Diagnosis and Management of Common Malformations of the Heart

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ACCURATE diagnosis and the proper management of children with malformations of the heart becomes increasingly important as more and more malformations become amenable to surgery. Even patients who cannot be helped by surgery should be encouraged to lead useful lives. In many instances it is quite as much the doctor's attitude as the malformation which makes an invalid out of the child.

Fortunately a number of malformations can be diagnosed with comparative ease by simple clinical methods, without recourse to cardiac catheterization or angiocardiography. The latter procedures are useful adjuncts, but by themselves seldom give the correct answer. Furthermore, they are not innocuous procedures; therefore they should not be used routinely, but only to obtain specific information on some particular point. The purpose of this paper is to review the common malformations of the heart which can be diagnosed clinically and to give the clinician a number of hints concerning the diagnosis and also some general principles concerning the management of these children.

I: Malformations of the Heart Which Show No Cyanosis

Patent Ductus Arteriosus

Persistent patency of the ductus arteriosus is usually a relatively easy diagnosis. The diagnosis is based on the occurrence of a continuous murmur over the pulmonary area in a person who shows no cyanosis. The murmur may vary considerably in intensity, but usually it has a systolic accentuation and the second sound is readily audible in the middle of the murmur. The electrocardiogram ordinarily shows a balanced axis or a tendency to left ventricular hypertrophy.

The x-ray commonly reveals fullness of the pulmonary conus and frequently there is evidence of slight left auricular enlargement. The size of the heart varies with the size of the ductus. In general, the larger the ductus, the larger is the heart and the more pronounced are the hilar markings. A hilar dance is, however, not common. The red blood cell count, the amount of available hemoglobin, and the hematocrit reading should be within normal limits.

The occurrence of cyanosis or polycythemia is strongly against the diagnosis of patency of the ductus arteriosus; even if there is patency of the ductus, either of these findings is almost invariably a contraindication to the surgical closure of the ductus arteriosus.

The fact that the continuous murmur seldom develops before 1 year of age and often not until the patient is 3 or 4 years of age, means that the diagnosis prior to this time is difficult. Fortunately in most instances the condition causes little or no disability. Therefore, in the vast majority of patients, early diagnosis is not necessary.

Occasionally, however, a large patent ductus arteriosus may cause great difficulty in early
infancy.\textsuperscript{1} Therefore an infant with great cardiac enlargement and evidence of excessive pulmonary blood flow who has a balanced electrocardiogram and has only a systolic murmur or both a systolic murmur and a mid-diastolic murmur but no continuous murmur, should be sent to a specialist for study.

When patency of the ductus arteriosus occurs as an isolated malformation, either ligation or division of the ductus arteriosus restores the heart to normal.\textsuperscript{5,6} Even if there is an additional malformation, provided there is no cyanosis and the oxygen saturation of the arterial blood is normal, in almost every instance closure of the ductus arteriosus lessens the strain on the heart.

The ideal age for operation is between 4 and 6 years; nevertheless, an infant or an adult in heart failure may be greatly benefited by operation.

\textit{Coarctation of the Aorta}

Coarctation of the aorta is usually easy to diagnose. The diagnosis is based upon the finding of a strong pulse in the upper extremities combined with a weak or absent pulse in the lower extremities. This finding should always suggest the possibility of a coarctation of the aorta, and should lead to the search for evidence of collateral circulation. Pulsating vessels are frequently palpable in the intercroupular region. The occurrence of a systolic murmur in this area which is not audible over the precordial is a relatively common finding in a coarctation of the aorta.

The most characteristic of all signs is notching of the ribs. Notching of the ribs may be seen as early as 3 years of age, but it is usually not apparent until 12 or 14 years of age. Its occurrence in adults is the rule. Indeed absence of notching of the ribs in an adult who has clinical evidence of coarctation of the aorta should always suggest the possibility of coarctation of the abdominal aorta. Under such circumstances either angiocardiography or arteriography is indicated to determine the location of the constriction. If arteriography is attempted, 35 per cent Diodrast should be used, as the injection of 70 per cent Diodrast directly into the aorta is an extremely dangerous procedure.

When coarctation of the aorta occurs proximal to the entrance of the ductus arteriosus, the low pressure in the descending aorta causes the blood to flow from the pulmonary artery to the descending aorta. Unless the ductus arteriosus undergoes prompt obliteration the infant may develop severe right-sided heart failure. If such an infant fails to respond to digitalis therapy, early operation is indicated.

Coarctation of the aorta may occur in combination with other anomalies. The occurrence of an additional malformation, especially that of aortic insufficiency, is usually an added indication for operation.

Coarctation of the aorta is another malformation in which operation may restore the heart and circulation to normal.\textsuperscript{5,6} Furthermore there is strong evidence that atherosclerotic changes begin at an earlier age in a patient with hypertension than in the normal individual. This means that operation is easier and more likely to be successful in children than in adults.

The author does not, however, believe that surgery is indicated for every patient with a coarctation of the aorta. Early operation is indicated if the child has symptoms referable to the malformation, if he has marked hypertension or if he has electrocardiographic evidence of left ventricular strain. The optimum time for operation is between 4 and 10 or 12 years of age. If, however, the patient is asymptomatic and has a normal blood pressure and the condition is picked up upon routine examination, he can well afford to wait until the surgical technic has been still further perfected.

\textit{Vascular Ring}

A vascular ring which encircles the trachea and esophagus may result from the persistence of both the right and left aortic arches which unite posteriorly to form the descending aorta, or it may be formed by the combination of a right aortic arch and left descending aorta when the ductus arteriosus or a strand thereof persists in its normal position and extends from
the pulmonary artery to the descending aorta on the left side. In the majority of instances, a vascular ring occurs as an isolated anomaly.

Stridor is the outstanding symptom. It is usually present from birth. Dysphagia is common and not infrequently the infant develops attacks of dyspnea during which he may become deeply cyanotic. These symptoms may be alleviated by hyperextension of the head and neck.

The combination of respiratory difficulty and difficulty in feeding, with the consequent failure to gain, render these infants very susceptible to tracheitis, bronchitis, and pneumonia.

† The condition can usually be readily diagnosed by x-ray or fluoroscopic examination. The retroesophageal aorta causes marked anterior displacement of the esophagus at the level of the aortic arch. A vascular ring which results from a double aortic arch commonly causes bilateral compression of the trachea and esophagus. The diagnosis may be established by a tracheogram. Delineation of the esophagus by a radio-opaque material in the lateral or oblique position usually suffices. If barium is given, the mixture should be extremely thin, because the dysphagia may precipitate an attack of dyspnea. If the infant has difficulty in swallowing, Lipiodol should be used, as aspiration of the Lipiodol does no harm.

The difficulty caused by a vascular ring can be greatly helped by operation. The division of the smaller segment abolishes the ring. Such an operation should cure the condition. Unfortunately, however, the trachea may be permanently constricted or the adhesions to the surrounding tissues may be such that the division of the vascular ring does not relieve the obstruction. The improvement is greater if the tissues of the anterior mediastinum are pulled up and sutured to the sternum. Even though operation is not always successful, if the baby is having attacks of dyspnea and cyanosis, operation is urgently indicated. On the other hand, when the patient is asymptomatic and the condition is picked up on routine examination, operation is not indicated.

**Auricular Septal Defect and the Lutembacher Syndrome**

Auricular septal defects and the Lutembacher syndrome are malformations which can be diagnosed with relative ease and accuracy. When a gross defect in the auricular septum occurs in combination with great dilatation of the pulmonary artery and congenital or acquired mitral stenosis, the condition constitutes a Lutembacher syndrome. Both malformations lead to great right-sided cardiac enlargement which frequently causes left-sided chest deformity. The patient often shows the frail build of the gracile habitus. The shunt is overwhelmingly left-to-right, and anything which increases the strain on the left side of the heart increases the volume of the shunt.

Inasmuch as the shunt is from left to right, there is no clubbing and no polycythemia; there may, however, be minimal cyanosis of the tips of the fingers and toes. The heart becomes tremendously enlarged both to the right and to the left. The apex beat is displaced down and out. A harsh basal systolic murmur with a snapping, reduplicated second sound over the pulmonary area is the rule. In addition there is usually both a systolic murmur and a low-pitched mid-diastolic murmur at the apex.

The electrocardiogram shows slight right axis deviation and widening of the QRS complex; the precordial leads generally show evidence of a right bundle branch block or the pattern of right ventricular strain.

The x-ray contour shows great right-sided cardiac enlargement and a small aortic knob; even in the presence of mitral stenosis there is seldom evidence of left auricular enlargement. In cases of a Lutembacher syndrome, there is, in addition, enormous dilatation of the pulmonary artery and a conspicuous hilar dance.

These patients frequently suffer from repeated attacks of bronchitis and pneumonia. In addition they are very susceptible to rheumatic fever and to all sorts of cardiac arrhythmias. The use of prophylactic chemotherapy or antibiotics lessens the frequency of respiratory infections and rheumatic fever. Digitalis and other measures may help to control the arrhythmias.
Closure of auricular defects is being attempted but has not yet been accomplished with sufficient success and a sufficiently low mortality rate to justify operation unless the patient is suffering from severe and intractable heart failure.

Most of these patients, in spite of the great cardiac enlargement, do well. The malformation causes remarkably little difficulty during pregnancy. Therefore sterilization or interruption of pregnancy is seldom justified. In most instances, relative longevity is the rule.

**Ventricular Septal Defect**

Ventricular septal defect, or maladie de Roger, is the probable diagnosis in a patient with a heart of normal size and no limitation of activity, who has a harsh systolic murmur and thrill maximal in the midsternal region. The murmur radiates in all directions and may be heard posteriorly. X-ray shows the heart to be of normal size and shape. A normal electrocardiogram is the rule.

Most of these patients are asymptomatic and require no treatment except for the general prophylactic measures which are indicated for all patients with malformations of the heart (see section III).

**II: Malformations Which Cause Persistent Cyanosis**

Cyanosis is the bluish tinge to the skin which results from the presence of reduced hemoglobin in the circulating blood. Cyanosis may, however, be due to increased deoxygenation of the blood in the peripheral tissues; under such circumstances the oxygen saturation of the arterial blood is normal. When cyanosis is due to a venous-arterial shunt the oxygen saturation of the arterial blood is reduced. In almost every such instance the degree of oxygen unsaturation increases still further with exercise. Therefore, when in doubt concerning the occurrence or significance of cyanosis, the effect of exercise should be tested. In a patient with a venous-arterial shunt the inhalation of oxygen may increase the oxygen saturation of the arterial blood, but usually it will not raise it to normal. In contrast to this, when cyanosis is due to inability to oxygenate the blood in the lungs, as in cases of cor pulmonale, the inhalation of oxygen causes prompt disappearance of the cyanosis and the arterial oxygen saturation rises to normal.

Persistent oxygen unsaturation of the arterial blood leads to polycythemia, which renders the cyanosis more readily apparent. The low arterial oxygen saturation and the polycythemia cause clubbing of the extremities.

Broadly speaking, malformations which cause persistent cyanosis place a strain on the right side of the heart. Either the aorta arises in part or completely from the right ventricle, or the malformation places a strain on the right side of the heart of such a nature that venous blood is shunted into the systemic circulation through the foramen ovale, or there is underdevelopment of the left side of the heart as in aortic atresia or marked hypoplasia of the aorta and the systemic circulation is maintained by the right ventricle through the ductus arteriosus.

The age at which cyanosis becomes apparent offers a clue to the diagnosis. Cyanosis which is present in early infancy is generally due to the direct shunting of venous blood into the systemic circulation. The commonest malformations in this group are complete transposition of the great vessels and pulmonary stenosis or atresia. Cyanosis which becomes apparent in early childhood, that is, between 2 and 6 or 7 years of age, is usually due to a venous-arterial shunt from right auricle to left auricle through the foramen ovale. In such instances the valve of the foramen ovale has been forced open by the high pressure in the right auricle, as for example valvular pulmonary stenosis with an intact ventricular septum or Ebstein's disease. Cyanosis which develops at or shortly after puberty is usually associated with pulmonary hypertension, as for example, an Eisenmenger complex or a cor pulmonale. Pulmonary hypertension may, however, occur in infants and young children. If pulmonary hypertension occurs early or is secondary to a malformation of the left side of the heart, either the ductus arteriosus or the foramen ovale may remain patent and thereby a right-to-left shunt is established.

In malformations which cause persistent
cyanosis, x-ray and fluoroscopic findings are usually of diagnostic aid, especially as regards the size and position of the pulmonary artery and the volume of the pulmonary blood flow.

The term "pulmonary conus" has been used to connote both the "main segment of the pulmonary artery" and the "infundibular region" or the "outflow tract" of the right ventricle. The term "pulmonary conus" means the first convexity in the cardiac shadow to the left of the sternum below the shadow of the aortic knob. Recent studies with Diodrast have shown that this shadow is usually cast by the main pulmonary artery as it arises from the right ventricle. When the pulmonary artery is large and normally placed, the pulmonary conus is full. A concave curve at the base of the heart to the left of the sternum in a patient with persistent cyanosis occurs when the pulmonary artery is diminutive, absent, or misplaced.

The volume of the pulmonary blood flow can be estimated by the vascularity of the lung fields. In patients with pulmonary stenosis or atresia, the lung fields are excessively clear. In contrast to this, large blotchy shadows which radiate from the hilar regions to the lungs are indicative of large pulmonary vessels and excessive pulmonary blood flow. Pulsations in the hilar shadows vary both with the size of the pulmonary arteries and with the pressure within these vessels.

When conspicuous hilar shadows are combined with a concave curve at the base of the heart to the left of the sternum, it means a large posteriorly placed pulmonary artery, that is, a complete transposition of the great vessels and a dilated pulmonary artery. This condition may occur with two ventricles or when there is only a single ventricle.

The Tetralogy of Fallot

The tetralogy of Fallot consists of pulmonary stenosis or atresia, combined with dextroposition of the aorta, a high ventricular septal defect, and right ventricular hypertrophy. As in almost all malformations of the heart, the severity of the abnormality is subject to wide variation. Thus the pulmonary stenosis may be so extreme that the pulmonary orifice is atretic; in other instances the pulmonary stenosis is slight and the condition may be compatible with life for a number of years.

Cyanosis may be present at birth. Frequently the cyanosis does not become apparent until 3 to 6 months of age, and it may not be appreciated until the infant starts to walk. Many infants with severe degrees of pulmonary stenosis suffer from attacks of paroxysmal dyspnea which may progress to loss of consciousness. For such infants the most critical period is from 6 to 18 months of age. From 18 months to 2 years of age, the infant generally holds his own, and thereafter he starts to improve.

Clubbing develops in early childhood and becomes progressively more pronounced. The red blood cell count, the amount of available hemoglobin, and the hematocrit reading are increased. The oxygen saturation of the arterial blood is reduced and falls still further with exercise. Polycythemia increases with age. Eventually the patient is handicapped not only by the reduction of the arterial oxygen saturation but also by the increased viscosity of the blood.

The heart is but slightly if at all enlarged. A basal systolic murmur and thrill are the rule. The second sound at the base of the heart to the left of the sternum is weak or absent; it is never reduplicated. The electrocardiogram shows right axis deviation and right ventricular hypertrophy. The x-ray shows a concave curve to the left of the sternum and diminished hilar markings; the aorta may arch to the right or to the left; the pulmonary window is clear. Arm-to-tongue circulation time is short.

In a tetralogy of Fallot, the more intense the cyanosis, the greater is the patient's incapacity, and the fainter is the murmur. A very cyanotic patient may have no murmur, but if there is a minimal cyanosis and no murmur it is not a tetralogy of Fallot. There may, however, be a loud systolic murmur, minimal cyanosis, a virtually normal red blood cell count, and yet the child may suffer from a rapid drop in the arterial oxygen saturation with exercise, and consequently may be extremely incapacitated. Such children may be able to walk only a few
feet or stand only for a few minutes before they have to squat.
These children, as well as those who show cyanosis and polycythemia, can be greatly helped by operation. The optimum time for operation is between 8 and 14 years. Any operation which increases the circulation to the lungs will be of benefit. A Blalock anastomosis\textsuperscript{12} gives excellent results, both immediate and long time.\textsuperscript{13} A Pott's procedure\textsuperscript{14} also gives good results.

\textit{Tricuspid Atresia}

Tricuspid atresia may occur in combination with a variety of malformations. The most common of these is defective development of the right ventricle and pulmonary stenosis. This malformation closely resembles that of a tetralogy of Fallot except that the electrocardiogram shows a left axis deviation and left ventricular hypertrophy. When the tricuspid valve is atretic, the blood cannot leave the right auricle in the normal manner. It must escape through an opening in the auricular septum to the left auricle. When this opening is small and difficulty is encountered in the expulsion of blood from the right auricle, the auricular contractions are transmitted to the liver and these pulsations are palpable at its margin.

When tricuspid atresia is associated with pulmonary stenosis or atresia, the management and treatment are the same as for a tetralogy of Fallot. Nevertheless, owing to the variety of associated anomalies, operation may lead to progressive cardiac enlargement and cardiac failure. The over-all mortality rate is nearly twice as high as for a tetralogy of Fallot. Consequently only the seriously incapacitated patient should be recommended for surgery.

\textit{Pure Pulmonary Stenosis}

Pure pulmonary stenosis, pulmonary valvular stenosis with an intact ventricular septum, can usually be diagnosed by simple clinical studies.\textsuperscript{15, 16} The diagnosis is based upon the finding of a harsh systolic murmur and thrill over the pulmonary area, combined with a weak or absent pulmonic second sound and electrocardiographic evidence of a marked right axis deviation and either right ventricular hypertrophy or a right bundle branch block. The size of the heart and the degree of dyspnea vary with the severity of the pulmonary stenosis. The obstruction at the pulmonary orifice causes difficulty in the ejection of blood from the right ventricle and increases the work required of this chamber. The pressure within the right ventricle gradually rises. There results right ventricular hypertrophy.

If the pulmonary stenosis is severe, the heart becomes greatly enlarged. The pulmonary artery distal to the stenosis becomes dilated and its main branches are large and conspicuous; nevertheless the pulsations within these vessels are minimal or absent. Sooner or later the increased pressure in the right ventricle renders it difficult for the right auricle to empty itself. The right auricle undergoes dilatation and hypertrophy. Not infrequently the forceful contractions of the right auricle cause the auricular pulsations to be palpable at the margin of the liver.

The presence or absence of cyanosis depends upon whether or not the foramen ovale is completely sealed. If the foramen ovale is completely sealed, there is no cyanosis. If, however, there is probe patency of the foramen ovale, the high pressure in the right auricle will force the valve covering the foramen ovale to open and a right-to-left shunt will be established. Although cyanosis of this origin may be present at birth, it usually develops insidiously and becomes manifest between 2 and 6 years of age.

The pulmonary stenosis prevents the normal increase in the pulmonary blood flow with exercise, and thus causes dyspnea on exertion. If the stenosis is extreme, the condition leads to progressive cardiac enlargement and eventually to severe right-sided cardiac failure with edema and ascites. As long as the heart is able to compensate for the increased work demanded of it, the patient remains asymptomatic. This means that cardiac enlargement precedes symptoms. For this reason patients suspected of this abnormality should be followed carefully for evidence of increase in heart size.

If the heart is normal in size and the patient is asymptomatic and the pressure within the
right ventricle is but slightly increased, operation is not indicated. If, however, the pressure in the right ventricle is higher than the systemic pressure, or if there is evidence of progressive cardiac enlargement, operation is indicated. The occurrence of cyanosis is also an indication for operation.

Valvulotomy\textsuperscript{17-19} relieves the pulmonary stenosis. Usually the valve is so fibrotic that cutting the valve does not cause it to become insufficient. Although the systolic murmur and thrill generally persist after operation, in most instances the cyanosis completely disappears and the oxygen saturation of the arterial blood rises to normal and the patient's exercise tolerance is greatly increased.

\textit{The Eisenmenger Complex}

The Eisenmenger complex consists of dextroposition of the aorta combined with a normally placed pulmonary artery which is of normal size or greatly dilated. The dextroposition of the aorta means that there is a high ventricular septal defect.

The difficulty caused by this malformation during infancy is due to excessive pulmonary blood flow. A number of infants die of cardiac failure. Those who survive, generally improve during childhood. Cyanosis is absent until late childhood or early adult life. Thereafter there is a progressive increase in the signs and symptoms. In the late stages, hemoptyses are relatively common.

Examination of the heart shows slight cardiac enlargement. Usually there is a systolic murmur and there are an early diastolic murmur. The x-ray shows fullness of the pulmonary conus, a prominent pulmonary artery and a hilar dance. The electrocardiogram usually shows a right axis deviation and right ventricular hypertrophy. Cardiac catheterization shows a high pressure in the right ventricle and evidence of pulmonary hypertension. The pressure in the pulmonary artery is approximately the same as that in the systemic circulation. The oxygen content of the blood in the pulmonary artery is higher than that in the right auricle and that in the right ventricle, and the oxygen saturation of the arterial blood is reduced. The malformation at the present time (1952) is not amenable to surgery.

\textit{The Taussig-Bing Malformation}

The Taussig-Bing malformation\textsuperscript{20} is a transposed aorta combined with a pulmonary artery which arises mainly from the right ventricle but partially over-rides the ventricular septum and thereby readily receives blood from both ventricles.

Patients with this malformation are cyanotic from birth; nevertheless, these infants do not suffer from paroxysmal dyspnea; children with this malformation are moderately incapacitated but seldom squat when tired.

The heart is but slightly enlarged. Usually there is a soft systolic murmur. The second sound at the base is generally accentuated and frequently reduplicated. The x-ray reveals fullness of the pulmonary conus and increased hilar shadows which frequently show expansile pulsations. The electrocardiogram shows a right axis deviation and evidence of right ventricular hypertrophy.

The findings on cardiac catheterization are closely similar to those of an Eisenmenger complex. Indeed, the oxygen saturation of the blood in the pulmonary artery is higher than that in the aorta. It is frequently possible to catheterize the aorta and pulmonary artery with equal ease. The outstanding difference between this malformation and an Eisenmenger complex is that the cyanosis dates from birth.

Patients with this malformation may be slightly benefited by the creation of an auricular septal defect. Such an operation generally increases the child's exercise tolerance but seldom causes a significant decrease in the red blood cell count, the amount of available hemoglobin, or in the hematocrit reading.

\textit{Complete Transposition of the Great Vessels}

Complete transposition of the great vessels means that the aorta arises from the right ventricle and the pulmonary artery from the left ventricle. Usually both great vessels are of normal size. The pulmonary artery, however, may be greatly dilated or it may be stenotic.

Although cyanosis may not be present at birth, it is always manifest at an early age and becomes progressively more intense. The infant breathes rapidly and may suffer from
episodes of loss of consciousness. Attacks of paroxysmal dyspnea are, however, rare.

A systolic murmur is usually audible, and a gallop rhythm frequently develops. There is engorgement of the liver and congestion in the lungs. In early infancy while the ductus remains patent, some of the oxygenated blood which is pumped from the left ventricle into the pulmonary artery flows through the ductus arteriosus to the lower extremities; hence the head and upper extremities are more cyanotic than are the feet and lower extremities.

The x-ray usually reveals that the heart is enlarged both to the right and to the left. In the anterior-posterior view the shadow cast by the great vessels is narrow and there is a concave curve at the base of the heart to the left of the sternum; in addition the vascular markings are increased and extend far into the lung fields.

The electrocardiogram usually shows right axis deviation and right ventricular hypertrophy. A left axis deviation may occur when a complete transposition of the great vessels is combined with tricuspid atresia.

The malformation usually leads to progressive cardiac enlargement with engorgement of the liver and congestion in the lungs. Death generally occurs at an early age. Occasionally a circulatory balance is established and the heart ceases to enlarge. Under such circumstances the condition may be compatible with life for a number of years.

The malformation causes marked stunting of growth. The child is usually severely incapacitated, and commonly squats when tired.

As the child grows and the diaphragm descends, the contour of the heart comes to resemble that of a tetralogy of Fallot. The x-ray, however, always reveals slight cardiac enlargement and the hilar markings are more pronounced than in a tetralogy of Fallot. Many blood vessels are viewed on end and the vascular markings extend nearly to the periphery of the lungs.

When the pulmonary artery is greatly dilated, there are large blotchy vascular shadows in both hilar regions, which upon fluoroscopy usually show expansile pulsations.

When the pulmonary artery is stenosed, the condition may be extremely difficult to differentiate from a tetralogy of Fallot unless the aorta is grossly misplaced. If the aorta arises from the normal outflow tract of the right ventricle and arches boldly to the left, the prominent arc to the left of the sternum in the region of the normal pulmonary artery combined with excessively clear lung fields seen upon x-ray or fluoroscopy, offers the clue to the correct diagnosis.

A complete transposition of the great vessels combined with pulmonary stenosis, may be helped by Dr. Blalock's double procedure,21 systemic-pulmonary artery anastomosis and the creation of an auricular septal defect.

Truncus Arteriosus

Truncus arteriosus means that there is but a single great vessel which receives the blood from both ventricles and directs the blood to the body and to the lungs. The condition is termed a "true" truncus arteriosus when the pulmonary arteries arise directly from the base of the common trunk or when there is but a single great vessel and no remnant of the pulmonary artery is found. When, however, any remnant of the pulmonary artery is found the condition is termed a "pseudo" truncus arteriosus.

Persons with this malformation show varying degrees of cyanosis and incapacity. The cyanosis and incapacity vary inversely with the adequacy of the pulmonary blood flow. Broadly speaking, the incapacity is less than in other malformations with the corresponding degree of cyanosis. These infants seldom suffer from paroxysmal dyspnea. Many children with moderate cyanosis can walk long distances at a slow pace; they do not squat when tired.

On physical examination the distinctive findings are a continuous murmur over the lungs and persistent cyanosis. The continuous murmur is not unlike that produced by a patent ductus arteriosus; it varies considerably with respiration, and is best heard when the breath is held at the end of inspiration. The intensity of the murmur is inversely proportional to the intensity of the cyanosis. When cyanosis is minimal, the murmur is loud and is usually widely transmitted throughout the lungs both front and back. The murmur may be audible only on one side. When it is maximal
over the pulmonary area, the malformation may be mistaken for that of a patent ductus arteriosus. In contrast to the murmur of a patent ductus arteriosus, this murmur seldom produces a thrill. When a thrill is present, it is widely transmitted and is usually readily palpable in the axilla or over the back.

In most instances the murmur is not very loud and cyanosis is moderately severe. Under such circumstances the murmur is frequently confined to a relatively small area, front, back or in the axilla. The location of the murmur depends upon the location and course of the vessels of the collateral circulation. The intensity of the murmur varies with the volume of blood coursing through these vessels. When cyanosis is intense and the pulmonary blood flow is minimal, there may be no murmur.

X-ray and fluoroscopic examination are frequently of aid in diagnosis. The heart is slightly to moderately enlarged and there is absence of fullness of the pulmonary conus. The large truncus arteriosus arches either to the right or to the left at an abnormally high level. The shadows cast by the pulmonary arteries are usually replaced by fine vascular markings which radiate from the hilar regions. In addition, delineation of the esophagus with barium in one or both oblique views may reveal the presence of retroesophageal vessels; in rare instances the esophagus is caught between vessels of the collateral circulation and displaced in a bizarre manner.

When the pulmonary blood flow is relatively adequate, there is no need for operation and no benefit can be derived therefrom. When the pulmonary blood flow is markedly reduced, these patients may be greatly helped by a Blalock-Taussig operation, provided there is a rudimentary pulmonary artery which is capable of conducting blood to the lungs. Those patients in whom the operation has been successfully performed have been greatly benefited. The over-all mortality rate, however, has been 25 per cent.

Pulmonary Hypertension and Persistent Patency of the Ductus Arteriosus (Reversed Ductus)

The occurrence of pulmonary hypertension combined with persistent patency of the ductus arteriosus causes a characteristic clinical syndrome.\textsuperscript{22, 23} The condition is frequently associated with malformations of the left side of the heart such as underdevelopment of the left ventricle, mitral stenosis, marked hypoplasia of the ascending aorta or extreme coarctation of the aorta. Whenever the pressure in the pulmonary artery is higher than that in the descending aorta, blood will flow from the pulmonary artery to the descending aorta. Consequently the feet will be more cyanotic than the hands. Frequently some blood from the pulmonary artery is forced back into the ascending aorta. Under such circumstances the left hand will be more cyanotic than the right.

The malformation places a strain on the right side of the heart. The electrocardiogram shows right axis deviation and evidence of right ventricular hypertrophy. The pulmonary valves frequently become insufficient as evidenced by a high-pitched, blowing diastolic murmur along the left sternal border. The difference in cyanosis between the upper and lower extremities offers the clue to the diagnosis. This can be confirmed by the simultaneous determination of oxygen content in the right brachial and femoral arteries. The oxygen content of the sample of blood from the right brachial artery will be normal and that from the femoral artery will be virtually the same as that of the pulmonary artery. Upon cardiac catheterization the pressure in the pulmonary artery will be elevated and it is usually possible to catheterize the descending aorta through the ductus arteriosus. Angiocardiogram will show simultaneous visualization of the descending aorta and the branches of the pulmonary artery.

Ligation of the ductus is generally contraindicated because of the extreme pulmonary hypertension and the left-sided heart lesion.

Dextrocardia without Situs Inversus and Situs Inversus with Levocardia

Dextrocardia without situs inversus and situs inversus with levocardia are usually extremely complicated malformations. A single ventricle is relatively common with dextrocardia. Anomalies of venous return are common when situs inversus is combined with levocar-
Dia. Although the malformation is seldom as simple as it appears to be, patients with these anomalies who show intense cyanosis, clear lung fields, and no hilar pulsations, may be helped by surgery.

III: General Rules in Regard to Management of Patients with Malformations of the Heart

1. Do not give a hopeless prognosis. Most of these children do better than the doctor anticipates. A poor prognosis not only increases the mother's anxiety but is likely to create a serious behavior problem in the child. Generally it is wise to tell the child he has a heart murmur but not to make an issue of it.

2. Allow the child to lead as normal and as active a life as is humanly possible. We all gain strength by using it. Moreover, almost every child with a malformation of the heart limits his own activity; there is seldom need for the doctor or for the parent to do so.

3. Urge the parents not to spoil the child. Warn the parents that the child may have to ask favors from his friends. The less the child is spoiled, and the better he is liked by his fellow playmates, the happier his life will be.

4. Immunization should be given at the usual time.

5. Prophylactic penicillin or some other antibiotic should always be given prior to dental extraction or tonsillectomy. The dosage recommended by the Council on Rheumatic Fever of the American Heart Association in 1952 is as follows: 400,000 units of crystalline penicillin G one-half hour prior to extraction; for the following three days the patient should receive either a daily dose of 400,000 units of slow-acting penicillin intramuscularly, or 400,000 units of oral penicillin three times daily one-half hour before meals.

6. Education is important. Whenever possible send the child to a regular school.

7. Death may result from anoxemia or occasionally from cardiac failure, subacute bacterial endocarditis, or brain abscess; but the usual childhood diseases or the ordinary intercurrent illnesses are generally well tolerated. Common colds and even pneumonia are seldom fatal.

8. Subacute bacterial endocarditis is now a curable disease. At the present time eight weeks of intensive antibiotic therapy is recommended. Whenever possible, the diagnosis should be established before treatment is started.

9. German measles in the first trimester of pregnancy is associated with a high incidence of congenital malformations of the heart, congenital cataracts and microcephaly in the offspring. The cardiac abnormality is the least serious of these three complications. The resultant malformation is frequently a patent ductus arteriosus. Microcephaly is a terrible tragedy. The earlier the infection occurs, the greater is the risk of injury to the fetus. If the infection occurs during the first month of pregnancy, the chance of a defective offspring is nearly 100 per cent. After the third month, German measles seldom if ever injures the fetus.

10. The incidence of malformations of the heart in siblings is so low that subsequent pregnancies can be recommended.

11. Women with malformations of the heart usually tolerate pregnancy better than is anticipated. Furthermore, the number of malformations amenable to surgery is constantly increasing; therefore only under exceptional circumstances is sterilization justified.

Special Considerations for the Cyanotic Child

1. An adequate fluid intake is important in order to lessen the danger of cerebral thrombosis. An infant should receive 1000 cc. of fluid per day; a child of 2 to 8 years, 1500 cc.; a child 8 to 12 years of age, 1500 to 2000 cc. Children over 12 years should receive a minimum of 2000 cc. Many young adults take 3000 cc. or even 4000 cc. of fluid per day. No patient with a polycythemia should go more than 12 hours without fluid. In case of vomiting, diarrhea, or excessive heat, the fluid intake should be increased.

2. The treatment of paroxysmal dyspnea: (a) Place the infant in the knee-chest position. This may be sufficient to end the attack. (b) Morphine sulfate (1 mg. per 10 pounds of body weight) is nearly specific for relief of paroxysmal dyspnea. (c) Inhalation of oxygen may help.

3. The treatment of the acute attacks of
dyspnea and cyanosis which occur in infants with a vascular ring may be alleviated by hyperextension of the head and neck.

4. Anoxemia causes retardation in growth and development, but does not cause mental retardation. The converse is also true: improvement of the circulation to the brain does not lessen mental retardation. Furthermore, a physically strong, mentally retarded child may get into far more trouble in this world than a mentally retarded child who is severely physically incapacitated.

5. In the evaluation of the advisability of early operation for a cyanotic infant, the degree of oxygen unsaturation of the arterial blood is far more important than is the intensity of the murmur. In most instances a malformation with a loud murmur carries a better prognosis than one in which there is no murmur. Indeed, the absence of a murmur is generally indicative of an extremely serious malformation. Under such circumstances if the lung fields are excessively clear, early operation is usually indicated.

6. Almost any anesthesia is well tolerated by a cyanotic patient who suffers from reduction of pulmonary blood flow provided he receives supplementary oxygen throughout the time of the anesthesia.

In conclusion, much can be done for the child with a malformed heart. An ever increasing number of malformations of the heart can be helped by surgery. Even if it is not now possible to help the child, it may yet be possible within the life span of the patient. Therefore, it is wrong to treat the child as if he were inevitably going to be a cardiac invalid throughout his life or die at an early age. Indeed, the child should be surrounded by a healthy, happy atmosphere, and the parents reminded that many persons have lived useful and significant lives without perfect health and strength.

REFERENCES


22 Damman, J. F., Berthrong, M., and Bing, R. J.: Reverse ductus: A presentation of the syndrome of patency of the ductus arteriosus, with pulmonary hypertension and a shunting of blood flow from pulmonary artery to aorta. (In preparation.)

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