pulmonary stenosis as a sole abnormality is a condition of great rarity. Those working in children's heart clinics will generally be struck by the number of cases which satisfy the diagnostic criteria usually applied to this anomaly. It is important to make a

FIG. 17.—Transposition of the vessels with ventricular septal defect. Pulmonary valve bicuspid. Marked cyanosis from birth.

separation between those cases that have a closed auricular septum and those which have a patent foramen ovale. In this latter group there is a shunt of blood from the right to the left so that a certain volume of blood is not returned to the lungs for oxygenation. Consequently, in some cases, cyanosis may appear relatively early and become marked.

Clinical Picture

In childhood there is no cyanosis unless the stenosis is of a severity amounting to functional atresia. The chief symptom is dyspnoea. Cyanosis usually develops in the teens in cases with a patent foramen ovale and gradually becomes permanent. It ultimately becomes marked and more severe than in the Eisenmenger anomaly and is accompanied by clubbing and polycythaemia. In the case without an auricular septal defect cyanosis is later still in appearance and depends to a great extent upon pulmonary vascular changes and pulmonary hypertension. The physical signs are a harsh systolic murmur and thrill in the second left space. The murmur is conducted towards the left clavicle and the pulmonary second sound may be diminished and sometimes absent. Occasion-

ally a diastolic murmur may be heard in the pulmonary area.

Radiology

The heart is enlarged, especially the right side. The pulmonary arc is convex and the hilar shadows a little smaller than usual (Fig. 9). There is some pulsation in the hila but no pulmonary congestion. The heart does not usually assume the coeur-en-sabot appearance of the tetralogy.

Electrocardiogram

This almost invariably shows a right axis deviation. The P waves are often tall and peaked (Fig. 10).

Diagnosis

It is important to make a distinction between the case with a patent foramen ovale and the case with a closed auricular septum. The latter properly belongs to the acyanotic group and cyanosis only develops late in life consequent upon pulmonary vascular changes. Pulmonary stenosis with open foramen ovale can be distinguished from the tetralogy by its radiological picture of a convex pulmonary arc and the presence of some hilar pulsation, and from the Eisenmenger complex by the large pulmonary artery and its branches. It

seems fair to say that in the present state of our knowledge the final and completely accurate diagnosis must depend upon the results of cardiac catheterization and angiocardiology.

Tricuspid Atresia

Until the present time tricuspid atresia has been regarded as a rather uncommon abnormality. The impetus given to the study of cyanotic children by the possibilities of surgical treatment has brought many of these cases to light.

There are several distinct anatomical types but the basic anomaly is a non-functioning right ventricle which is virtually cut off from the rest of the heart. The right ventricle may be entirely absent; it may exist as a blind cavity or it may communicate by a small orifice with the left ventricle (Figs. 11 and 12). The tricuspid valve may be entirely absent and represented only by a dimple in the floor of the right auricle, or it may show extreme hypoplasia. Corresponding to the defect of the tricuspid valve the pulmonary artery is commonly atresic or else arises in an abnormal situation. The right auricle is hypertrophied and the foramen ovale widely open. The auricular septum may be so defective that it makes the heart functionally biloculate. A patent ductus arteriosus is generally present in order to maintain a pul-

FIG. 18.—Transposition of the vessels with ventricular septal defect. Male aged 20 years.

FIG. 19.—Transposition of the vessels with ventricular septal defect. Male aged 13 years. Marked right axis. Tall P waves.

FIG. 20.—Persistent truncus arteriosus. Female aged 12 years, cyanotic since birth. At operation an enormous collateral circulation through bronchial and mediastinal vessels and no visible pulmonary artery. (a) Posteroanterior. (b) Left anterior oblique.

monary circulation and is usually of large dimension. In some cases transposition of the vessels coexists and may result in some prolongation of life.

The effects upon the circulation are such that the only means of escape of blood from the right auricle is through an auricular septal defect into the left auricle. A pulmonary blood supply is maintained through a patent ductus arteriosus in cases where there is pulmonary atresia, and in those cases where a stenotic and hypoplastic pulmonary artery arises from the rudimentary right ventricle some blood reaches the lungs through a defect in the ventricular septum and hence the pulmonary artery. A transposed pulmonary artery arising from the left ventricle sometimes affords a pulmonary circulation. Not only is there a mixture of arterial and venous bloods in the auricles but an inadequate volume of blood is passed to the lungs, consequently cyanosis is marked and severe.

Clinical Picture

Cyanosis of marked degree is the rule and

clubbing is commensurate with the cyanosis. The heart may be normal in size or slightly enlarged. A systolic murmur is occasionally present but more often murmurs are absent. It is very rare to hear a ductus murmur. The pulmonary second sound is well heard and never reduplicated.

Radiology

In the postero-anterior view the silhouette of the heart resembles that of the tetralogy (Figs. 13a and 13b). There is a concavity of the pulmonary arc, small root shadows with an ischaemic lung and an upturned apex reminiscent of the *coeur-en-sabot*. In the left anterior oblique view it is at once evident that the right ventricle is very small and that the left ventricle is hypertrophied. The pulmonary window is abnormally clear.

Electrocardiogram

The electrocardiogram consistently shows a left axis and for a long time this change was thought to be pathognomonic of tricuspid atresia (Figs. 14 and 15). It is now clear that other cyanotic cases may equally show a left axis, but the finding

of a left axis is strong evidence in favour of tricuspid atresia, as it is more frequent in this than in any other cyanotic congenital abnormality. Occasionally a normal axis is present (Fig. 16).

Diagnosis

The combination of cyanosis with left axis in the electrocardiogram and radiological evidence of a non-functioning right ventricle is presumptive evidence of tricuspid atresia. The only other congenital heart conditions in which a left axis occasionally may be associated with cyanosis are a single ventricle with diminutive outlet chamber, a persistent truncus arteriosus and transposition of the vessels. Cardiac catheterization will demonstrate the impossibility of entering the pulmonary artery in tricuspid atresia, whereas in the single ventricle and persistent truncus it is possible to observe the catheter moving freely within the cavity of the large ventricle. Complete transposition of the vessels with tricuspid atresia is usually associated with a large interventricular septal defect so that for a time an adequate volume of blood reaches the lungs and cyanosis may develop late. Like other cases of transposition without tricuspid atresia, the heart hypertrophies and failure develops.

Complete Transposition of the Great Vessels

This is a severe form of congenital abnormality only compatible with life in certain anatomical circumstances. The position of the great vessels is reversed so that the aorta arises from the right ventricle and the pulmonary artery from the left, both vessels being of normal dimensions (Fig. 17).

FIG. 21.—Persistent truncus arteriosus in an infant, with a 'sitting duck' silhouette.

This places the subject at a very grave disadvantage for although an adequate volume of blood reaches the lungs for oxygenation it cannot be returned to the systemic circulation unless the foetal passages remain open, or unless there is a ventricular septal defect. Accordingly, life is governed by the relative efficiency of either a patent ductus arteriosus, patent foramen ovale, or ventricular septal defect, alone or in combination with each other. From a practical point of view, only those cases with a ventricular septal defect survive for any length of time.

Clinical Picture

This depends to a great extent upon the associated abnormalities accompanying the transposed great vessels and their efficiency in allowing communication between the two circulations. If the ventricular septum is closed cyanosis is intense and progressive. Attacks of dyspnoea often occur. The heart rapidly enlarges and the patient dies of anoxaemia in days or weeks before there is time to develop clubbing or marked polycythaemia.

When there is an associated ventricular septal defect cyanosis may not be much in evidence at birth but soon appears and may be evident with sucking or crying. Once apparent it is progressive and accompanied by enlargement of the heart. Life may be prolonged for a year or in exceptional

FIG. 22.—Persistent truncus arteriosus. Female aged 14 years. Major deflection downwards in all three leads.

cases for several years. Prolonged survival may depend upon the position of the pulmonary artery vis-a-vis the septal defect.

In both groups of cases there may be a harsh systolic murmur but this has no definite characters and may be absent. A systolic murmur is more frequent and obvious when there is a ventricular septal defect.

Radiology

This is of great importance in diagnosis. Progressive enlargement of the heart over a short space of time has been stressed by Taussig (1947). In the antero-posterior view there is a narrow vascular pedicle which broadens as the patient is rotated into the oblique position (Fig. 18). Hilar pulsation is increased.

Electrocardiogram

Right axis deviation is generally present. The P waves tend to be large (Fig. 19).

Diagnosis

The clinical picture is one of severe cyanosis and dyspnoea occurring at birth. The radiological evidence is most important from the point of view of diagnosis. This discloses an enlarged heart with a narrow vascular pedicle which latter increases in width as the patient is rotated into the oblique position. There is pulmonary congestion. Serial photographs show rapid and progressive cardiac enlargement. The presence or absence of a systolic murmur is of small diagnostic importance. A bruit is most likely to be heard when a ventricular septal defect is present.

Persistent Truncus Arteriosus

A persistent truncus arteriosus is an abnormally large vessel occupying the site of the aorta and pulmonary artery and provided with a variable number of cusps. It overrides a high septal defect and the heart as a whole is enlarged, particularly the right ventricle. There are two important anatomical varieties distinguished by their pulmonary arteries either separately, or from a persistent truncus gives off right and left pulmonary arteries either separately or from a common stem; in the other group the pulmonary circulation is maintained through bronchial arteries and other collaterals. The type of pulmonary circulation influences the clinical picture so that cyanosis may not be apparent in those cases where the pulmonary arteries arise directly from the truncus unless these vessels are very small, but may be very marked and severe in the cases where a pulmonary circulation is through collateral channels such as the bronchial arteries.

Clinical Picture

As mentioned previously the presence of cyanosis depends upon the type of pulmonary circulation. Clubbing is proportional to the cyanosis, especially in cases with a bronchial artery circulation. There may be particularly severe dyspnoea in these latter. The heart is enlarged in all cases, whether cyanosis is present or absent, and may be greatly enlarged with deformity of the left chest in some cases. A systolic murmur and thrill is usually present at the base. A continuous murmur has been occasionally described and its site of maximum intensity is at a lower level than in the patent ductus arteriosus. The second sound is abnormally loud and is not reduplicated.

Radiology

In infancy there is gross enlargement of the heart with a cardiac silhouette which bears some resemblance to a sitting duck. The heart is enlarged to both left and right with a concave pulmonary arc, prominent aortic knuckle and an upturned apex from left ventricular hypertrophy (Fig. 21). In the left anterior oblique view the right ventricular border passes like a shelf from the base of the aorta towards the anterior chest wall. The hilar shadows in the presence of a bronchial artery circulation are small and peppered, and the lung fields abnormally clear. As the child grows older the thorax lengthens and the heart becomes more vertical; there is a change in the shape of the heart so that it comes to resemble the silhouette of the tetralogy so closely that distinction from it is difficult. A left anterior oblique view may furnish a clue to the diagnosis at this age but not always (Fig. 20a and b).

Electrocardiogram

This may show a right axis. Occasionally there is a left axis. In several cases there has been a bizarre electrocardiogram with the major deflection downwards in all three leads (Fig. 22).

Diagnosis

The radiological picture in infancy is characteristic with an extremely large heart and a peculiar shelf-like projection of the right ventricle in the left oblique view. The vascular pedicle is wide. The point may be made that this abnormality presents the largest heart of any congenital malformation, the only other comparable abnormality being the infantile type of coarctation. In older cases diagnosis is more difficult because the heart silhouette may so closely resemble the tetralogy that distinction from it is impossible on radiological grounds alone. Catheterization of the heart shows that the tip of the catheter may be freely moved about and a blood sample may have

the same oxygen content as blood from the femoral artery, demonstrating a single ventricle in the functional if not always in the completely anatomical sense. Angiocardiography may also have obvious advantages in diagnosis.

Single Ventricle with Rudimentary Outlet Chamber

This may be regarded as a rather rare anomaly of which there are several anatomical types. It is of importance because one of these types may benefit from surgical operation. Embryologically the anomaly represents persistence of the bulbus cordis as a rudimentary chamber. Anatomically, the rudimentary chamber occupies the site of the normal pulmonary conus. It is separated from a single ventricle by a band of muscular tissue, and communicates freely with it. Both auriculo-ventricular orifices open into the single ventricle. There is thus a functional cor biatriatum triloculare, and if there is associated maldevelopment of the auricular septum, a functional cor biloculare results. One or both of the great vessels may arise from the rudimentary chamber, and depending upon whether there is associated maldevelopment of the great vessels, in normal or in transposed relationship. The great vessel arising from the rudimentary cavity is usually small. An aorta arising from the single ventricle, and the pulmonary artery from the rudimentary outlet chamber, is an uncommon abnormality, usually known as the Holmes' heart, of which but few examples are recorded (Fig. 23). More commonly the vessels are transposed.

The effects of the abnormality upon the circulation will naturally vary according to the anatomical arrangement of the great vessels. In all cases, because both auricles empty into the single ventricle there is a mixture of arterial and venous bloods; in consequence the inhalation of oxygen can never bring oxygen saturation to a normal level. If both great vessels arise from the rudimentary chamber and are equal in size, equal volumes of blood reach the respective circulations and arterial and venous bloods are mixed in about equal proportions and one finds some degree of cyanosis. When the pulmonary artery arises from the single ventricle and a smaller aorta from the diminutive chamber, a greater volume of blood reaches the lungs than the volume reaching the systemic circulation and consequently cyanosis is usually not apparent. An aorta arising from the single ventricle and a small pulmonary artery arising from the rudimentary chamber, entails a diminished volume of blood to the lungs for oxygenation together with a large proportion of venous blood mixing with arterial blood in the single ventricle, hence cyanosis is marked and

FIG. 23.—Single ventricle with diminutive outlet chamber. The great vessels are not transposed. A probe is placed in the small foramen communicating between the single ventricle and the small outlet chamber. An anastomotic operation has been performed.

severe. It is in this latter type that benefit may be expected from surgical operation. Obviously in the case where the pulmonary artery arises from the single ventricle the volume of blood supplied to the lungs is adequate and surgery can be of no value. A clinical manifestation of this type of case is that the subject tends to be underdeveloped because of the small volume of blood reaching the systemic circulation through a small aorta. There remains the type in which both vessels may arise from the small chamber with pulmonary stenosis, and if this latter is at all severe, benefit may be expected from surgery provided that there is a low pulmonary artery pressure. If pressures are about the same in both aorta and pulmonary artery, which they will be if there is not marked pulmonary stenosis, an anastomosis is useless.

Clinical Picture

There are very great difficulties in the recognition of this type of case. Cyanosis may be marked where the pulmonary artery arises from the rudimentary chamber or where there is pulmonary

FIG. 24.—Single ventricle with diminutive outlet chamber. The great vessels not transposed. Pulmonary artery arises from the diminutive chamber. Marked cyanosis and left axis in the electrocardiogram. Female aged 7.

stenosis with both vessels arising from the small chamber. In other cases clinical cyanosis may be absent, but in all cases, whether cyanotic or not, there is an increased level of reduced haemoglobin in the arterial blood and a normal oxygen saturation cannot be produced by the inhalation of oxygen. The heart is not enlarged. Murmurs may or may not be present, and if present are systolic in time and have no definite characters or localization.

Radiology

X-ray examination furnishes important diagnostic clues. In the very young child there may be prominence of the pulmonary conus due to the fact that the rudimentary chamber occupies this site. At a later stage of childhood, as the length of the chest increases, the heart becomes more vertical and this apparent prominence disappears, so there may be little in the postero-anterior view to distinguish it from the tetralogy (Fig. 24). In the left anterior oblique view there is no enlargement of the right ventricle towards the chest wall and the line of the aorta is virtually continuous as a straight line with that of the heart border. When the pulmonary artery arises from the single ventricle pulmonary congestion is present.

Electrocardiogram

There may be no deviation of the electrical axis. Right axis has been reported. Left axis occurred in a case of the author (Fig. 25).

Diagnosis

The difficulties in differential diagnosis were well exemplified in a personal case of a child aged 7. She had been cyanotic since birth and was markedly cyanotic with clubbing when first seen. Her X-ray examination disclosed a small or absent right ventricle and her electrocardiogram a left axis. A confident diagnosis of tricuspid atresia was made and following further investigation she was submitted to operation. This was successfully done, but the child died suddenly and unexpectedly. Autopsy disclosed that she had a single ventricle with diminutive outlet chamber from which the pulmonary artery arose, a situation identical with that of the Holmes heart.

The lesions most likely to cause difficulty in differential diagnosis are tricuspid atresia and the tetralogy of Fallot in cases with marked cyanosis. It is evident that the age of the patient will, to some extent, determine the radiological picture and in infancy an enlarged pulmonary arc, with or without congestion of the lung fields, would exclude the tetralogy. In later childhood the

FIG. 25.—Single ventricle with diminutive outlet chamber. Great vessels not transposed. Left axis, tall pointed P waves, deep Q3.

THE PRINCIPAL CLINICAL FINDINGS SET OUT FOR CONVENIENT REFERENCE

	Tetralogy of Fallot	Eisenmenger	Pulmonary Stenosis F.O. Patent	Tricuspid Atresia	Transposition of Vessels	Persistent Truncus Arteriosus	Single Ventricle Diminutive Outlet Chamber Vessels Not Transposed
Cyanosis ..	Early and marked	Slight to moderate and late	Appears early, progressive and marked	Marked	Marked	Marked	Marked
Clubbing ..	Marked	Slight	Slight to marked	Marked	Marked in surviving cases	Marked	Marked
Polycythaemia ..	Severe	Later stages	Severe	Severe	Severe in surviving cases	Severe	Severe
Symptoms ..	Severe	None till late childhood	Present early	Present early	Present early	Present and severe	Severe and early
Physical capacity ..	Grossly impaired	Little impaired	Severely impaired	Grossly impaired	Grossly impaired	Grossly impaired	Grossly impaired
Dyspnoea ..	Severe, Squats	Some	Severe	Severe, Squats	Severe	Severe	Severe
Systolic murmur ..	In pulmonary area. May be absent	Pulmonary area	Pulmonary area	Absent; if present not characteristic	Often absent; present with V.S. defect	Present or absent—not characteristic—continuous murmur in 4th I.C.S. Occasional	Present or absent; not characteristic
Diastolic murmur ..	Rarely present	Frequent from aortic or pulmonary incompetence	Occasionally present	Absent	Absent	Occasional	Absent
Pulmonary Second Sound ..	Often unaltered	Accentuated	Often diminished	Loud and not reduplicated	Unaltered	Loud and pure	Unaltered
X-ray Enlargement	Normal heart size	Slight to moderate	Slight enlargement	Often present	Enlargement	Gross enlargement	Moderate
Pulmonary arc ..	Concave	Convex and prominent	Convex and prominent	Concave	Concave	Concave in late childhood	Early slight convexity, late concavity
Hilar shadows ..	Small	Normal or large	Normal or slightly smaller	Small	Normal	Small	Small
Hilar pulsation ..	Nil	Present	Present	Absent	Present	Absent	Absent
Pulmonary congestion ..	None	May be present	None	None	Present	None	None
Ecg R axis ..	Marked	Marked	Marked	Left axis	Right axis usual occasional normal axis	Right axis; occasional main deflections downwards—1, 2 and 3 enlarged	Right or left axis
P waves ..	Tall and peaked	Enlarged	Enlarged	Enlarged	Enlarged	Enlarged	Enlarged
Circulation time ..	Shortened	Shortened	Unaltered	Unaltered	Unaltered	—	—

difficulties are very real, particularly in the case with a left axis in the electrocardiogram and a left anterior oblique view closely resembling that of the non-functioning right ventricle of tricuspid atresia. In tricuspid atresia it will be impossible to introduce the catheter into the pulmonary artery. In

cases with a common ventricle the catheter moves freely within the heart and may be readily introduced into the aorta if this arises from the common ventricle. Further, a sample of blood from the common ventricle would have the same oxygen content as blood from the femoral artery.

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