CONGENITAL HEART DISEASE AS IT IS ENCOUNTERED IN THE ADULT PATIENT

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INTRODUCTION

Some understanding of congenital heart disease and its varied presentations is necessary for anyone expected to care for adult patients with heart disease. Malposition of the heart, cyanosis, bacterial endocarditis, arrhythmias, pulmonary hypertension, left and/or right ventricular failure, recurrent bacterial pneumonias, heart block, syncope, angina, etc may all be problems in the adult that stem from the patient having underlying congenital heart disease. Thus, some knowledge of those congenital heart problems that one is likely to encounter in adult cardiology seems necessary and this presentation will be devoted to that end. Table I represents a clinical classification of congenital heart disease which has previously been developed by Perloff to provide an overview of congenital heart disease.

TABLE I

Classification of Congenital Heart Disease

(from Perloff, J.K. The Clinical Recognition of Congenital Heart Disease, W.B. Saunders Company, Philadelphia, 1970 and modified 1977)

General

Innocent or normal murmurs

Congenital complete heart block

Congenitally corrected transposition of the great vessels

Congenital positional anomalies of the heart -- The cardiac malpositions

Acyanotic without a Shunt

Malformations originating in the left heart

1. Aortic stenosis
   a. Valvular
   b. Discrete subvalvular
   c. Muscular subvalvular
   d. Supravalvular
2. Coarctation of the aorta
3. Congenital mitral incompetence
   a. Endocardial cushion defect
   b. Congenitally corrected transposition of the great vessels

c. Primary endocardial fibroelastosis
d. Anomalous origin of the left coronary artery from the pulmonary artery
e. Miscellaneous (double orifice mitral valve, congenital perforations, accessory commissure with anomalous chordal insertion, congenitally short or absent chordae, cleft posterior leaflet, Marfan's syndrome, etc.)

4. Primary endocardial fibroelastosis
5. Congenital obstruction to left atrial flow
   a. Cor triatriatum
   b. Mitral stenosis
   c. Pulmonary vein stenosis
6. Congenital aortic incompetence

Malformations originating in the right heart or pulmonary artery

1. Pulmonic stenosis
   a. Valvular
   b. Infundibular
   c. Supravalvular (stenosis of the pulmonary artery and its branches)
   d. Subinfundibular
2. Idiopathic dilatation of the pulmonary artery
3. Congenital pulmonary valve incompetence
4. Primary pulmonary hypertension
5. Ebstein's anomaly of the tricuspid valve
6. Hypoplastic right ventricle

Acyanotic with a Shunt (Left to Right)

Shunt at atrial level

1. Atrial septal defect (isolated)
   a. Ostium secundum
   b. Ostium primum
   c. Sinus venosus

2. Atrial septal defect with mild pulmonic stenosis
3. Total anomalous pulmonary venous connection with low pulmonary vascular resistance
4. Partial anomalous pulmonary venous connection with intact atrial septum
5. Atrial septal defect with mitral stenosis (Lutembacher's syndrome)

Shunt at ventricular level

1. Ventricular septal defect (isolated)
   a. Infracristal
   b. Supracristal
   c. Muscular
   d. Endocardial cushion location
2. Ventricular septal defect with mild pulmonic stenosis (acyanotic Fallot's tetralogy)
3. Ventricular septal defect with right ventricular origin of both great vessels
4. Ventricular septal defect with congenitally corrected transposition of the great vessels
5. Ventricular septal defect with aortic incompetence
6. Ventricular septal defect with left ventricular to right atrial shunt
7. Ventricular septal defect with complete interruption of the aortic arch

Shunts between aortic root and right heart

1. Coronary arteriovenous fistula
2. Ruptured sinus of Valsalva aneurysm

Shunt at aorticopulmonary level

1. Patent ductus arteriosus
2. Aorticopulmonary septal defect
3. Anomalous origin of the left coronary artery from the pulmonary artery
4. Truncus arteriosus with large pulmonary arteries and low pulmonary vascular resistance

Shunts at more than one level

1. Complete endocardial cushion defect (complete persistent common artrioventricular canal)
2. Ventricular septal defect with patent ductus arteriosus
3. Ventricular septal defect with atrial septal defect

Cyanotic with a Shunt (Right to Left)

Normal or decreased pulmonary blood flow

A. Normal or low pulmonary arterial pressure
   1. Dominant right ventricle
      a. Pulmonary stenosis or atresia with ventricular septal defect and right to left shunt (cyanotic Fallot's tetralogy)
      b. Pulmonic stenosis with right to left interatrial shunt
      c. Complete transposition of the great vessels with severe pulmonic stenosis and large ventricular septal defect
      d. Pulmonic stenosis with right ventricular origin of both great vessels
      e. Pulmonic stenosis with single ventricle and inversion of the infundibulum (electrical dominance)
      f. Pulmonary artresia with intact ventricular septum and dilated right ventricle
      g. Truncus arteriosus with hypoplastic or absent pulmonary arteries
   2. Dominant left ventricle
      a. Tricuspid atresia
      b. Ebstein's anomaly with right to left interatrial shunt (mechanical dominance)
      c. Pulmonary atresia with intact ventricular septum and diminutive right ventricle
      d. Congenital vena caval to left atrial communication
      e. Single ventricle with pulmonic stenosis and noninversion of the infundibulum
      f. Large pulmonary arteriovenous fistula in infancy

3. Normal or nearly normal ventricles
   a. Pulmonary arteriovenous fistula
   b. Congenital vena caval to left atrial communication

B. Elevated pulmonary arterial pressure (pulmonary hypertension)
   1. Ventricular septal defect with reversed shunt (Eisenmenger's complex)
   2. Patent ductus arteriosus or aorticopulmonary septal defect with reversed shunt
   3. Atrial septal defect with reversed shunt
   4. Right ventricular origin of both great vessels with high pulmonary vascular resistance
   5. Hypoplastic left heart (aortic atresia, mitral atresia, complete interruption of the aortic arch)
   6. Complete transposition of the great vessels with high pulmonary vascular resistance
   7. Single ventricle with high pulmonary vascular resistance
   8. Total anomalous pulmonary venous connection with pulmonary venous obstruction

Increased pulmonary blood flow

1. Complete transposition of the great vessels
2. The Taussig-Bing anomaly (right ventricular origin of both great vessels with supracristal ventricular septal defect or right ventricular aorta with biventricular pulmonary trunk)
3. Truncus arteriosus with large pulmonary arteries
4. Total anomalous pulmonary venous connection
5. Single ventricle with low pulmonary resistance and no pulmonic stenosis
6. Common atrium
7. Fallot's tetralogy with pulmonary atresia and increased bronchial arterial flow
8. Tricuspid atresia with large ventricular septal defect and no pulmonic stenosis

This clinical classification allows one to ask and answer certain questions about a patient with potential congenital heart disease. Included in these questions might be: 1) Is the patient cyanotic? 2) Is pulmonary arterial blood flow normal, diminished or increased? 3) Does the abnormality originate in the left or right heart? 4) Is pulmonary hypertension present? Table II allows a somewhat simpler and working classification of congenital heart disease that might be utilized as a directional guide in answering the questions posed above.

TABLE II

A Simplified Classification of Congenital Heart Disease

(from Perloff, J.K. The Clinical Recognition of Congenital Heart Disease, W.B. Saunders Company, Philadelphia, 1970 and modified)

General

Innocent or normal murmurs
Congenital complete heart block
Corrected transposition of the great vessels
Cardiac malpositions

Acyanotic without a shunt

Malformations originating in the left heart
Malformations originating in the right heart

Acyanotic with a shunt (left to right)

Shunt at atrial level
Shunt at ventricular level
Shunt between aortic root and right heart
Shunt between aorta and pulmonary artery
Shunts at more than one level

Cyanotic (right to left shunt)

Normal or decreased pulmonary blood flow

A. Normal or low pulmonary arterial pressure
   1. Dominant right ventricle
   2. Dominant left ventricle
   3. Normal or nearly normal ventricles

B. Pulmonary hypertension

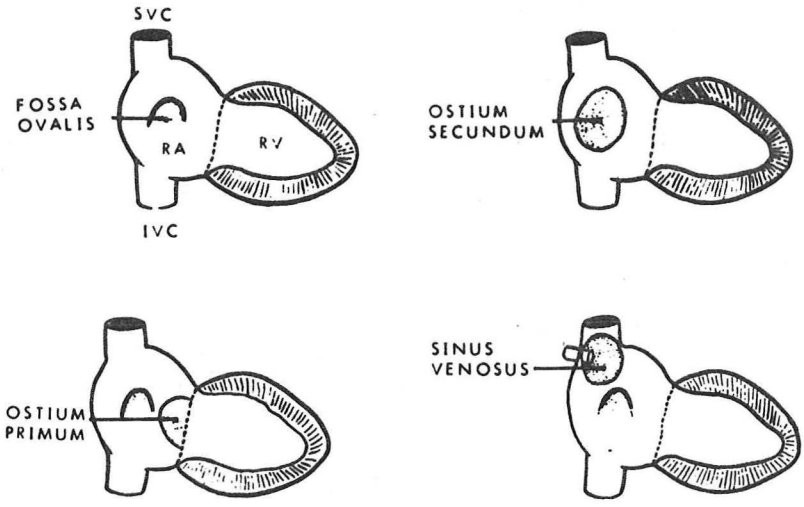
Increased pulmonary blood flow

1. Complete transposition
2. Truncus arteriosus (large pulmonary arteries)
3. Taussig-Bing anomaly

The ability to ask the appropriate clincal questions and subsequently the correct interpretation of the physical examination, the electrocardiogram, the chest roentgenogram (and now both the echocardiogram and in some instances radionuclide blood pool scintigrams) allow one to direct attention and further diagnostic and therapeutic efforts appropriately in almost every patient. We will now proceed with a more detailed evaluation of some of the more important types of congenital heart disease that one is likely to encounter in adult patients.

Atrial Septal Defects

Defects in the atrial septum may occur in the locations shown in Figure 1.



Schematic illustration of location of the normal fossa ovalis and of an ostium secundum, ostium primum and sinus venosus atrial septal defect. In the sinus venosus defect note the relationship of the right superior pulmonary vein to the defect. (Modified after Bedford et al.: Amer. J. Cardiol. 6:568, 1960, and Arnfred, E.: J. Cardiov. Surg. 7:349, 1966.)

Figure 1

Atrial septal defects may occur 1) in the region of the fossa ovalis (ostium secundum); 2) inferior to the fossa, i.e. between the fossa and the inferior vena cava; 3) in the upper part of the atrial septum (sinus venosus); 4) in the lower part of the septum (ostium primum); and 5) in the position normally occupied by the coronary sinus. The commonest location for an atrial septal defect is in the region of the fossa ovalis; this and is referred to as an ostium secundum defect (Fig. 1). Occasionally the defect lies between the fossa and the inferior vena cava with the lower margin bordering the caval orifice. Rarely, the defects are located superior to the fossa just beneath the entrance to the superior vena cava; these defects are sometimes called "sinus venosus defects". Ostium primum defects are located in the lower part of the septum, occupying the site they would ordinarily occupy if they occurred as part of an endocardial cushion malformation (Fig. 1). While ostium primum defects may occur as isolated abnormalities, they are more commonly found coexisting with other features of the endocardial cushion defect, especially cleft mitral valve and mitral incompetence. Finally, there is a rare type of atrial septal defect that is found in the position normally occupied by the coronary sinus. Defects in this location may occur as part of a developmental complex consisting of atrial septal defect, absent coronary sinus and drainage of a left superior cava into the left atrium. Ordinarily an atrial septal defect is found in only one of the above described locations, but occasionally separate defects exist at more than one site. A patent foramen ovale is a type of interatrial communication, but it should not be considered an atrial septal defect. The patent foramen is distinguished from a true atrial septal defect in which septal tissue is absent. Of course, under ordinary circumstances the foramen ovale and its valve permit right to left flow in utero, but the foramen closes prior to or shortly after birth. Under certain conditions, a left to right shunt may occur across an otherwise essentially normal foramen ovale; the usual explanation for this phenomenon is a high left atrial pressure with dilatation of the left atrium resulting in stretching of the foramen making its ostium incompetent. Therefore, any condition that results in a distended left atrium with increased pressure could theoretically produce a left to right shunt through a patent foramen ovale. Such conditions as mitral insufficiency, mitral atresia, and acquired mitral stenosis have been associated with a patent foramen ovale and a left to right shunt that is ordinarily quantitatively small.

Physiologic Consequences of Atrial Septal Defects

The physiologic consequences of an atrial septal defect depend upon its size, the magnitude of the left to right shunt and the subsequent behaviour of the pulmonary vascular bed. It should be emphasized that often the pressure difference between the atria does not adequately account for the left to right shunt seen in this condition, but instead reduced compliance (increased stiffness) in the left atrium initially is believed to be primarily responsible for the left to right shunt. A large atrial septal defect often results in free communication between the atria, but little or no pressure gradient and a small atrial septal

defect may result in a larger pressure differential but a smaller shunt. *Compliance alterations in the two ventricles as well as in the left atria appear to be the major determinant of the size and direction of the shunt.* Flow occurs in the direction of lesser resistance with atrial septal defects. The right ventricle is normally more distensible than the left so left atrial blood flows across the defect establishing a left to right shunt. A small defect permits only a small shunt, but with a large defect the magnitude of the left to right shunt depends on compliance characteristics in the right ventricle. When the right ventricle fails and its compliance decreases (and stiffness increases), the left to right shunt decreases in magnitude and may be replaced by a right to left shunt across the atrial septal defect. One should recall that in the fetus the direction of interatrial flow is from right to left (whether as a consequence of flow through a patent foramen ovale or through a true atrial septal defect). This is accounted for by the high pulmonary vascular resistance in the fetal pulmonary bed and the relatively thick walled right ventricle. At birth there is little or no shunt in either direction because the compliance characteristics of the two ventricles are essentially similar. During infancy, the pulmonary arteries involute and the relatively thick walled neonatal right ventricle become thinner. In individuals with atrial septal defects this alters physiological conditions so that left to right shunting across an atrial septal defect occurs. A sizable left to right shunt at the atrial level can then exist with normal pulmonary artery pressure and vascular resistance. The development of pulmonary hypertension in patients with atrial septal defects is then ordinarily the consequence of late intimal pulmonary arteriolar vascular alterations. *This emphasizes the point that the development of pulmonary hypertension in individuals with large left to right shunts is relatively rare during childhood and reaches its peak incidence of about 15% during young adulthood (i.e. at approximately the age of 18 or 20 years).* This also emphasizes the fact that if normal pulmonary artery pressure and vascular resistance are present in a young child with a large atrial septal defect, one generally need not be concerned that significant pulmonary hypertension will subsenquently rapidly develop and that the atrial septal defect need be closed in the early years of life to prevent this complication. Instead, the defect can be closed during young adulthood electively if the shunt is of large enough size to warrant this approach.

As time passes and individuals with atrial septal defects age, the shunt pattern may follow one of two courses. The right ventricle may become progressively more stiff because of ventricular failure or the late development of elevated pulmonary artery pressure and vascular resistance. This will reduce the magnitude of the left to right shunt and result in the possible development of right to left shunting. The second possible approach (and a more common one) is that the adult patient develops an increase in left ventricular stiffness resulting in an increase in the left to right shunt and an even greater volume load in the right heart. Decreases in left ventricular compliance (or increases in stiffness) may be the result of acquired left ventricular diseases such as systemic hypertension, coronary artery disease, or alterations

of aging. This increased left to right shunt may, of course, also result in the later development of significant pulmonary artery hypertension and ultimately lead to right ventricular failure and eventually a balanced or reversed shunt.

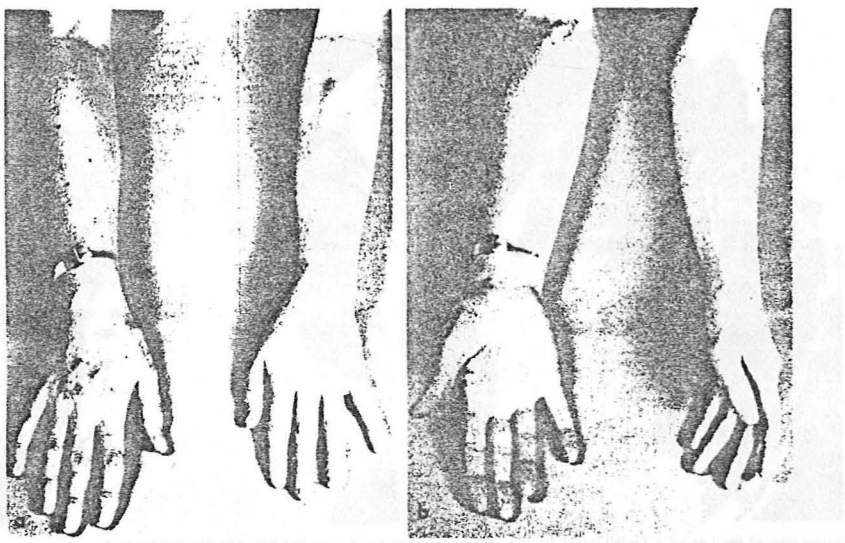
Pulmonary Venous Drainage in Individuals with Atrial Septal Defects

It has long been recognized that anomalies of pulmonary venous drainage may coexist with atrial septal defects. One or more pulmonary veins may enter either the right atrium or a systemic vein directly. This circumstance has been designated "anomalous pulmonary venous drainage" to emphasize that the pulmonary vein(s) drain into the right atrium or its tributaries rather than as expected into the left atrium. Partial anomalous venous return typically involves the veins from the right lung. The anomalous connections may occur through drainage into the superior vena cava or into the right atrium. Occasionally connection is with the inferior vena cava; in this circumstance abnormalities of the right lung often coexist. These abnormalities may include hypoplasia of the right lung with displacement of the heart and abnormal systemic arterial supply to the right and/or abnormalities of pulmonary parenchyma and bronchi. The consequences of anomalous pulmonary venous drainage coexisting with atrial septal defects depend on the number of veins involved and the magnitude of this additional left to right shunt. *This possibility should always be considered and specifically searched for in the evaluation of patients with atrial septal defects.*

Natural History of Atrial Septal Defects

The most common type of atrial septal defect is an ostium secundum one. This defect develops more often in females than males with the sex ratio varying from 1.5 to 3:1. These defects may be familial and may recur through a number of generations. The mode of inheritance is somewhat uncertain and is variously described as being autosomal dominant or recessive depending upon the series selected. Autosomal dominant inheritance with familial occurrence is recognized in individuals with atrial septal defects and the Holt-Oram syndrome (Fig. 2). In this syndrome the thumb is hypoplastic and often has an accessory phalanx that may give it a crooked appearance. The thumb also lies in the same plane as the other digits so that apposition may be difficult. In some individuals the thumb is rudimentary or absent. Some individuals also have absence of an arm or underdevelopment of a portion of the arm. Hypoplastic abnormalities may extend to the radius making full supination of the hand difficult.

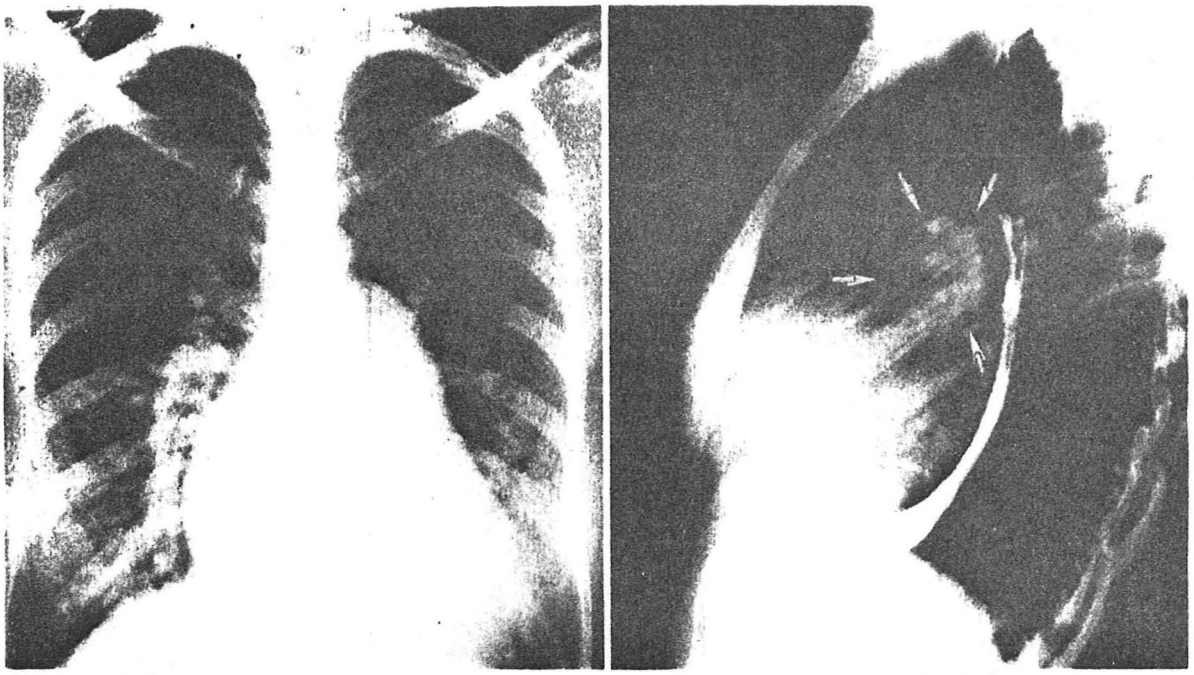
Atrial septal defects may go undetected for years. This is primarily because they may be relatively subtle from a physical examination point of view and they may produce no problems for many years. It is not uncommon for an atrial septal defect to be initially suspected on the basis of a routine chest x-ray that is obtained (Figs. 3-5).



Photographs of the hands of a 34 year old woman with the Holt-Oram syndrome and a large ostium secundum atrial septal defect. *a*, The left thumb is moderately hypoplastic and the left arm is shorter than the right. *b*, When the palms are turned up, hypoplasia of the radius prevents full supination of the left hand, and the "crooked" appearance of the small thumb becomes apparent. Note also that the fingertips of the upturned right hand are erythematous as a result of a small right to left shunt (arterial oxygen saturation 90 per cent).

Figure 2

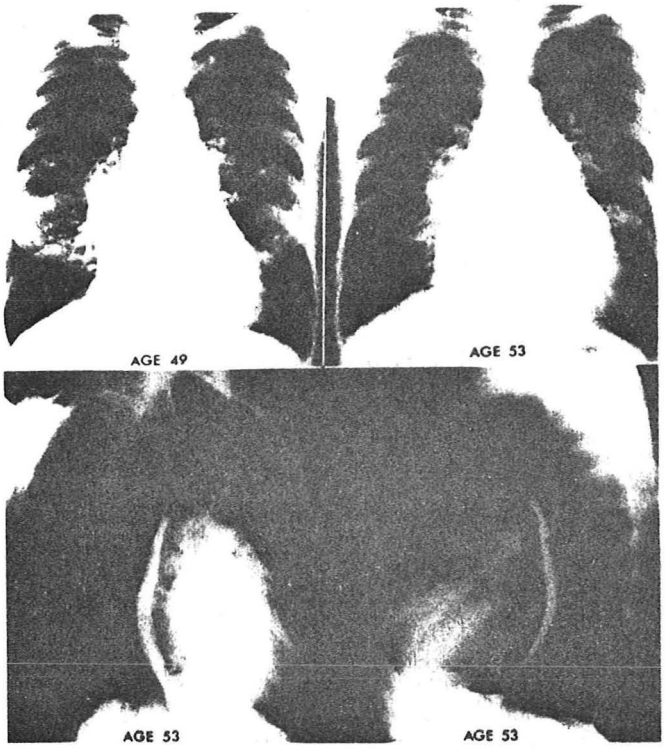
(Modified from Perloff, J.K. The Clinical Recognition of Congenital Heart Disease, W.B. Saunders Company, Philadelphia, 1970.)



X-rays from a 31 year old woman with an ostium secundum atrial septal defect, a 3.2 to 1 left to right shunt and normal pulmonary arterial pressure. The posteroanterior view shows increased vascularity extending to the periphery of the lung fields. The pulmonary trunk and its right branch are strikingly enlarged, but the ascending aorta is inconspicuous. There is a prominent right atrial shadow to the right of the vertebral column, and a dilated right ventricle occupies the cardiac apex. The lateral projection shows prominent anterior bowing of the upper third of the sternum. The arrows outline the dilated right pulmonary artery.

Figure 3

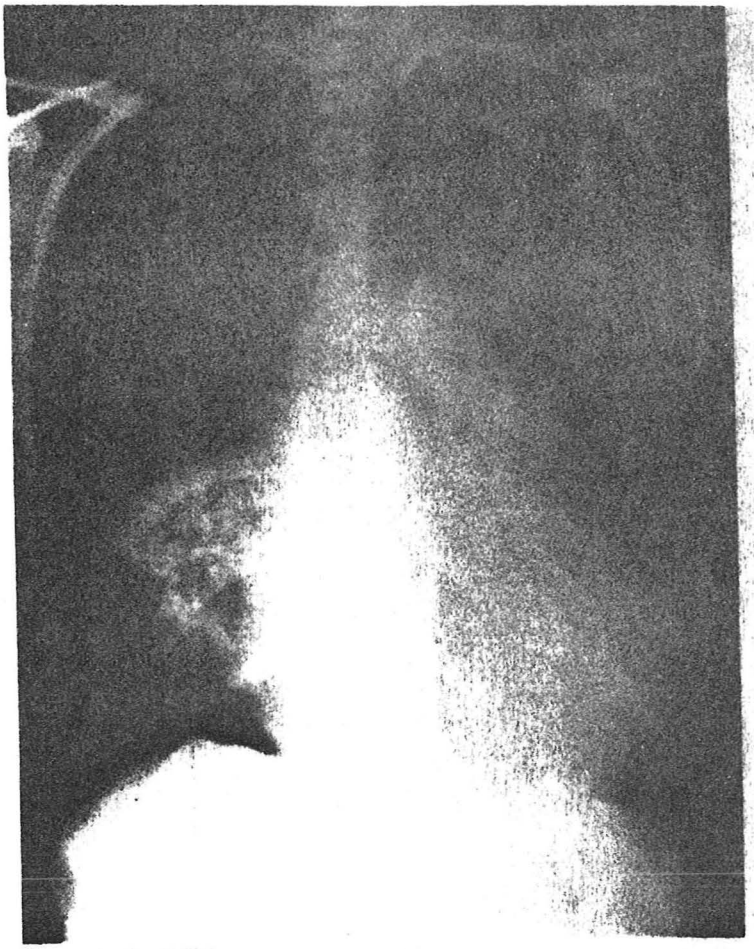
(Modified from Perloff, J.K. The Clinical Recognition of Congenital Heart Disease, W.B. Saunders Company, Philadelphia, 1970.)



X-rays from a woman with an ostium secundum atrial septal defect before and after the onset of atrial fibrillation and cardiac failure. At age 49 the patient was relatively well and her rhythm was normal sinus; the x-ray (upper left) shows increased pulmonary vascularity, slight prominence of the pulmonary trunk and its right branch, and a moderate increase in the right atrial shadow. At age 53 she was referred with a diagnosis of rheumatic mitral valve disease after the onset of atrial fibrillation and progressive clinical disability. The posteroanterior x-ray (upper right) shows a considerable increase in cardiac dimensions; the pulmonary trunk, right atrium and right ventricle are much larger than before and the right oblique and lateral films (below) show retrodisplacement of the esophagus by a dilated left atrium. The left to right shunt at this time was 2.2 to 1 and the pulmonary arterial systolic pressure 40 mm. Hg.

Figure 4

(Modified from Perloff, J.K. The Clinical Recognition of Congenital Heart Disease, W.B. Saunders Company, Philadelphia, 1970.)



X-ray from a mildly cyanotic 64 year old woman with a pulmonary hypertensive ostium secundum atrial septal defect and a small persistent left to right shunt. The lung fields are clear although the film is overpenetrated. The pulmonary trunk and its right branch are aneurysmal and the right branch contains calcium. In addition, the large left branch can be seen behind the pulmonary trunk. The ascending aorta is inconspicuous. A right atrial shadow is seen to the right of the vertebral column and the dilated right ventricle occupies the cardiac apex.

Figure 5

(Modified from Perloff, J.K. The Clinical Recognition of Congenital Heart Disease, W.B. Saunders Company, Philadelphia, 1970.)

Survival to advanced ages is not at all uncommon and some individuals may be in their eighth or ninth decade of life when the atrial septal defect is discovered. Survival, of course, depends in part on whether significant pulmonary hypertension develops during adulthood; the development of this complication markedly shortens longevity. Death when it occurs may be related to the underlying atrial septal defect and congestive heart failure is the most common expression of an important complication of atrial septal defects. Death is also known to be associated with the development of pulmonary arterial thrombosis, recurrent pulmonary infections, brain abcess and even rupture of the pulmonary artery. Complications of the atrial septal defect often arise in patients with left to right shunts of 2:1 or greater and almost all patients surviving to the sixth decade with left to right shunts of this magnitude are symptomatic. When complaints develop, effort dyspnea and fatigue are the most common. A history of recurrent pulmonary infections is not unusual in such individuals and occasional patients die of pneumonia. *Bacterial endocarditis is very rare in individuals with otherwise uncomplicated atrial septal defect.* The rarity of this complication is believed to be due to the absence of jet lesions or turbulence because of the low velocity of flow across the interatrial communication. An interesting additional problem related to atrial septal defects with left to right shunts of 2:1 or greater is that adults with the abnormality may have atrial arrhythmias including fibrillation, flutter and/or paroxysmal atrial tachycardia. These arrhythmias may be annoying and also may contribute to more rapid development of congestive heart failure in the aging patient.

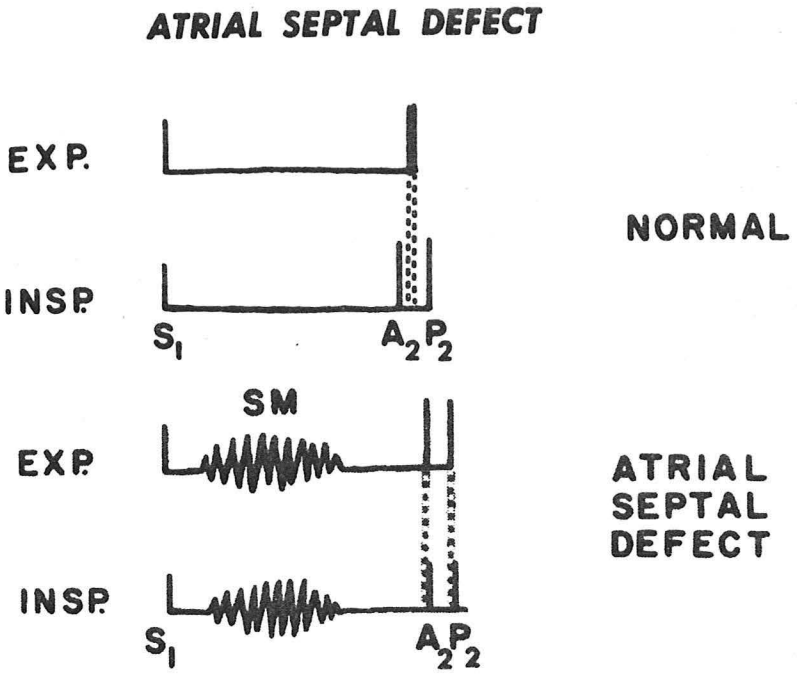
The development of pulmonary hypertension and increased pulmonary vascular resistance has already been mentioned as a significant complication ordinarily developing after the age of 20 years in individuals with atrial septal defects and left to right shunts of 2:1 or greater. However, the history of patients with these abnormalities should take into account the altitude at which the patient was born. A higher incidence and earlier onset of pulmonary hypertension has been described in individuals with atrial septal defects born at high altitude. This presumably reflects the influence of high altitude per se on pulmonary artery pressure and vascular resistance. It has long been recognized that the decreased partial pressure of oxygen at relatively high altitudes results in some degree of pulmonary vasoconstriction and coupled with left to right shunts of the magnitude already described may produce transient (and possibly permanent although this matter is not totally clear presently) pulmonary hypertension that occurs earlier and to a more severe degree than in similar individuals born at sea level. Complaints related to the development of pulmonary hypertension may include dyspnea and fatigue; cyanosis may develop either at rest or with effort. Chest pain resembling angina may occur and sometimes this is the major complaint. Hemoptysis may also be noted. Symmetrical cyanosis of the hands and feet and clubbing occur when pulmonary hypertension develops, right ventricular failure ensues and the shunt becomes right to left. Exercise often serves to accentuate the cyanosis so that appearance should be assessed both at rest and after effort.

Physical Examination of the Patient with an Atrial Septal Defect

Examination of the jugular venous pulse demonstrates that the A and V waves tend to be equal instead of the slight dominance of the A wave found in normal individuals. Once right ventricular failure develops, jugular venous pressure may or may not rise depending on whether adequate decompression of right atrial venous pressure is possible by right to left shunting. The development of pulmonary hypertension may result in an increased force of right atrial contraction and a dominant A wave which may reach impressive levels (although if the septal defect is large enough, a prominent A wave may not be visualized even when it should for physiological reasons be present). Tricuspid regurgitation may develop in the failing right ventricle and this leads to large V waves in the jugular venous pulse.

Palpation of the precordium identifies a prominent right ventricular impulse in patients with atrial septal defects and 2:1 left to right shunts or greater. Characteristically, the right ventricular impulse is forceful and of short duration. This impulse is understandable since the left to right shunt distends the right ventricle during diastole and the chamber then contracts vigorously ordinarily against a relatively low resistance in the pulmonary vascular bed. The dialated pulmonary artery may also be recognized by detecting a pulmonary artery impulse in the second left intercostal space. *A systolic thrill in the second left intercostal space identifies a large shunt or coexisting pulmonic stenosis.* Once pulmonary hypertension develops, the right ventricular thrusting impulse may be more sustained and less dynamic. Pre-systolic right venticular distention may be palpable; this is the consequence of increased force of right atrial contraction. With the development of pulmonary arterial hypertension, the palpable pulmonary artery becomes even more striking and one should also be able to feel a grossly exaggerated pulmonic valvular closure sound.

The auscultatory signs of atrial septal defect without pulmonary hypertension include some of the following. The first heart sound is often "split" and the second component of the split first heart sound relatively pronounced. If the pulmonary artery is dilated, an ejection click may also be heard. The typical murmur of atrial septal defect begins immediately after the first heart sound and is ejection in quality reaching a peak in mid-systole and ending well before the second heart sound (Fig. 6). Rapid ejection of the large right ventricular stroke volume into a dilated pulmonary artery accounts for this murmur. The atrial septal defect itself does not ordinarily produce a murmur. The systolic ejection murmur is soft enough that it is occasionally missed or attributed to being an "innocent murmur" and the possibility of an atrial septal defect not seriously considered. *When the murmur is loud exceeding grade 3, one often finds either associated pulmonic stenosis or an unusually large shunt.*

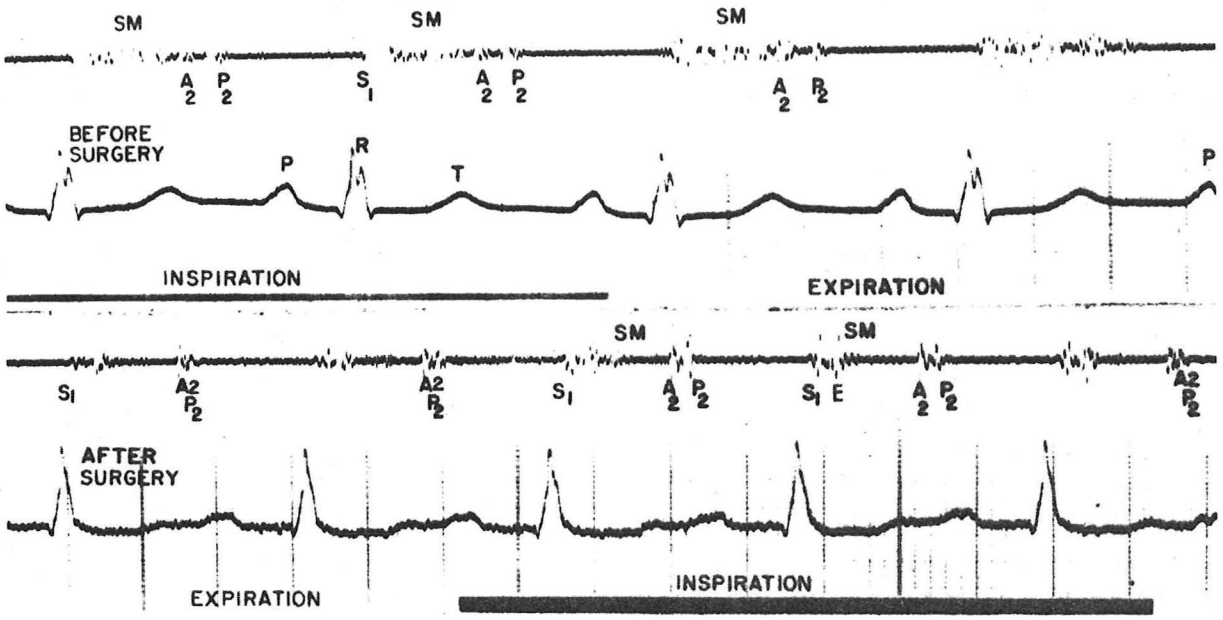


Schematic illustration of the normal respiratory behavior of the second heart sound and the behavior in uncomplicated atrial septal defect. Normal inspiratory splitting is due chiefly to a delay in pulmonary valve closure (P_2) and to a lesser extent to movement of aortic closure (A_2) in the opposite direction. In atrial septal defect the second sound is widely split even during expiration because P_2 is late. The split remains "fixed" during active breathing because both components of the second heart sound move equally and in the same direction or do not move at all. (Perloff, J. K.: Heart Bull. *18*:28, 1969. Copyrighted by The Medical Arts Publishing Foundation, 1603 Oakdale Street, Houston, Texas 77004.)

Figure 6

(Modified from Perloff, J.K. The Clinical Recognition of Congenital Heart Disease, W.B. Saunders Company, Philadelphia, 1970.)

One of the classical findings on physical examination of individuals with atrial septal defects is a "fixed splitting" of the second heart sound (Fig. 7). That is, the aortic and pulmonic components are widely



Phonocardiograms before and after surgery in a 17 year old girl with an ostium secundum atrial septal defect, a 2.6 to 1 left to right shunt and normal pulmonary arterial pressure. Prior to operation, there was a grade 3 pulmonic systolic murmur followed by wide, fixed splitting of the second heart sound. After closure of the atrial septal defect, the pulmonic systolic murmur became soft, and the second sound split normally. A pulmonic ejection sound (E) appears in the postoperative tracing. Note that there is PR interval prolongation in lead 2 of the electrocardiogram.

Figure 7

split during expiration and show little or no change in the degree of splitting with inspiration or with any other provocative maneuver. The wide splitting is caused by a delay in pulmonic valve closure itself a result of increased duration of right ventricular ejection. Since the right ventricle receives both shunted blood and the systemic venous return, its stroke volume is increased and its ejection time prolonged. In young children with atrial septal defects, splitting of the second heart sound may be normal (rather than fixed) since a relatively rapid rate of right ventricular ejection may partially offset the effect of a large stroke volume. The duration of diastole is another determinant of the degree of splitting of the second heart sound in individuals with atrial septal defects since the left to right shunt occurs primarily during cardiac diastole. As heart rate increases, diastole shortens and

the left to right shunt per beat decreases. This results in reduction in the right ventricular stroke volume and splitting narrows. Slowing of heart rate exerts exactly the opposite effect. With atrial fibrillation, the splitting normally tends to vary inversely with the duration of the preceding cardiac cycle. The patient's postion should also be considered before drawing conclusions concerning splitting of the second heart sound, especially in children. In the supine position a normal young adult (or child) may exhibit relatively wide splitting of the second heart sound which disappears as the upright position is assumed. Sitting or standing decreases systemic venous return and consequently reduces the stroke volume and ejection time of the right ventricle. Pulmonic valve closure occurs earlier and the correct impression of the normal respiratory behaviour of the second heart sound is made possible. Fixed splitting of the second heart sound which is a feature of some patients with atrial septal defects (but not all as already emphasized) is characterized not only by wide splitting, but also constant splitting with essentially no variation in the degree of splitting of the second heart sound during inspiration, expiration, sitting, standing, the Valsalva maneuver, etc. This fixed splitting occurs as emphasized above because of 1) the left to right shunt and 2) the increase in systemic venous return during inspiration in this setting. The degree of splitting remains fixed primarily because phasic changes in systemic venous return during respiration are associated with reciprocal changes in the volume of the left to right shunt, thus minimizing the respiratory differences in right and left ventricular filling. An inspiratory decline in left to right shunting has been demonstrated both in experimental animals and in human subjects with atrial septal defects. Thus, the respiratory sequence in individuals with atrial septal defects appears to be as follows: inspiration results in an increase in venous return so that right ventricular filling is either maintained or increased; the left to right shunt diminishes reciprocally so that left ventricular filling is maintained or increased by the same amount. Accordingly, the relative durations of right and left ventricular ejection remain constant and the degree of splitting remains unchanged.

Two types of diastolic murmurs occur in individuals with atrial septal defects with left to right shunt. One is the classical mid-diastolic murmur which results from the large volume of flow across the tricuspid valve; this murmur may be thought of as reflecting "relative tricuspid stenosis". The second type of diastolic murmur occurring in individuals with atrial septal defects is due to pulmonary valvular regurgitation, which may occur as a consequence of pulmonary arterial hypertension, *but occurs in some patients with a dilated pulmonary artery but without significant increases in pulmonary arterial pressure or vascular resistance.*

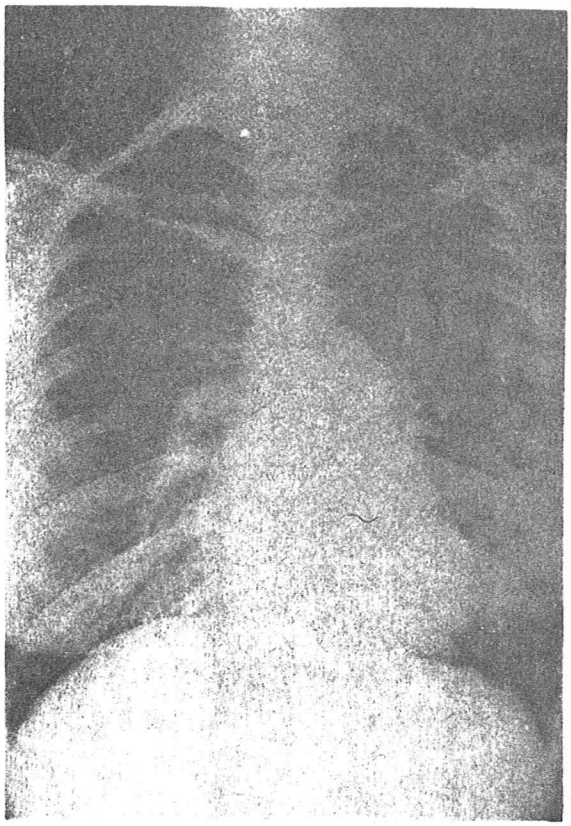
With the development of pulmonary hypertension the auscultatory findings change in patients with atrial septal defects. The second heart sound increases appreciably in intensity and particularly that component due to pulmonic valve closure. The splitting of the second

heart sound may become quite narrow and in fact the second sound may be single as pulmonary artery pressure and vascular resistance reach systemic arterial levels. The systolic murmur disappears as the left to right shunt becomes balanced or the shunt is reversed to a right to left one; the systolic ejection murmur may be replaced by a holosystolic one that increases with inspiration and reflects tricuspid regurgitation which identifies the presence of right ventricular failure. As noted above, a diastolic decrescendo murmur representing pulmonary valvular regurgitation and a consequence of pulmonary hypertension may also be noted in patients with severe increases in pulmonary artery pressure. Systolic ejection clicks are expected in patients with pulmonary arterial hypertension whatever the eitology and they may be noted in this circumstance. As the right ventricle fails, gallop sounds may be audible including both third and fourth heart sounds; these sounds may increase with inspiration allowing one to assign them to originating in the right ventricle.

The electrocardiogram in patients with atrial septal defects ordinarily demonstrates sinus rhythm during the first three decades of life, but later the incidence of atrial arrhythmias increases. Atrial fibrillation is the commonest rhythm distrubance in later adult years in individuals with this entity. During sinus rhythm, the P-R interval is ordinarily at the upper limits of normal, but sometimes is prolonged. Patients with atrial septal defect and the Holt-Oram syndrome often have conduction and rhythm disturbances irrespective of age. The Wolff-Parkinson-White electrocardiographic pattern has been reported in a few patients with atrial septal defects. Right bundle branch block occurs in a significant percentage of patients with otherwise uncomplicated atrial septal defects when the left to right shunt is 2:1 or greater. With the development of pulmonary arterial hypertension other electrocardiographic manifestations may develop including the development of ECG evidence of right ventricular hypertrophy, right bundle branch block, and right axis deviation. Patients with ostium primum atrial septal defects may be distinguished from septal defects in other locations by noting that most of these individuals have left axis deviation and right bundle branch block. Indeed, the combined presence of left axis deviation and right bundle branch block in a young patient with no other reason for this electrocardiographic pattern and with other findings consistent with atrial septal defect suggests ostium primum atrial septal defect.

Chest X-ray

As noted earlier, a chest roentgenogram, especially in adults, is often distinctive enough to suggest the diagnosis of atrial septal defect in its own right when the left to right shunt is 2:1 or greater (Figs. 3,4,5 & 8). Once pulmonary hypertension develops in patients with atrial septal defects the radiographic manifestations will be those of pulmonary hypertension and it becomes more difficult to distinguish precise etiologies. In the patient with an atrial septal defect with a large shunt but relatively normal pulmonary artery pressures, dramatic



X-ray from a 22 year old woman with an ostium secundum atrial septal defect, a 2.4 to 1 left to right shunt and normal pulmonary arterial pressure. The increased pulmonary vascularity extends to the periphery of the lung fields. The main pulmonary artery and its right and left branches are appreciably enlarged, but the ascending aorta is inconspicuous. There is a prominent right atrial shadow to the right of the vertebral column, and a dilated right ventricle occupies the cardiac apex.

Figure 8

enlargement of the proximal arteries occurs and marked increase in pulmonary vascularity may be detected and followed far out into both lung fields producing a pattern referred to as "shunt vascularity" (Fig. 8). As mentioned earlier, often the initial recognition of atrial septal defect is on the basis of a chest x-ray that is obtained and identifying the rather characteristic appearance of the proximal pulmonary artery, a particularly enlarged right pulmonary artery and evidence of shunt vascularity. The presence of anomalously connecting pulmonary veins may sometimes be suspected on the plain chest x-ray when these channels communicate with the inferior vena cava. These venous anomalies are often restricted to the right lower lobe, but occasionally the venous drainage from the entire right lung converges to form a large venous trunk that empties into the inferior vena cava. The abnormal vascular shadow that is produced may be recognized in a frontal chest film as a density to the right of the cardiac silhouette. Its density takes a downward course parallel to or behind the right side of the heart and may be crescent-shaped. The contour of this density may resemble a curved Turkish sword so that the x-ray picture has occasionally been referred to as "the scimitar sign".

With the development of pulmonary arterial hypertension, the conspicuous peripheral pulmonary vasculature is replaced by relatively oligemic lung fields. The proximal pulmonary arteries are huge, but they rapidly taper and rather than producing a pattern of peripheral "shunt vascularity" a relative oligemia of the peripheral lung fields devlops (Fig. 5).

The atrial silhouettes also deserve some comment. Right atrial enlargement is often seen as a rightward convexity in the posterior anterior projection of a plain chest film, but the left atrium in any view is often inconspicuous in individuals with atrial septal defects. *In acyanotic patients with large left to right shunts, the absence of left atrial enlargement and the presence of a conspicuous right atrial shadow suggests an intracardiac shunt at the atrial level.* Dilatation of the right ventricle is an expected roentgenographic feature of individuals with atrial septal defect. Left ventricular enlargement is not a roentgenographic feature of uncomplicated atrial septal defect (although it might well coexist in older individuals with other disease processes responsible for the LV enlargement).

Lutembacher's Syndrome

Lutembacher published the first complete account of atrial septal defect with mitral stenosis. This combination of lesions has since become known as Lutembacher's syndrome. Lutembacher originally proposed that the mitral stenosis was congenital in this syndrome, but more recently the term has been applied to acquired mitral stenosis; the latter is ordinarily the result of previous rheumatic carditis. I believe that it is appropriate today to accept a congenital defect in the atrial septum coexisting with either acquired or congenital mitral stenosis as representing Lutembacher's syndrome. Varying degrees of mitral incompetence may also be present, but this is not a feature of the basic syndrome.

When atrial septal defect and mitral stenosis coexist, each lesion modifies the hemodynamics and clinical expression of the other. Clinical signs and symptoms depend largerly upon the size of the atrial septal defect and the severity of mitral stenosis and the compliance characteristics of the pulmonary vascular bed and the right ventricle.

Let us briefly consider the physiological interaction of these two lesions. The left to right shunt in uncomplicated atrial septal defect is determined by the compliance characteristics of the right and left ventricle and the size of the septal defect. Mitral stenosis increases the resistance to the flow of blood from left atrium into left ventricle and consequently increases left to right shunting through an atrial septal defect. When a defect in the atrial seputm is large and the mitral stenosis severe, diastolic flow from left atrium into right heart is markedly augmented. When a small atrial septal defect occurs with severe mitral stenosis, the left to right shunt, although comparatively small, may become continuous since the pressure difference across the interatrial communication exists throughout the cardiac cycle.

An atrial septal defect constitutes an alternative exit for left atrial blood and if large enough, may effectively decompress a high pressure left atrium resulting from severe mitral stenosis. This may allow left and right atrial pressures to equalize and the severity of the gradient across the mitral valve to decrease, to disappear altogether or be present only during exercise. These hemodynamic benefits are real, but they are acquired at a price, i.e. reduced inflow into the left ventricle and a consequently reduced cardiac output. The hemodynamic principle that an interatrial communication might exert a favorable effect on the hemodynamics of mitral stenosis is not new. Indeed, in 1949 Bland and Sweet surgically treated mitral stenosis by anastomosing a right pulmonary vein to the azygous vein after which the left atrial pressure fell. In patients with pulmonary hypertension, atrial septal defects and mitral stenosis the right ventricle ultimately becomes stiffer (less compliant), the left to right shunt diminishes, more left atrial blood enters the left ventricle and the systemic output rises.

Lutembacher's syndrome occurs mostly in females; this is predictable since both atrial septal defects and rheumatic mitral stenosis are more common in females. *The presence of atrial fibrillation is more common than it is in isolated atrial septal defects and there is a risk of endocarditis since mitral stenosis coexists with the atrial septal defect and bacterial infection of the mitral valve is a distinct possibility in individuals with this entity.*

Physical Examination

Examination of the jugular venous pulse in individuals with Lutembacher's syndrome and moderately severe mitral stenosis and an important left to right shunt and sinus rhythm often reveals elevated mean jugular venous pressure and an increased jugular venous A wave even in individuals without gross right ventricular failure or pulmonary hypertension.

Physical examination further reveals a loud first heart sound, an opening snap (if the mitral valve is not heavily calcified) and a middiastolic murmur. One may, of course, also document the soft systolic ejection murmur previously mentioned as occurring in individuals with atrial septal defects and either a diastolic rumble (at the left sternal border) related to torrential tricuspid valve flow or a diastolic decrescendo murmur resulting from a dilated pulmonary artery with relatively normal pulmonary artery pressure or more commonly the presence of severe pulmonary hypertension. An interesting additional characteristic is the presence of a continuous murmur in some patients with small atrial septal defects and severe mitral stenosis: this murmur originates at the site of the interatrial communication when high left atrial pressure results in blood flow across the small atrial septal defect throughout the cardiac cycle. This murmur tends to be loudest along the lower right sternal edge and it may increase with inspiration and diminish or vanish with the Valsalva maneuver. The inspiratory increase in the intensity of the murmur is attributed to a late rise in left atrial

pressure during inspiration; marked reduction or disappearance of the interatrial pressure gradient has been demonstrated during straining (i.e. Valsalva maneuver).

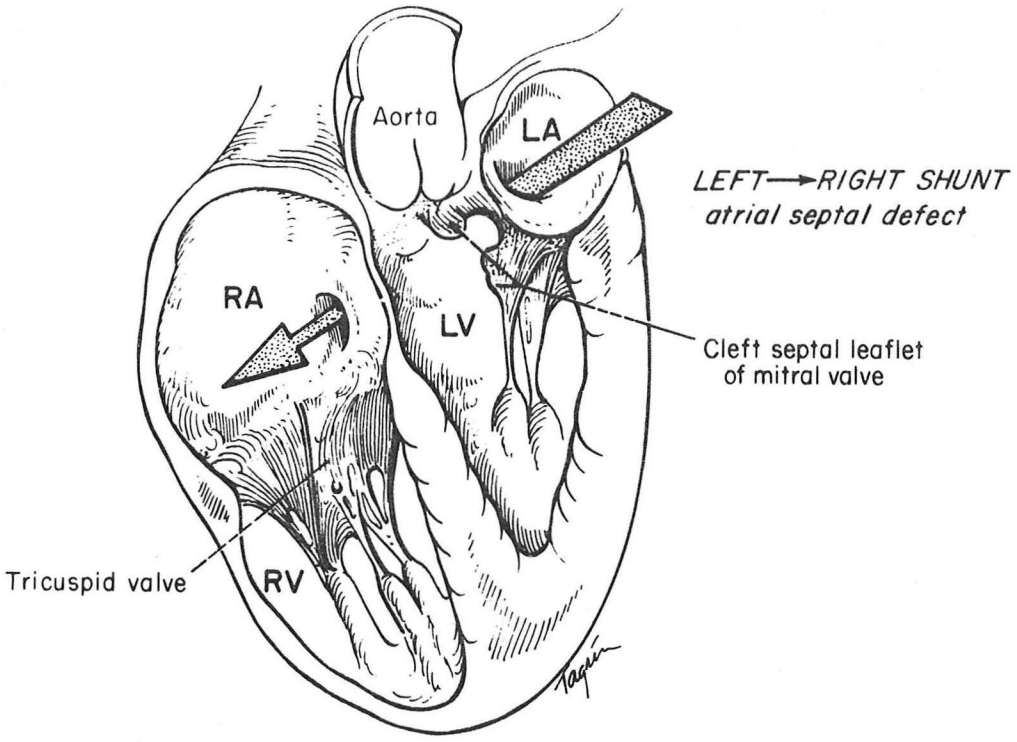
The electrocardiogram often demonstrates the same findings as those found in individuals with uncomplicated ostium secundum atrial septal defects and a left to right shunt. However, particular attention should be paid to the appearance of the P waves in individuals with Lutembacher's syndrome since atrial fibrillation is relatively frequent in individuals with Lutembacher's syndrome and in patients with sinus rhythm combined atrial enlargement may be identified. The frontal QRS electrical axis is ordinarily directed to the right and ECG evidence of right ventricular hypertrophy is common.

The chest x-ray in patients with Lutembacher's syndrome demonstrate signs of pulmonary arterial plethora but usually not pulmonary venous congestion. This is because the left atrium may decompress itself through the interatrial defect. In addition, radiologic evidence of left atrial enlargement is less marked than one would ordinarily expect for the same degree of isolated mitral stenosis.

Endocardial Cushion Defects

The endocardial cushions of the embryo contribute to the development of the mitral and tricuspid valves and to the growth and development of both the atrial and ventricular septa. Endocardial cushion defects may occur at a critical region where the atrial and ventricular septa join the two atrioventricular valves. Complete endocardial cushion defects consist of a defect in the lower part of the atrial septum and clefts in the anterior mitral and septal tricuspid leaflets; a defect in the ventricular septum is also present (Fig. 9). The partial or incomplete form of endocardial cushion defect is represented ordinarily by an ostium primum atrial septal defect and a cleft mitral valve. In this abnormality, the ventricular septum is intact and the tricuspid valve is normal or may occasionally also have a cleft.

We will not discuss complete endocardial cushion defects in any detail today, but rather will concentrate on the other relatively common type of atrial septal defect which is the ostium primum one. The interatrial communication is located in the lowermost portion of the atrial septum (Figs. 1 and 9). The upper margin of the defect is crescentic and the lower margin is formed by the atrioventricular valve tissue. With ostium primum defects and a cleft anterior mitral leaflet, the physiological consequences depend largely upon the size of the atrial defect and the degree of functional alteration of the mitral valve. A cleft anterior mitral leaflet may be functionally competent if its mobility and chordal support allow satisfactory apposition with a posterior mitral leaflet. Alternatively, inadequate chordal support can produce severe mitral incompetence even in valves that are only partially cleft. Clefts divide the anterior mitral leaflet into two parts (and when present in the tricuspid valve divide the septal tricuspid leaflet



Schematic illustration of endocardial cushion defect—primum atrial septal defect with cleft mitral valve leaflet.

Figure 9

into two parts). The anterior leaflet of the mitral valve may be partially or complete cleft. Accesssory chordae tendineae are common anatomic features of the cleft leaflet. These accessory chordae arise from the margins of the cleft and insert directly into the ventricular septum; accessory chordae are not found in the normal heart. These accessory chordae support the edges of the cleft which would otherwise be flail. When the anomalous chordae are short, thick or fused, the mobility and function of the cleft leaflet is seriously disturbed and this may result in mitral regurgitation. It is important to remember that in normal hearts the chordae of the anterior mitral leaflet arise from its free edge and insert into papillary muscles not into the ventricular septum. This arrangement permits the mitral cusp to move away from the ventricular septum during systole. However, accessory chordae tend to hold the cleft anterior leaflet relatively close to the ventricular septum and by so doing may produce subaortic obstruction or a characteristic deformity of the left ventricular outflow tract often referred to as a "goose neck" alteration.

If the ostium primum atrial septal defect is large and severe mitral regurgitation exists as well, the effects of the mitral incompetence are ordinarily transmitted to the right side of the heart with correspondingly little alteration of the left cardiac chambers. When the interatrial communication is small, the hemodynamic consequences of important mitral regurgitation are reflected in the left atrium and left ventricle and the picture becomes one dominated by the signs and symptoms of mitral regurgitation including enlargement of the left atrium and left ventricle, the characteristic holosystolic murmur at the apex and at least in some instances an "S_3-rumble complex".

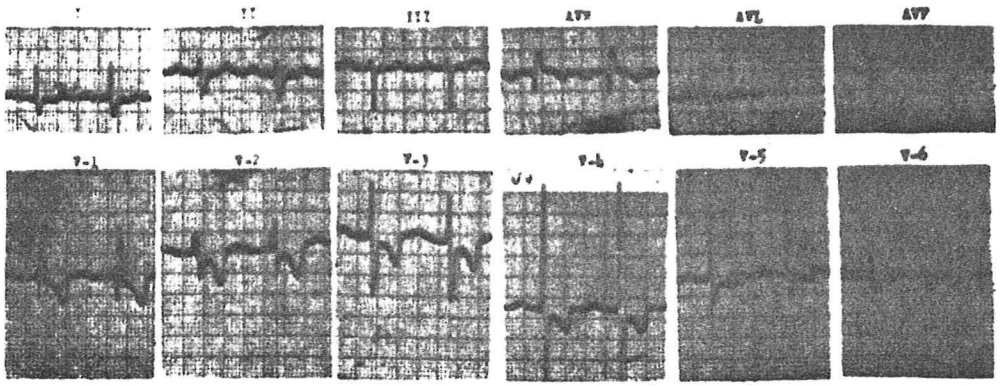
When the ostium primum atrial septal defect is large and the left to right shunt 2:1 or greater, the physical findings are essentially similar to those of isolated ventricular septal defect.

Females are affected more commonly than males with a sex ratio that is approximately the same as with ostium secundum septal defects. Cleft mitral or tricuspid leaflets are susceptible to bacterial endocarditis and this is an important feature that distinguishes ostium primum septal defects with cleft atrial ventricular valves. Mongolism in a child with congenital heart disease should immediately arouse concern of the presence of an endocardial cushion defect. In individuals with complete endocardial cushion defects the incidence of Mongolism is estimated to be as high as 50%.

The physical findings in patients with large ostium primum atrial septal defects and a cleft mitral valve resemble those found in individuals with ostium secundum atrial septal defects with the exception of the addition of the murmur of mitral regurgitation.

Sinus rhythm is the rule in individuals with ostium primum atrial septal defects, but there is a high incidence of PR interval prolongation. Arrhythmias and conduction disturbances are relatively common with this abnormality and there is an appreciable incidence of atrial fibrillation, atrial flutter and complete heart block.

The QRS complex deserves special attention in individuals with ostium primum atrial septal defects. Individuals with the abnormality typically have left axis deviation and right bundle branch block (Fig. 10); this electrocardiographic pattern helps one to distinguish patients with ostium primum from those with ostium secundum atrial septal defects. It should be emphasized however, that occasional individuals with ostium primum atrial septal defects have normal or rightward electrical axis and absence of left axis deviation so the absence of the classical ECG pattern does not categorically exclude this diagnostic possibility. There are potentially multiple reasons for the particular ECG pattern in those individuals with primum defects but slight alterations in the sequence of ventricular activation appear to be the most likely unifying explanation.

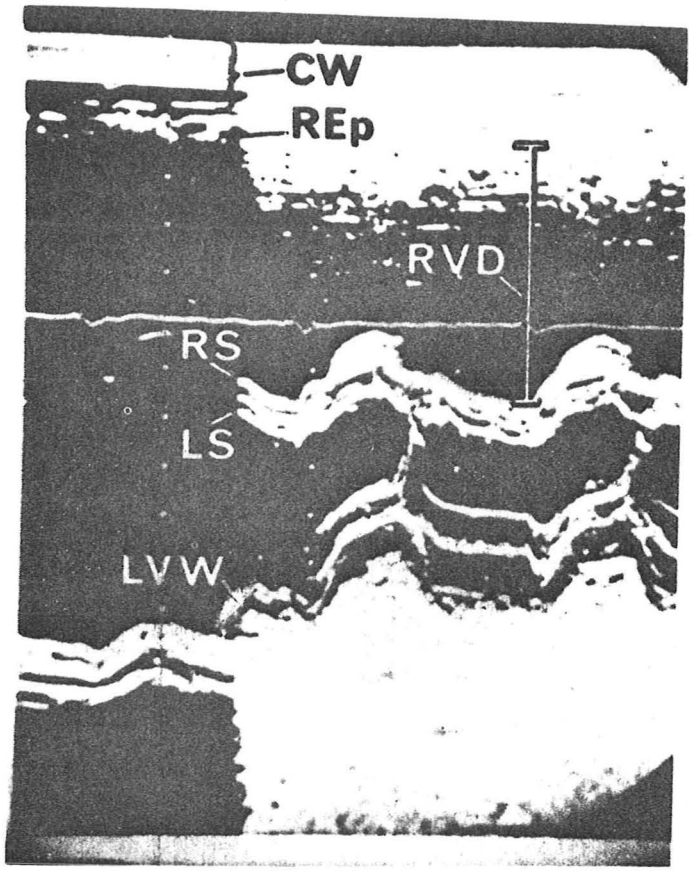


Electrocardiogram from patient with endocardial cushion defect (ostium primum atrial septal defect). There is left axis deviation in the frontal plane and a right ventricular conduction disturbance evident in Lead V_1. The P-R interval shows prolongation for rate.

Figure 10

The chest x-ray of individuals with ostium primum defects with a cleft mitral valve resembles that found in patients with ostium secundum atrial septal defects. Pulmonary plethora is striking and the aorta tends to be small. The pulmonary trunk is large with evidence of shunt vascularity ordinarily obvious. Left atrial dilatation is sometimes seen, particularly in patients with important mitral regurgitation.

Echocardiography is also a useful tool that one may utilize to help establish the presence of an atrial septal defect (Fig. 11). Classically, one expects to find "paradoxical septal motion" on the echocardiogram and enlarged right ventricular end diastolic dimensions. Neither of these observations is pathognomonic of atrial septal defect since they may also be present in right ventricular overload states of other etiologies but they are supportive of a presumptive diagnosis of atrial septal defect.

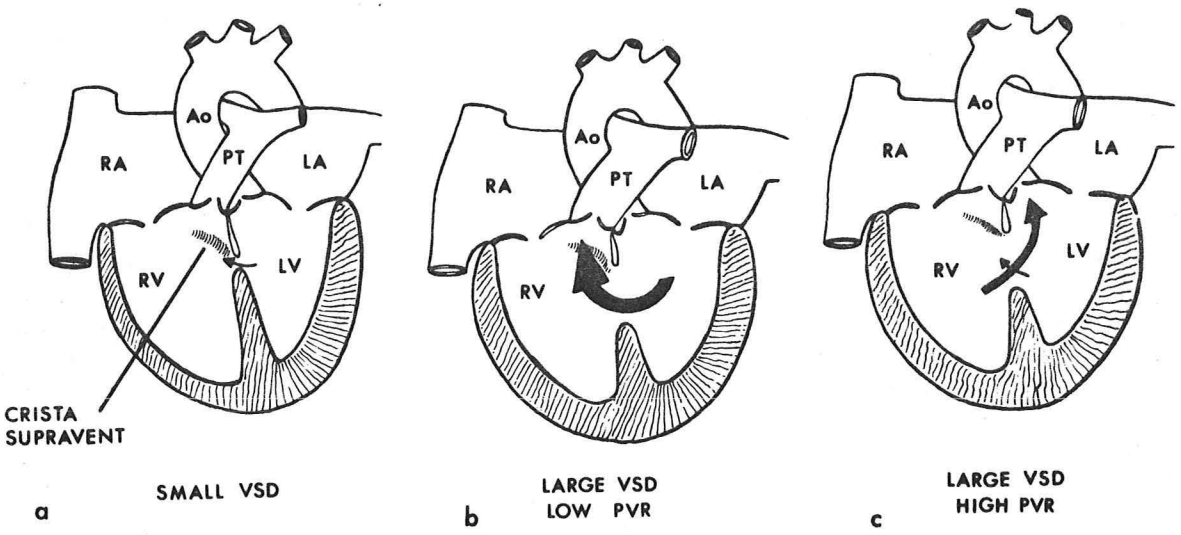


Echocardiogram from a patient with an atrial septal defect. The right ventricular dimension (RVD) is large and the interventricular septum (RS and LS) shows paradoxical motion during ventricular systole. (From Diamond, Dillon, Haine, Chang, and Feigenbaum: Echocardiographic features of atrial septal defect. Circulation, 43, 129, 1971. By permission of the American Heart Association, Inc.)

Figure 11

Ventricular Septal Defects

Ventricular septal defect is probably the commonest congenital anomaly of the heart seen clinically. The defect varies in anatomic location and is probably best considered in relationship to morphologic landmarks in the normal heart. One important landmark is the crista supraventricularis; this is a muscular ridge that separates the main portion of the right ventricular cavity from the infundibular or outflow portion (Fig. 12). Ventricular septal defects lie either above or below



Schematic illustrations of:
a, Small infracristal ventricular septal defect with small left to right shunt and normal pulmonary arterial pressure.
b, Large infracristal ventricular septal defect with low pulmonary vascular resistance and large left to right shunt.
c, Large infracristal ventricular septal defect with high pulmonary vascular resistance and reversed shunt.

Figure 12

this ridge; those positioned above the ridge are termed "supracristal" and those lying below are called "infracristal". Supracristal ventricular septal defects are situated immediately beneath the pulmonary orifice so that the valve forms part of the superior margin of the interventricular defect. Viewed from the left side, a supracristal ventricular septal defect is found just below the commissure joining the left and right aortic cusps. Infracristal septal defects may be located either in the

area of the membranous or muscular septum or in the site that would be occupied if the defect occurred as part of an endocardial cushion abnormality. Infracristal ventricular septal defects are the commonest variety and the most common type lies below and posterior to the crista in the region of the membranous septum. These defects are often partly hidden by the septal leaflet of the tricuspid valve and when viewed from the left side, they are found just beneath the aortic valve close to the commissure joining the right and noncoronary cusps. Ventricular septal defects in this position are generally referred to as "membranous" although they usually involve an adjacent portion of the muscular septum. The less common type of infracristal defect is located in the muscular portion of the septum so that its margins are made up entirely of the septal muscle. These defects may be small or large, single or multiple and they may be represented by relatively thin muscular septum that is honeycombed with fenestrations. Occasionally, an infracristal ventricular septal defect is found in the position that it would occupy if it were part of a complete endocardial cushion abnormality.

Sometimes a defect in the membranous septum exists as a blind congenital aneurysm. Such an aneurysm protrudes into the right heart in one of several different directions. It may protrude above the tricuspid valve and into the right atrium, into the septal leaflet of the tricuspid valve, or below the tricuspid leaflets and into the right ventricular cavity. These aneurysms tend to be small, but on rare occasions may be quite large producing obstruction to right ventricular outflow.

There is the possibility of spontaneous closure of ventricular septal defects. This is most likely to occur during the first few years of life when the normal growth rate of the heart is most rapid and the size of the ventricular septal defect remains essentially the same or enlarges less rapidly than the rest of the heart. This results in a tendency for the relative size of the defect to diminish particularly during the first two years of life. Septal muscle may also contract around the defect further diminishing the size of the interventricular communication. The tendency for ventricular septal defects to decrease in size may result in spontaneous complete closure of the defect. Sometimes the defect remains anatomically patent but functionally of no great importance since the left to right shunt is small or essentially nonexistent. Spontaneous closure of ventricular septal defects is not thought to be uncommon and in fact has been estimated to occur in 25-40% of ventricular septal defects during the first few months of life.

Physiologically, the consequences of ventricular septal defect depend upon the size of the defect and the degree of elevation of pulmonary arterial pressure and vascular resistance. It needs to be kept in mind that both of these variables may change with time and the physiologic manifestations will vary accordingly.

A small ventricular septal defect causes little or no functional disturbance since the shunt is negligible. With larger ventricular septal defects and specifically with left to right shunts of 2:1 or

greater several potential problems may develop including volume overload of the right heart and ultimately the possible development of significant increases in pulmonary artery pressure and vascular resistance. A large ventricular septal defect results in equilibration of systolic pressures in the two ventricles, which then behave physiologically as a common chamber. Blood is ejected into the systemic and pulmonary circuits at the same pressure so pulmonary hypertension is obligatory. In this circumstance the amount of flow into the pulmonary and systemic circulation depends upon the vascular resistance in each bed; a low pulmonary vascular resistance results in a large left to right shunt. However, as resistance rises in the pulmonary vascular bed the left to right shunt decreases and as the pulmonary vascular resistance equals or exceeds systemic the shunt becomes balanced and the left to right shunt may ultimately be replaced by a right to left one ("Eisenmenger's syndrome"). At this time, pressure overload of the right ventricle develops and the pulmonary vasculature resembles that described in the previous discussion of atrial septal defects with pulmonary hypertension.

For further insight into the vascular responses in the pulmonary arterial bed a brief review of pulmonary vascular physiology in this setting is in order. Infants born with large ventricular septal defects have a high neonatal pulmonary vascular resistance that subsequently is altered in one of four ways. 1) The high pulmonary vascular resistance may undergo normal regression with a proportionate fall in resistance, 2) delayed or incomplete regression may occur, 3) recurrence of an elevated pulmonary vascular resistance after complete or partial regression, or 4) persistence of the high pulmonary vascular resistance may be noted.

There are several regulatory mechanisms that may affect the volume and direction of the shunt at the ventricular level. These include 1) pulmonary vascular pressure and resistance as described above; 2) the relative alteration in size of the ventricular septal defect during the first few years of life; and 3) whether or not obstruction to right ventricular outflow develops, i.e. acquired infundibular pulmonic stenosis.

In children with moderate size ventricular septal defects there is a tendency for the left to right shunt and the pulmonary artery pressure to decline during the first few years of life and this tendency may continue well beyond this period. This is probably the result of a relative decrease in size of the defect which is accompanied by a fall in pulmonary vascular pressure and resistance. It should be emphasized that all patients do not experience such a favorable course, however, and pulmonary artery pressure and pulmonary vascular resistance may remain modestly elevated from the outset. Excessive volume overload of the left and right heart persists in this circumstance unless there is a marked rise in pulmonary vascular resistance. If the latter occurs, there is a subsequent favorable effect of reciprocally decreasing the left to right shunt but if the progression of increase in pulmonary

artery pressure and vascular resistance is not interrupted by surgical intervention pulmonary vascular pressures and resistance reach systemic levels, the shunt becomes bidirectional and finally at this stage, Eisenmenger's syndrome exists.

History and Physical Examination

Ventricular septal defects occur with equal frequency in males and females. Familial occurrence has been described, but does not appear to be commonly a factor. Most ventricular septal defects are discovered in early infancy primarily because of the loud murmur that accompanies the abnormality. The murmur classically appears after the relatively high neonatal pulmonary vascular resistance has returned to normal or to the level sufficient to allow left to right shunting through the defect that is present. Relatively few adults are discovered clinically with ventricular septal defect primarily because most often their communications have spontaneously closed or diminished to the point that the defects are clincally unrecognizeable. It should be kept in mind, however, that acute myocardial infarction (in particular, anterior infarcts) may produce a ventricular septal defect and sometimes large left to right shunts. Trauma has also been associated with the development of a ventricular septal defect in adults. In patients with large ventricular septal defects, mortality is highest in early childhood. In these patients the large left to right shunts do not diminish nearly enough as time passes and death results from congestive heart failure and/or from the development of severe pulmonary hypertension and subsequent congestive heart failure. Children with large ventricular septal defects have chronic cardiac failure, poor growth and development, frequent respiratory infections and very small children have feeding difficulties.

A characteristic clinical sequence consists of absence of a murmur at birth, a prominent murmur several weeks later and cardiac failure during the first week of life followed later by improvement. The majority of such patients get better because the relative size of the defect decreases and the pulmonary flow, vascular resistance and pressure fall. Consequently, both the volume overload of the left heart and the pressure and volume load in the right heart are reduced. *Such a history is sufficiently classic in its own right to raise suspicion concerning the presence of a ventricular septal defect.* A decrease in volume overload of the left heart with clinical improvement may also be a consequence of the development of infundibular pulmonic stenosis or to a rise in pulmonary vascular resistance. These regulatory mechanisms also serve to improve the hemodynamic burden on the heart since the right heart tolerates an elevated systolic pressure better than the left ventricle tolerates a large volume load. A rising pulmonary vascular resistance reduces the volume load in the left heart, but if pulmonary vascular resistance continues to rise and ultimately reaches near systemic levels or greater the left to right shunt is abolished, a balanced or net right to left shunt appears and with it cyanosis and all

the complications of pulmonary hypertension. This pattern may develop during childhood although sometimes it is present from infancy on. As mentioned earlier, the eponym "Eisenmenger's syndrome" describes this sequence of large ventricular septal defect, subsequent development of severely increased pulmonary vascular resistance and reversed or bi-directional shunting. Dr. Victor Eisenmenger described this entity in 1897 in a paper entitled "Congenital Defects of the Ventricular Septum".

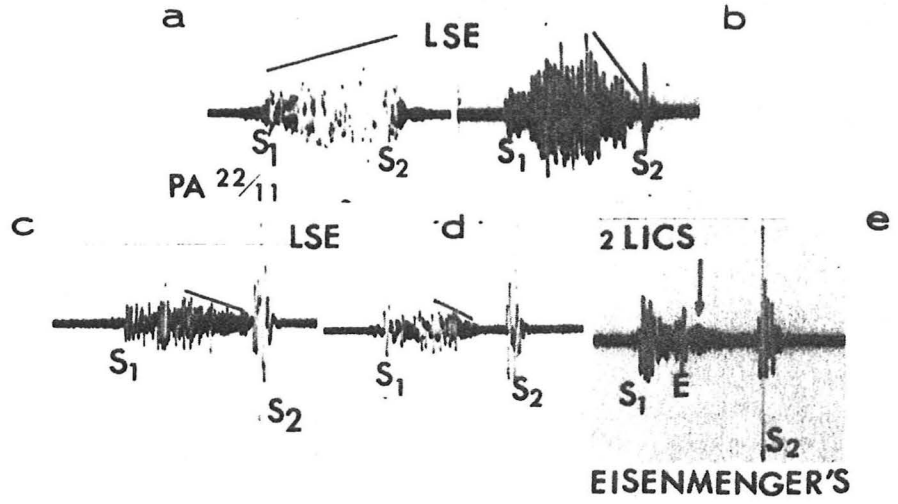
Bacterial endocarditis is a major risk for patients with ventricular septal defects. These defects are particularly susceptible to endocarditis because the high velocity shunt results in a traumatic endocardial lesion at the site of impact. This traumatic lesion is most likely to occur when the septal defect is relatively small and a considerable pressure difference exists between the left and right ventricles.

Physical Examination

One seeks to establish whether patients with ventricular septal defects have cyanosis and clubbing which would, of course, be suggestive of right to left shunting and the presence of important pulmonary hypertension. Patients with pulmonary hypertension and balanced shunts may be cyanotic only after exercise or emotion. Large ventricular septal defects with pulmonary hypertension are associated with elevations of mean jugular venous pressure; prominent V waves may develop in those individuals that develop right ventricular failure. Prominent A waves are often not present in patients with large ventricular septal defects even though pulmonary hypertension and right ventricular failure may be present. In addition, when the shunt is right to left and the ventricular septal defect large, prominent A and V waves may also be absent since the right ventricle has the ability to decompress itself.

Left to right shunts of 2:1 or greater often have a palpable systolic thrill along the lower left sternal border. Small ventricular septal defects may occur without thrills. When the ventricular septal defect is located above the crista supraventricularis, the shunt is directed into the pulmonary artery; the accompanying thrill is maximal in the second left intercostal space and may even radiate into the neck. When sizable left to right shunts occur with little or no pulmonary hypertension, the left ventricular impulse is dynamic and displaced outward and down. The right ventricular impulse may be relatively unimpressive. However, the development of pulmonary hypertension with a large left to right shunt results in a heaving right ventricular impulse and a pulsation over the pulmonary trunk. The sound of pulmonic valve closure may also be palpated as well.

The classical murmur of a ventricular septal defect is a holosystolic one heard best along the lower left sternal border (Fig. 13). When the ventricular septal defect is located below the crista supraventricularis, the murmur is maximal in the third and fourth intercostal spaces to the left of the sternum. When the defect is located above the crista supraventricularis, the accompanying murmur is maximal in the

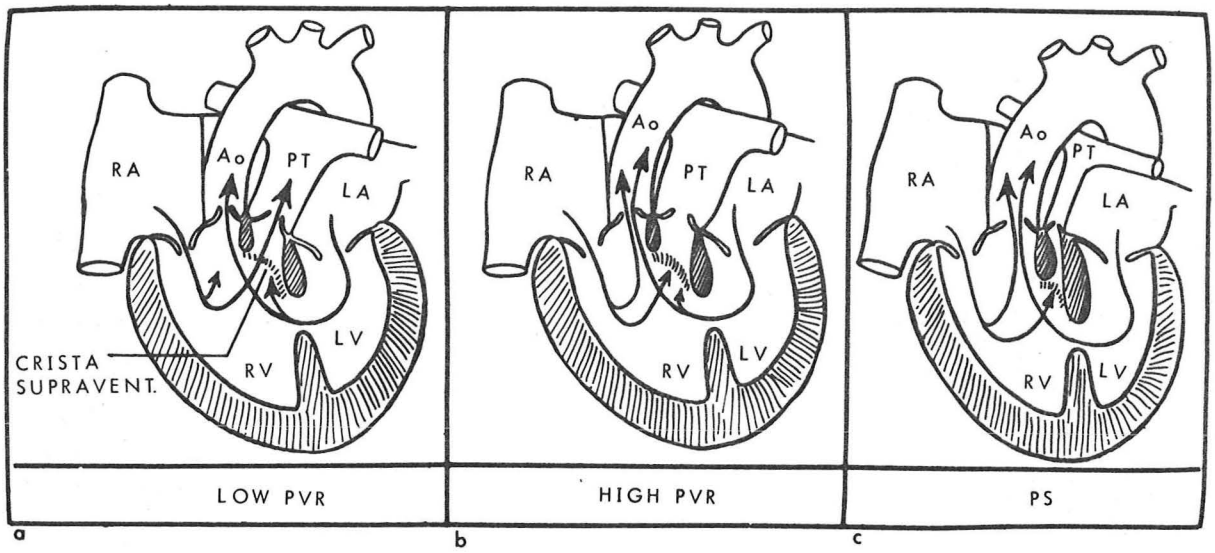


Phonocardiograms from five patients with ventricular septal defects and pulmonary arterial pressures ranging from normal to systemic levels. The tracings show the modifications of the ventricular septal defect murmur from non-pulmonary hypertensive left to right shunt (*a*) to pulmonary hypertensive right to left shunt (*e*). In Eisenmenger's complex, the VSD murmur is absent altogether, and a pulmonic ejection sound (E) introduces a short midsystolic murmur caused by ejection into a dilated pulmonary trunk. (Perloff, J. K.: Progr. Cardiov. Dis. *9*:303, 1967.)

Figure 13

second left intercostal space. Small ventricular septal defects tend to generate high frequency early systolic murmurs that are prominent. As the size of the defect increases, the murmur remains loud, harsh and holosystolic as long as a significant pressure difference exists between the left and right ventricle throughout systole. As pulmonary hypertension develops and the pressure gradient between the left and right ventricle disappears, the systolic murmur becomes progressively softer and ultimately disappears altogether as the shunt becomes balanced or right to left (Fig. 13). Flow into a dilated pulmonary artery resulting from pulmonary hypertension ordinarily produces an ejection sound and a short and soft systolic ejection murmur that is maximal in the second left intercostal space. A murmur of pulmonic insufficiency, a high frequency diastolic decrescendo murmur best heard in the second left intercostal space, is heard in some patients who develop significant pulmonary hypertension. A diastolic apical rumble that is short and localized to the point of maximal impulse is heard in some patients with left to right shunts of 2:1 or greater and provides a way at the bedside to estimate the severity of the shunt through a ventricular septal defect.

The electrocardiogram is a relatively accurate monitor of the hemodynamic disturbance in the sense that it is affected chiefly by the size of the defect, the level of pulmonary vascular resistance and the degree of volume overload of the left and pressure overload of the right heart. Large left to right shunts result in electrocardiographic evidence of left atrial enlargement. The QRS electrical axis is generally directed downward; once severe pulmonary hypertension develops, a rightward shift in the axis occurs. A left axis deviation in an individual with a ventricular septal defect suggests the presence of an endocardial cushion defect and a ventricular septal defect at that location (or that both great vessels originate from the right ventricle) (Fig. 14). In



Schematic illustrations of right ventricular origin of both great vessels with ventricular septal defect *below* the crista supraventricularis. The septal defect is so aligned that left ventricular blood is directed toward the aorta.

a, When the pulmonary vascular resistance (PVR) is relatively low, a sizable portion of left ventricular blood enters the pulmonary trunk so that pulmonary flow is increased. Blood from the right ventricle flows principally toward the pulmonary artery so that cyanosis is minimal or absent.

b, As the pulmonary vascular resistance rises, right ventricular blood is diverted toward the aorta so that cyanosis develops. A left to right interventricular shunt remains obligatory, but less left ventricular blood enters the pulmonary bed so that pulmonary flow declines.

c, In the presence of severe pulmonic stenosis (usually infundibular), right ventricular blood is diverted from the pulmonary artery to the aorta so that cyanosis is present. The left to right interventricular shunt remains obligatory, although little or no left ventricular blood enters the pulmonary bed.

Figure 14

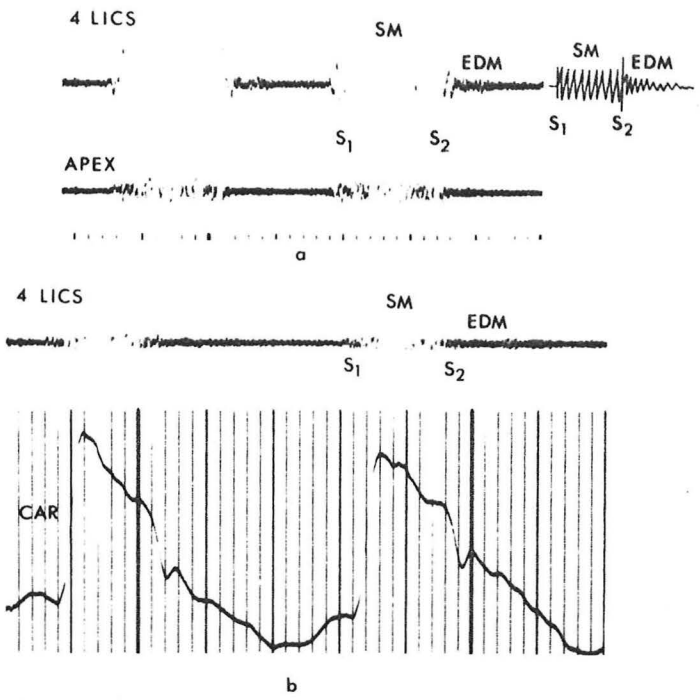
(Modified from Perloff, J.K. The Clinical Recognition of Congenital Heart Disease, W.B. Saunders Company, Philadelphia, 1970.)

the setting of the large left to right shunt, evidence of left ventricular hypertrophy and strain develops; once severe pulmonary hypertension develops then the electrocardiographic indications of right ventricular

hypertrophy, right atrial enlargement and as noted earlier, right axis deviation. Small ventricular septal defects are generally associated with normal chest x-rays. Large ventricular septal defects with persistent left to right shunts are associated with left ventricular enlargement radiographically and the development of "shunt vascularity" as described earlier. The development of important pulmonary hypertension also results in radiographic changes that were described previously, but include marked enlargement of the main pulmonary trunk, important enlargement of the proximal pulmonary arteries and rapid tapering of the pulmonary vasculature so that the peripheral pulmonary artery radicals appear small and the distal lung fields relatively oligemic.

Ventricular Septal Defect and Aortic Regurgitation

Occasionally individuals are seen that have both a ventricular septal defect and clinical evidence of aortic valvular regurgitation. The ventricular septal defect in this setting may be located either above or below the crista supraventricularis. The commonest anatomic arrangement is for the ventricular septal defect to be located below the crista and in the region of the membranous septum and as such is positioned just beneath the right coronary and noncoronary aortic cusps. The aortic insufficiency ordinarily develops years after birth, but it is believed to arise as a result of a congenital abnormality in aortic leaflet support. With infracristal ventricular septal defects, the aortic regurgitation appears to be most commonly related to underdevelopment of an aortic commissure, generally the right coronary-noncoronary commissure. The leaflets are poorly supported, leading to a fault in leaflet apposition. Often, the right coronary cusp is prolapsed and distorted, or both the right and the noncoronary cusps are prolapsed. It should be emphasized that most clinical observations of individuals in this setting have suggested that aortic regurgitation develops after birth and progresses as time goes on consistent with the above description. The physical findings in patients with both ventricular septal defects and aortic regurgitation are those that represent a combination of these two lesions in their classical forms (Fig. 15).



a, Phonocardiograms from an eight year old boy with ventricular septal defect (2 to 1 left to right shunt) and mild aortic incompetence. In the fourth left intercostal space (4 LICS) a prominent holosystolic murmur (SM) is followed by a soft, high frequency, early diastolic murmur (EDM). These murmurs are schematically illustrated to the right. At the apex the holosystolic murmur is well recorded, but the early diastolic murmur is virtually absent.

b, Tracings from a 17 year old boy with ventricular septal defect and aortic incompetence. The left to right shunt was 1.3 to 1 and the brachial arterial pressure 135/30 mm. Hg. (The wide pulse pressure is not well reflected in the indirect carotic tracing shown here.) The phonocardiogram in the fourth left intercostal space shows the high frequency holosystolic murmur of small ventricular septal defect and the soft early diastolic murmur of aortic incompetence.

Figure 15

(Modified from Perloff, J.K. The Clinical Recognition of Congenital Heart Disease, W.B. Saunders Company, Philadelphia, 1970.)

Tetralogy of Fallot

The entity is considered the most common cyanotic cardiac lesion in patients surviving infancy. The entity as originally described by Fallot (1888) consists of four anatomic lesions: 1) pulmonary stenosis, 2) ventricular septal defect, 3) right ventricular hypertrophy and 4) overriding of the aorta. Pulmonary stenosis is usually of the

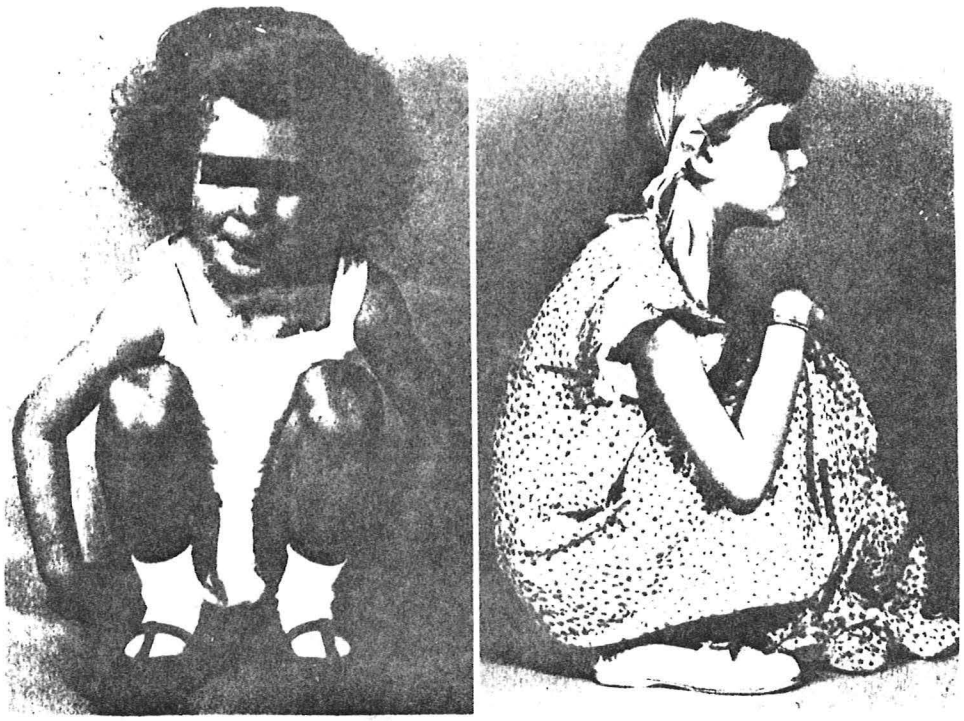
infundibular type, occasionally is located at the valvular level, or may be a combination of both. Rarely, pulmonary valve atresia or absence of the left main pulmonary artery have also been noted. Typically, the ventricular septal defect is large and lies just above the crista supra-ventricularis in close proximity to the medial leaflet of the tricuspid valve. The aorta is overriding as noted and the degree of right ventricular hypertrophy depends on the severit of right ventricular outflow obstruction.

Individuals with tetralogy of Fallot have a right to left shunt through the ventricular septal defect; this shunt is the consequence of the presence of important right ventricular outflow obstruction and the septal defect. The shunt is further enhanced by the overriding aorta which allows direct streaming of blood from the right ventricle into the aorta. The ventricular septal defect ensures that right ventricular systolic pressure equals but does not exceed systemic arterial pressure and thus, the right ventricle is protected from performing pressure work beyond its capacity. This explains why young individuals with tetralogy of Fallot rarely exhibit cardiac failure (although it must be kept in mind that some individuals with tetralogy of Fallot have other valvular or myocardial abnormalities that may lead or contribute to the development of heart failure).

When a reduction in pulmonary blood flow and the right to left shunting are responsible for chronic systemic hypoxia, the systemic hypoxia encourages the development of polycythemia and the development of collateral vascular supply.

History

Males and females are affected equally by this entity. Most patients with tetralogy of Fallot are symptomatic and the symptoms are the result of the reduction in pulmonary blood flow and the right to left shunting through the ventricular septal defect. Symptoms include dyspnea on exertion and variable degrees of exercise intolerance. Cyanosis usually develops within the first few months of life and slow growth and development. Patients with severe right ventricular outflow obstruction commonly have episodes of severe cyanosis sometimes associated with loss of consciousness and occasionally convulsions. These attacks are referred to as "anoxic spells" and it is believed that these spells are the result of additional right ventricular outflow obstruction resulting from reactive spasm associated with emotional crises; this additional right ventricular outflow obstruction enhances the right to left shunt producing more cyanosis and a further reduction in systemic oxygen tensions and loss of consciousness and/or convulsions. Individuals with this entity (particularly young children) commonly squat in an attempt to increase systemic arterial oxygen saturation (Fig. 16). Squatting increases systemic vascular resistance thereby decreasing the magnitude of the right to left shunt and encouraging the flow of blood from right ventricle to pulmonary artery. This allows for potential increases in systemic arterial oxygen tension. The squatting also decreases venous return from the legs and minimizes the effects of orthostatic hypotension.



Photographs showing the squatting posture in two children with cyanotic Fallot's tetralogy. The position was assumed effortlessly.

Figure 16

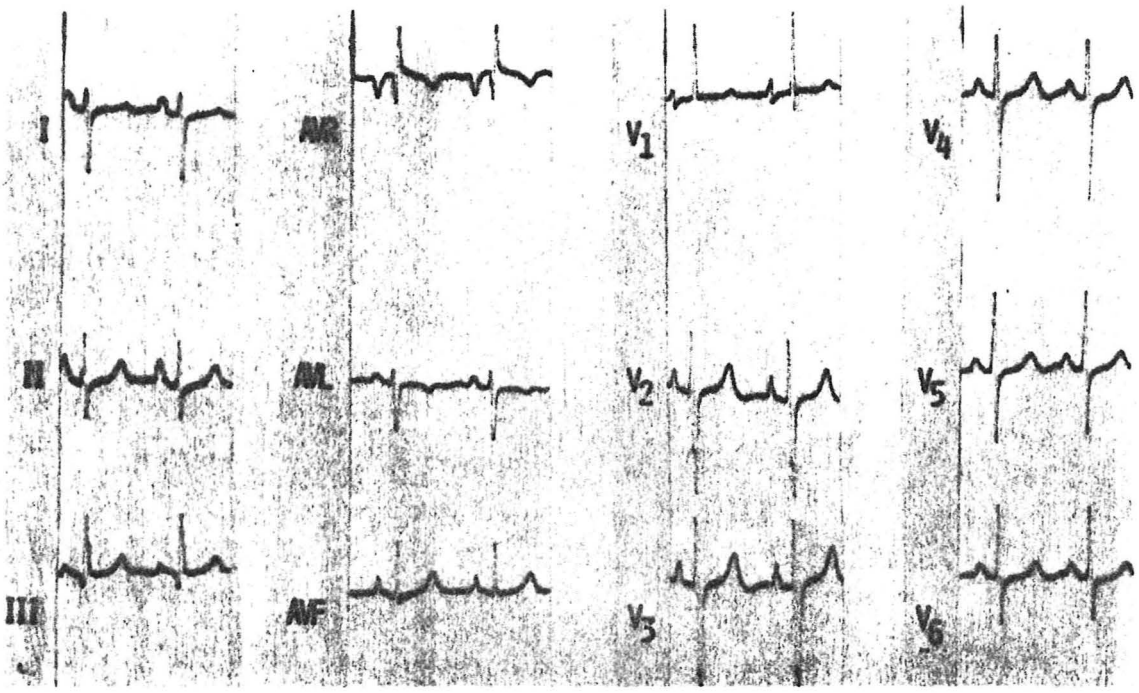
(Modified from Perloff, J.K. The Clinical Recognition of Congenital Heart Disease, W.B. Saunders Company, Philadelphia, 1970.)

Physical Examination

Cyanois and clubbing are generally present in individuals with tetralogy of Fallot. A right ventricular lift with no clear left ventricular enlargement is ordinarily also present. A systolic ejection click is often audible; this may be detected in patients with pulmonary valvular stenosis in which case the ejection click diminishes with inspiration or disappears altogether. In some individuals with severe right ventricular outflow obstruction, a hypoplastic pulmonary trunk and a relatively large aortic root the click may originate in the aortic root. The second sound is ordinarily single representing aortic valve closure. Pulmonic valve closure is rarely audible. This is a result of the severe rightsided outflow obstruction which is present. A systolic ejection murmur of a varying intensity is heard maximally in the third and fourth left intercostal spaces; with severe degrees of right ventricular outflow obstruction and the presence of a large VSD the murmur becomes

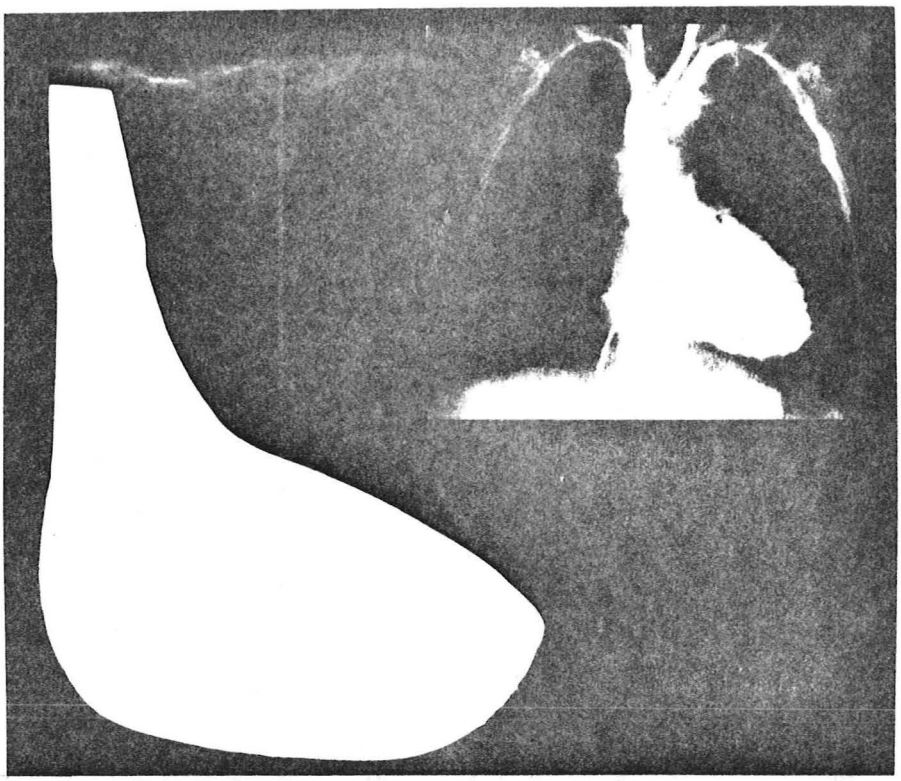
shorter and softer and where shunting is entirely right to left the VSD murmur disappears. A continous murmur representing blood flow through enlarged bronchial collaterals is often heard over the back and in the second right and left intercostal spaces.

The electrocardiogram discloses right axis deviation and right ventricular hypertrophy (Fig. 17). Characteristic chest x-ray consists of a relatively normal sized heart with a right ventricular contour, a concavity in the region of the main pulmonary artery and decreased pulmonary vascularity; the typical appearance as demonstrated in Figure 18 has been referred to as the coeur en sabot. The aortic arch is often rightsided; in particular the more severe the right ventricular outflow obstruction the more likely the aortic arch to be in the right chest (Figs. 18 & 19).



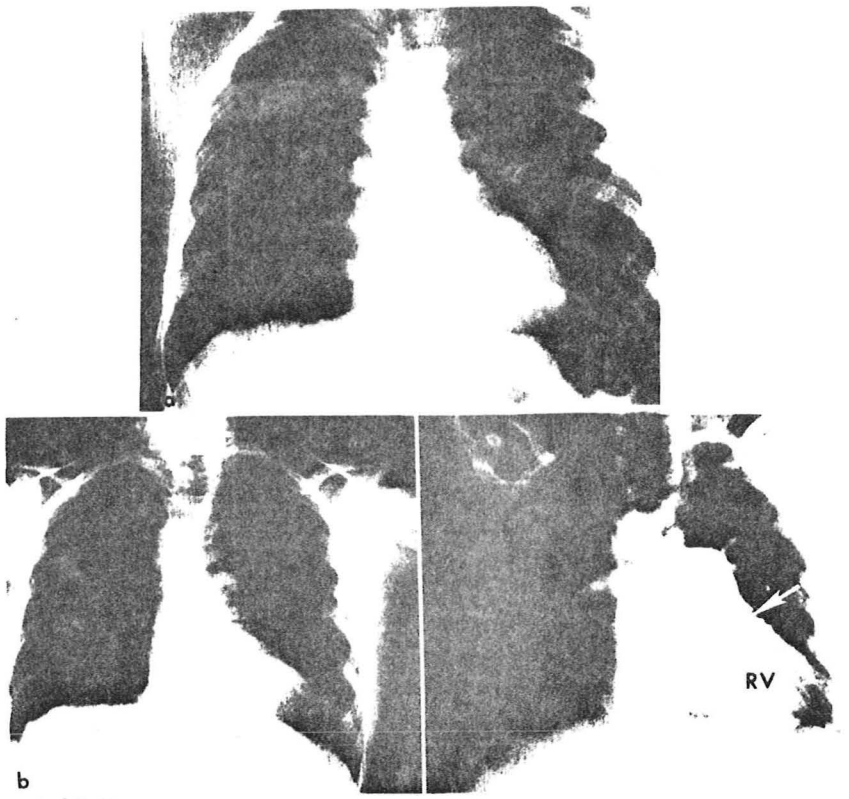
Electrocardiogram from patient with tetralogy of Fallot. There is right axis deviation, right atrial enlargement, and right ventricular hypertrophy (R wave in V_1). Note early transition in V_2 and V_3.

Figure 17



Silhouette of a golf club (wood) showing the similarity to the cardiac silhouette (insert) in pulmonary atresia with right aortic arch. The angiocardiogram is from the 20 month old cyanotic boy referred to in Figure 18-6. (The golf club silhouette is published through the courtesy of Dr. Gerold G. Schiebler. University of Florida.)

Figure 18



a, X-ray from a nine and a half year old girl with Fallot's tetralogy and mild to moderate cyanosis. The pulmonary vascularity appears diminished. A right aortic arch indents the right side of the trachea. The cardiac size is normal. The apex is rounded rather than boot-shaped because the left ventricle is well formed.

b, X-ray and angiocardiogram from a five year old girl with Fallot's tetralogy and mild to moderate cyanosis. The pulmonary vascularity appears diminished, although the film is overpenetrated. However, the overpenetration makes it easier to see a right aortic arch indenting the right side of the trachea. The pulmonary trunk is slightly convex. The cardiac apex is rounded rather than boot-shaped because the left ventricle is well formed. The angiocardiogram (injection into right ventricle) shows infundibular pulmonic stenosis (arrow), a well developed pulmonary artery and a right aortic arch with a right descending aorta. The rounded apex is radiolucent because the left ventricle forms this contour (compare with plain film). Note that the right subclavian and right common carotid arteries originate from the aortic arch.

Figure 19

Course and Prognosis

The anoxic spells that occur in patients with tetralogy of Fallot have already been discussed. These episodes tend to occur during early childhood and are rare after the fifth year of life. Treatment should be instituted immediately when they occur and should consist of oxygen administration, placing the individual in the knee chest position to increase systemic vascular resistance and the administration of morphine. Morphine is given because of its central nervous system sedative effect and because it tends to relax the right ventricular infundibulum. Beta blocking agents such as propanolol have also been used in an effort to decrease the reactive right ventricular infundibular obstruction.

All patients with cyanotic congenital heart disease (and particularly those with tetralogy of Fallot) are at risk to develop brain abscess. The most frequent organism encountered in these abscesses is the alpha streptococcus. The most common site for abscess development in the brain is the right parieto-occipital region. Treatment consists of intense antibiotic therapy and drainage of the abscess.

Other potential complications occurring with tetralogy of Fallot are bacterial endocarditis, polycythemia and very late in the course the development of right or biventricular failure.

Individuals with tetralogy of Fallot that are severely cyanotic at birth with markedly reduced pulmonary blood flow and severe systemic arterial unsaturation rarely survive the first year of life without surgical intervention. The largest group of patients with tetralogy of Fallot are those who become cyanotic during the first year of life. These individuals have moderately severe obstruction to right ventricular outflow and significant, but not severe systemic arterial hypoxia. As children they are limited and their exercise tolerance is poor. They squat frequently. Without some form of surgery, either palliative or corrective, they rarely survive beyond the third decade of life. Death usually results from cerebro vascular accidents, brain abscess, endocarditis, anoxia or pulmonary hemorrhage.

Surgical treatment consists of either a palliative procedure to increase pulmonary blood flow and thus, relieve severe systemic hypoxia or total correction of the malformation. The palliative procedure consists of a shunt from a systemic to a pulmonary artery; some of these operations are described below. The Blalock-Taussig operation consists of an anastomosis of the subclavian artery to the side of the pulmonary artery, the Waterston operation consists of a side to side anastomosis of the ascending aorta to the right pulmonary artery and the Potts procedure consists of an anastomosis of the aorta to the main pulmonary artery. Today, the most common operation used for palliative shunting is the Waterston procedure. The shunt procedure often provides considerable clinical and hemodynamic benefit to these patients for several years. Operative mortality for the shunt procedures varies depending on the status and size of the patient, but it is approximately 5% in most

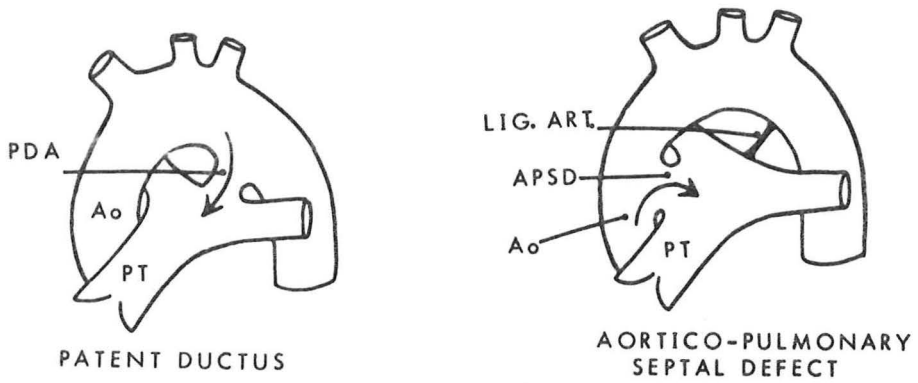
centers. Complications of the shunt procedure include thrombosis of the shunt, endocarditis, pulmonary artery hypertension (when the shunt that is created is too large) and localized hemorrhagic pulmonary edema. Another procedure which should also be mentioned which is also palliative and improves pulmonary blood flow is the Brock procedure which consists of relief of the right ventricular outflow obstruction by pulmonary valvulotomy and infundibular resection.

Complete repair of this defect requires cardiopulmonary bypass. The infundibular obstruction is resected, the pulmonary valvular stenosis (if present) is relieved by open valvulotomy and a patch is placed in the ventricular septal defect. In some instances it is necessary to provide a patch that enlarges the pulmonary outflow tract to further relieve the obstruction. In several academic centers total correction of this entity can be accomplished with an operative mortality of approximately 10%. This procedure is presently being performed in individuals at relatively young ages in some centers (3-5 years of age) while others prefer to wait until the child is slightly larger before attempting it. The complications of the procedure include heart block, a residual defect in the ventricular septum, the development of right ventricular failure secondary to either significant residual right ventricular infundibular obstruction or to pulmonary arteriolar changes resulting from chronic low pulmonary blood flow and polycythemia.

Patent Ductus Arteriosus

Patent ductus arteriosus is an arterial channel connecting the left (and rarely the right) pulmonary artery to the aorta just distal to the left subcalvian artery (Fig. 20). Embryologically, the ductus consists of the distal portion of the sixth left aortic arch. During fetal life it allows the pulmonary artery blood to bypass the lungs and enter the descending aorta. The ductus arteriosus ordinarily closes during the last weeks of intrauterine life or the first several weeks after birth. Closure is thought to be a progressive event rather than an instantaneous one and the mechanism for normal closure is thought to be primarily an increase in the oxygenation of the fetal blood. Prostaglandins may also play some role in persistent patency of the ductus. Persistent hypoxia and maternal rubella have also been implicated as possible etiologic factors in persistent patency of the ductus after birth. Patients born at high altitudes have a higher incidence of persistence of this fetal connection.

At birth, pulmonary artery pressure and vascular resistance decreases with initation of respirations and systemic vascular resistance increases with occlusion of the umbilical cord. The direction of flow through the ductus changes so that blood moves from the aorta to the pulmonary artery through the ductus. With normal pulmonary artery pressures, the flow through the ductus occurs in both systole and diastole giving rise to the classical continuous murmur. This "runoff" occuring during both



Schematic illustrations of patent ductus arteriosus (PDA) and aorticopulmonary septal defect (APSD). The aortic end of the ductus lies just beyond the origin of the left subclavian artery; the pulmonary orifice of the ductus is located immediately to the left of the bifurcation of the pulmonary trunk.

The aorticopulmonary septal defect consists of a round or oval communication between adjacent parts of the ascending aorta and pulmonary trunk. The ligamentum arteriosum is shown as a helpful landmark.

(Modified after Edwards, J. E., et al.: Congenital Heart Disease.)

Figure 20

phases of the cardiac cycle results in a wide systemic pulse pressure and a relatively low diastolic arterial blood pressure. With large shunts through the ductus, the left ventricular stroke volume is markedly increased and the work of the left ventricle is so increased that congestive heart failure may ensue. In a small percentage of patients, probably less than 10%, pulmonary arterial hypertension and pulmonary vascular obstruction develop in association with the patent ductus arteriosus. In those circumstances in which pulmonary artery pressure exceeds systemic pressure, the direction of the shunt is reversed and becomes right-to-left through the ductus.

Clinical Features and History

Isolated patent ductus arteriosus occurs approximately three times as freqeuntly in females as in males. A small patent ductus arteriosus is ordinarily well tolerated and symptoms may be absent or not become evident until childhood or early adult life. However, with large shunts through the ductus left ventricular failure develops.

On physical exam one expects to find evidence of left ventricular enlargement with a sizable left to right shunt. Bounding peripheral pulses with a widened pulse pressure will also be found. A systolic or systolic and diastolic thrill is palpable in the suprasternal notch and second left interspace in many individuals with the abnormality. Auscultation reveals a normal first heart sound followed by an aortic ejection sound at the apex and left sternal border. The second heart sound is often obscured by the murmur, but is narrowly split. The pulmonic component may become accentuated as pulmonary artery pressure rises. The third heart sound is occasionally heard. The characteristic "machinery murmur" is heard best in the second left interspace with radiation to the anterior chest, neck and back. The murmur begins after the first sound, persists through the second and fades away gradually during diastole. It begins softly and increases in intensity so as to reach its peak at or immediately after the second sound and from that point gradually wanes until its termination. When a large shunt is present, an apical middiastolic murmur may be noted secondary to increased flow across the mitral valve. The systolic murmur of increased flow across the aortic valve is heard with large left to right shunts and when severe pulmonary hypertension develops the diastolic decrescendo murmur of pulmonary regurgitation may also be audible. Patent ductus arteriosus is one cause of continous heart murmurs. Table III lists other causes of continuous murmurs that should be considered in the differential diagnosis.

TABLE III

Differential Diagnosis of Continuous Murmurs*

(from Clinical Cardiology, edited by Willerson, J.T. and Sanders, C.A., Grune & Stratton, June, 1977)

Location	Differential Diagnosis
First to second left intercostal spaces (and under left clavicle)	1. Patent ductus arteriosus
Second to fourth intercostal spaces	2. Aorticopulmonary septal defect
Usually best heard in the second to third left intercostal spaces; occasionally may be best heard at the right of the sternum in the same area	3. Surgical shunts such as aorto-pulmonary anastomoses
Usually best heard along the lower left sternal border though it may be audible over the entire precordium	4. Rupture of sinus of Valsalva aneurysm

Audible over the left precordium	5. Coronary arteriovenous fistulae
May be audible anywhere that they occur	6. Arteriovenous fistulae

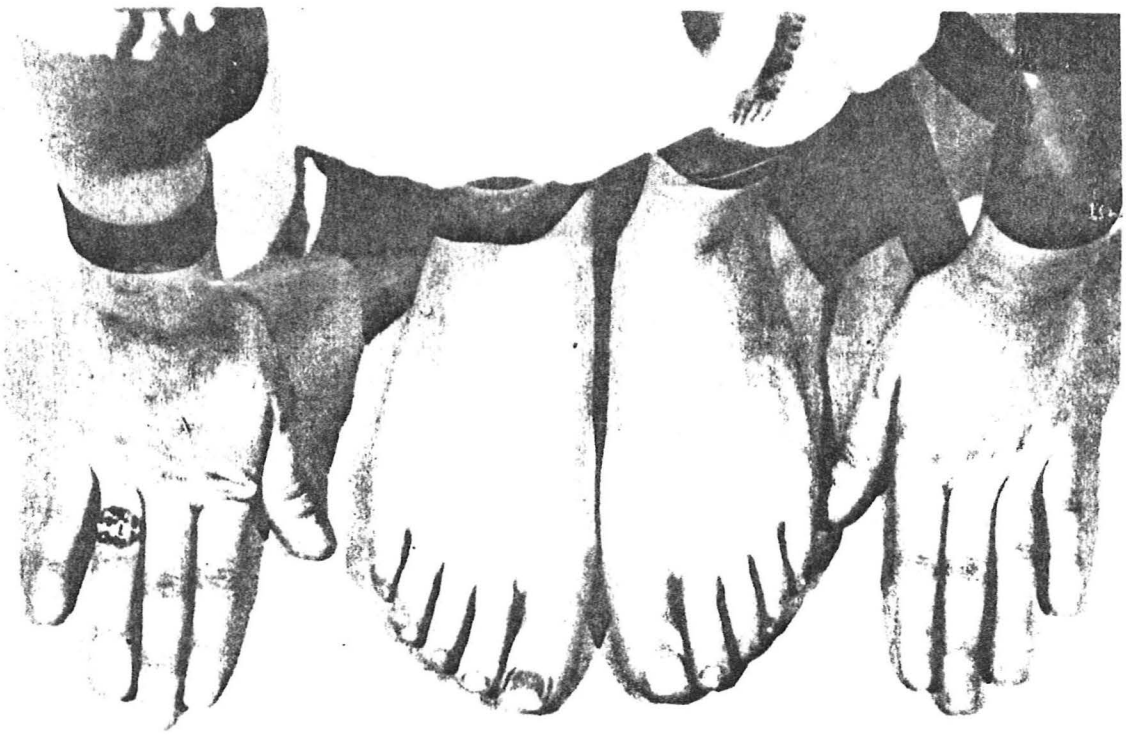
*This refers to murmurs that increase in intensity at the end of systole, envelop the second sound, and diminish in diastole. The generation of this type of murmur depends on a continuous flow of blood from a chamber or a vessel of higher pressure to one of lower pressure with persistence of pressure differences in both systole and diastole.

Left ventricular hypertrophy and left atrial enlargement are ordinarily obvious with large left to right shunts through the ductus. Right ventricular hypertrophy, if present, is indicative of the significant increase in pulmonary artery pressure. The chest x-ray demonstrates left ventricular and left atrial enlargement and a prominent main pulmonary artery. Chest fluoroscopy ordinarily reveals a pulsating aortic arch, increased pulmonary vascular markings and increased pulsations in the hilar shadows. With pulmonary hypertension, right ventricular enlargement will also be noted. Cardiac catheterization may prove the presence of the ductus arteriosus by identifying an oxygen saturation stepup from the right ventricle to the pulmonary artery with the highest readings being recorded from the left pulmonary artery. Peripheral systemic arterial saturation is normal except in those cases with a reversal of flow secondary to marked increases in pulmonary vascular resistance and pressure. At the time of cardiac catheterization one may be able to pass the catheter from the pulmonary artery through the ductus into the descending aorta which also proves the presence of the ductus arteriosus.

Patent ductus arteriosus may be associated with several different cardiac abnormalities although most commonly with a ventricular septal defect, coarctation of the aorta, aortic stenosis and pulmonic stenosis. In some patients, the ductus arteriosus may be the primary source of blood flow to the lungs, i.e. in pulmonary atresia or it may be the primary source of blood flow to the distal aorta, i.e. in complete interruption of the aortic arch.

Course and Prognosis

Patent ductus arteriosus will rarely close spontaneously when it is still patent after a few months of life. Potential complications include bacterial endarteritis and congestive heart failure. The occurrence of severe pulmonary artery hypertension and pulmonary vascular obstruction may occur relatively late in the adult. Reversed flow (right to left shunt) through a patent ductus arteriosus is a cause of differential cyanosis, i.e. cyanosis and clubbing of lower extremities and lack of such changes in the upper extremities (Fig. 21). Rupture of a patent ductus is a rare complication -- usually associated with the development of an aneurysm or calcification of the structure. The life



Photograph of a 14 year old girl with patent ductus arteriosus, pulmonary hypertension and reversed shunt. The girl is squatting with her hands placed alongside her feet in order to compare the fingers (which were acyanotic) with the toes (which were cyanotic). In fact, she had painted her toenails (see right great toe) to hide their cyanotic color.

Figure 21

expectancy of a patient with patent ductus arteriosus who does not undergo surgical treatment depends on the size of the ductus, but on the average is about 40 years. A small shunt through a ductus arteriosus is compatible with a normal life expectancy.

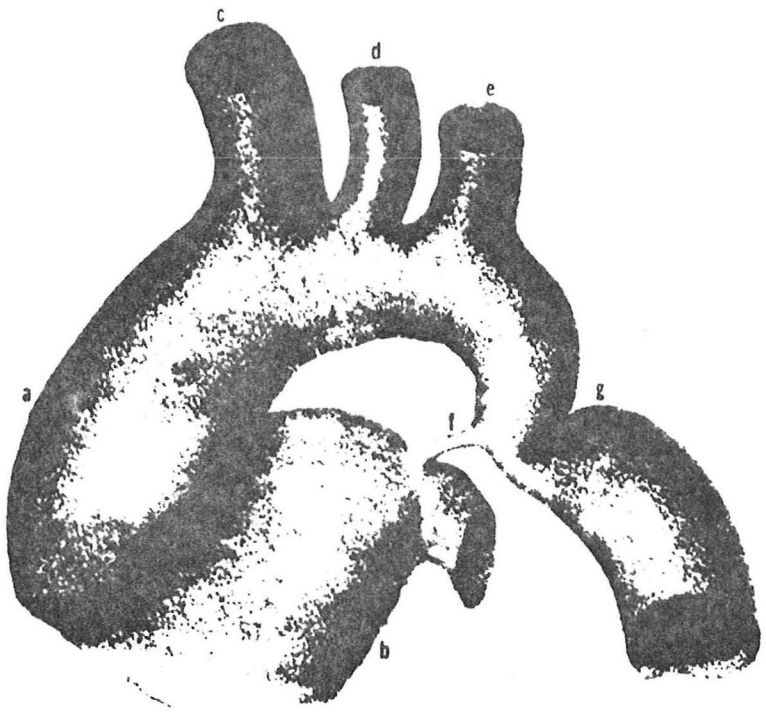
Medical treatment in the past has been directed at potential complications including antibiotic prophylaxis against bacterial endarteritis and measures to treat congestive heart failure (if present). More recently, there is evidence that prostaglandin synthesis inhibitors (indomethacin) may be capable of producing closure of a patent ductus in small children, but the frequency with which this occurs remains to be established in larger series of patients.

In 1938, Gross performed the first surgical correction of a patent ductus arteriosus. The surgical closure of the ductus is ordinarily a relatively simple procedure and is accomplished by simple ligation or division of the ductus. Elective surgery has been advised for all patients over one year of age; symptoms make earlier surgery a procedure to be considered, but as noted above it may well be that the use of

prostaglandin synthesis inhibitors will also be an effective means, at least in some patients, of producing ductal closure soon after birth. Surgical therapy carries a very small operative risk (approximately 0.5%) and is highly successful. Once severe pulmonary hypertension and and pulmonary vascular obstructive disease develops, then surgery is contraindicated. Surgery may also be contraindicated in those individuals in whom inoperable associated anomalies are present and particularly in those in whom the ductus serves as the essential pathway connecting the pulmonary and arterial systems.

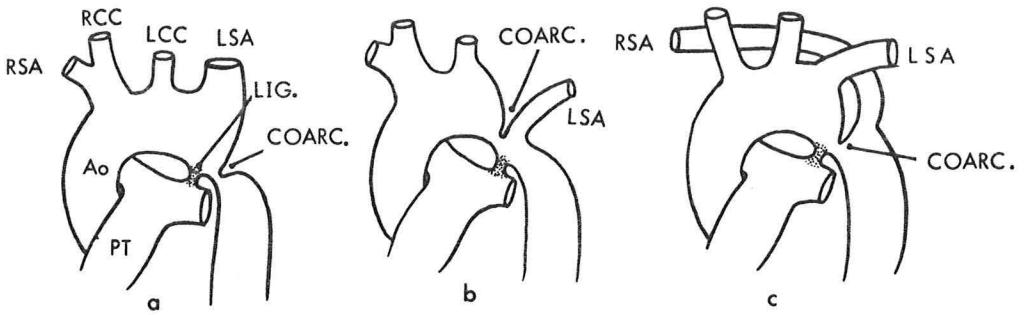
Coarctation of the Aorta

Coarctation of the aorta is a narrowing of the aortic lumen usually in the region of the ligamentum arteriosus (Fig. 22, Panels A & B). The lesion may occasionally occur higher in the aortic arch, the descending thoracic aorta or even in the abdominal aorta. Historically, coarctation of the aorta was classifed into two groups: infantile (preductal) and adult (postductal). The former is characterized by a relatively long constricted segment of aorta situated proximal to the ductus arteriosus. The adult type consists of a localized narrowing at or just distal to the ductus arteriosus. This classification is now of most value to the



An anatomic sketch of coarctation of the aorta published in 1834. Compare with Figure 7-1 *a*. (Nixon, R. L. Dublin J. Med. *In* Chem. Sci. 5:386-400. 1834). (Courtesy of Dr. Saul Jarcho.)

Figure 22
Panel A



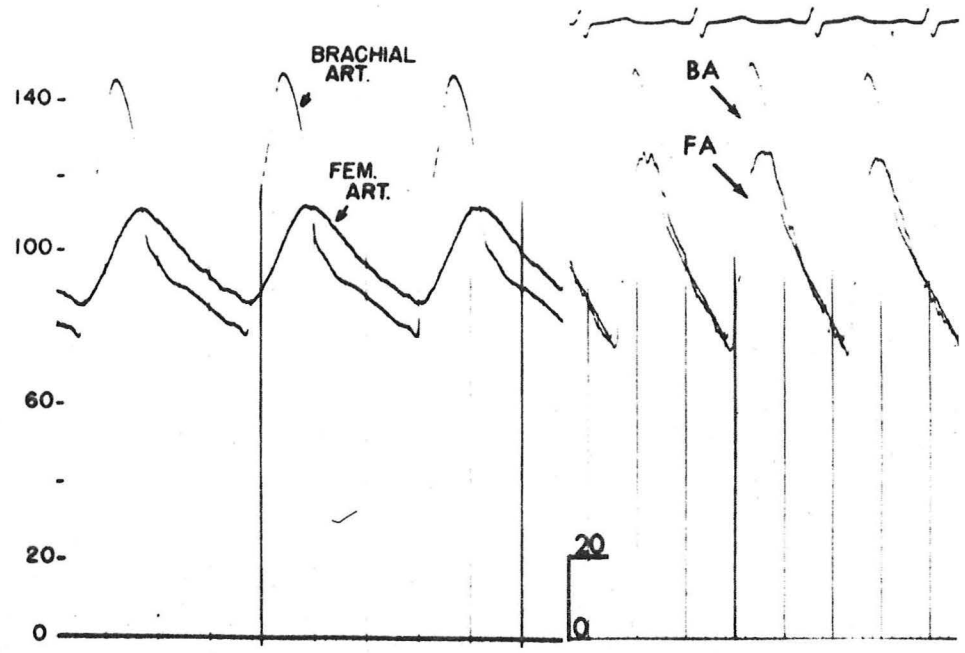
Schematic illustration of the classic variety of coarctation of the aorta and two anatomic variations.
a, In the classic variety the zone of coarctation is located immediately beyond the left subclavian artery (LSA) which tends to be enlarged. The descending aorta is often locally dilated distal to the coarctation. (RSA-right subclavian artery; RCC and LCC-right and left common carotid arteries; LIG-ligamentum arteriosum; Ao-ascending aorta; PT-pulmonary trunk.)
b, The site of coarctation is shown just proximal to the left subclavian artery, which is in the low pressure zone. Under these circumstances the left subclavian is not dilated.
c, The *right* subclavian artery (RSA) arises anomalously below the coarctation and is therefore in the low pressure zone.

Figure 22
Panel B

pathologist, but to a physiologist and clinician this anatomic differentiation has little meaning since the lesion may fit one group anatomically and another functionally. Hence, a newer classification has been developed in which coarctation of the aorta is divided as follows: 1) those in which the left ventricle supplies the entire systemic circuit, and 2) those in which the left ventricle supplies the upper half of the body and the lower half is supplied by the right ventricle through a patent ductus arteriosus. This classification takes into account not only the ductus above and below the coarctation, but also those inserting at the site of aortic narrowing. The latter group -- coarctation of the aorta with systemic left and right ventricles -- will not be further discussed here since its occurrence is ordinarily limited to the first year of life.

The major hemodynamic problem created by coarctation of the aorta is the maintenance of adequate blood flow to the regions distal to the obstruction, most particularly the kidneys. This is accomplished by an

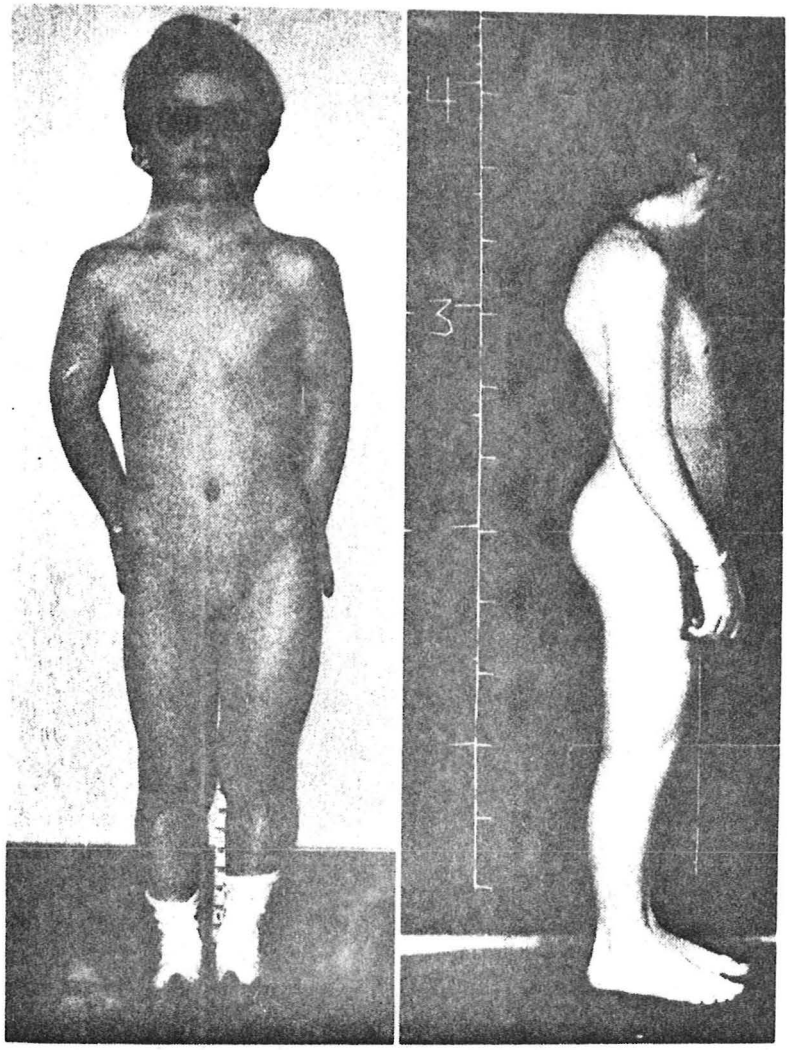
elevation of the systolic blood pressure in the proximal aortic segment (and to a much lesser degree the diastolic blood pressure although the diastolic pressure often remains normal) (Fig. 23) and by utilizing collaterals in the chest wall or a ductus emptying below the site of the obstruction.



Simultaneous brachial and femoral arterial pressure pulses from two boys aged 10 and 4 years with mild to moderate coarctation of the aorta. In both, the femoral pulses were palpable but smaller than the brachials. The important differences in upper and lower extremity pressures are the *systolic* and *not* the diastolic levels.

Figure 23

Aortic coarctation may coexist with patent ductus arteriosus, aortic arch, and aortic valve abnormalities (in particular a bicuspid aortic valve). Associated intracardiac abnormalities may include ventricular septal defect, congenital mitral regurgitation, and endocardial fibroelastosis. Coarctation of the aorta has been relatively frequently found in association with Turner's syndrome (Fig. 24).

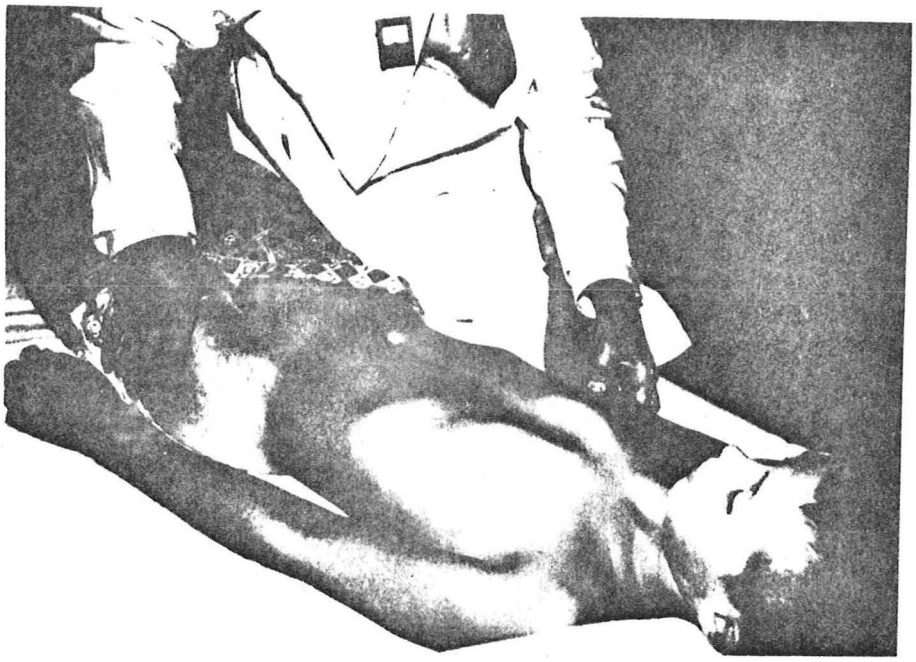


Two girls aged 13 and 14 years, respectively, with Turner's syndrome and coarctation of the aorta. Both patients exhibit short stature, webbing of the neck, absent pubic hair, wide set nipples and small chin. In addition, both wore bangs to cover their low hairlines.

Figure 24

Clinical Features

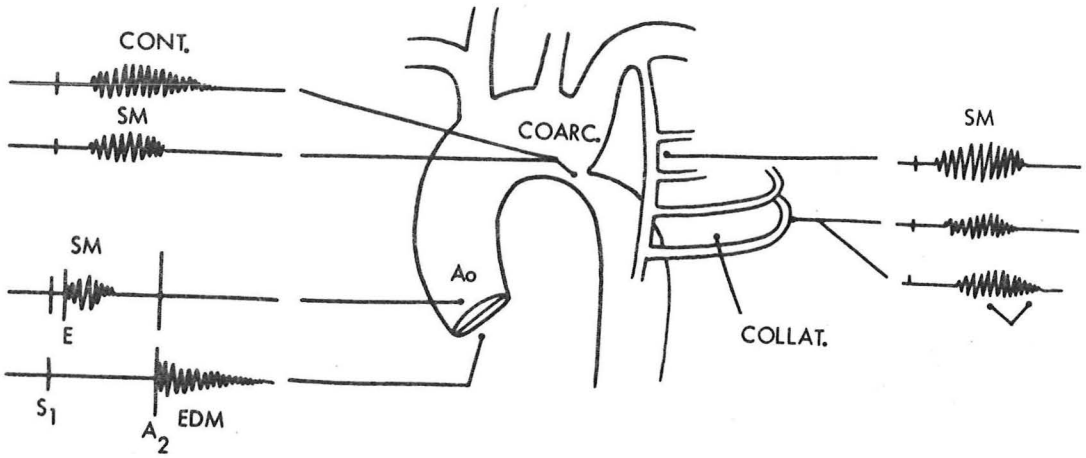
The majority of children and young adults with the classical type of coarctation are asymptomatic. Males are more commonly afflicted than females with a ratio 2:1. Diagnosis of aortic coarctation is often made during the routine physical examination at which time systemic arterial hypertension is observed in the upper extremities in association with absent or diminished femoral arterial pulsations (Fig. 25). As noted earlier, the hypertension is chiefly systolic. Symptoms when present include those associated with hypertension, i.e. headache, epistaxis, dizziness and palpatations. Occasionally decreased blood flow to the lower extremities will result in intermittent claudication during exercise. A relatively small percentage of patients develop congestive heart failure in association with aortic coarctation.



Simultaneous palpation of the brachial and femoral arteries is the most useful and convenient way of comparing the upper and lower extremity pulses. Application of the thumbs as illustrated allows either simultaneous or rapidly sequential palpation, which permits detection of subtle differences in amplitude and timing. (Perloff, J. K.: G. P. *33*:78, 1966.)

Figure 25

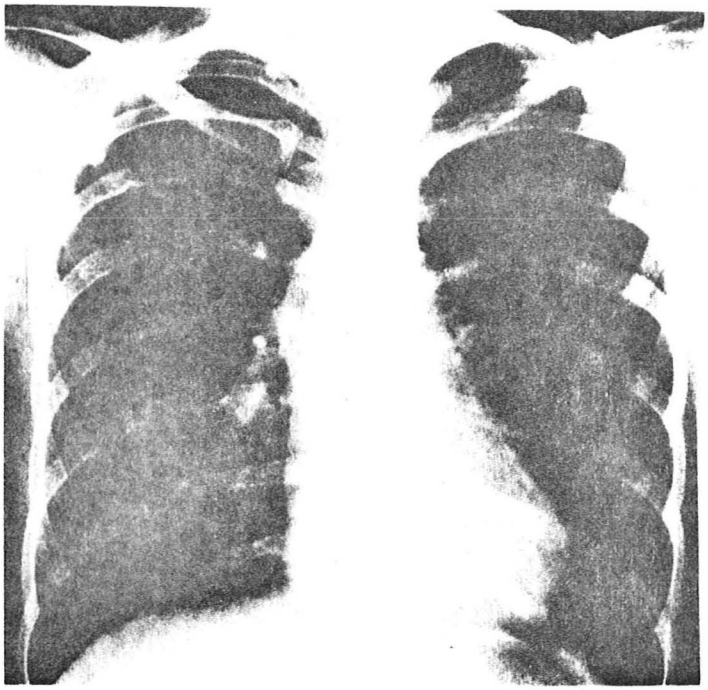
On physical examination patients with coarctation are ordinarily well developed. The esential feature for the diagnosis of coarctation of the aorta is a higher systolic arterial blood pressure in the arms than in the legs. The diastolic pressures are ordinarily the same. Thus, the upper extremities have a widened pulse pressure. Palpation of the femoral and brachial pulses simultaneously discloses a weaker or absent or delayed femoral pulsation (Fig. 25). Pressure should be measured in both upper extremities to identify possible involvement of the left subclavian artery by the coarctation. A systolic thrill is often palpable in the suprasternal notch. Left ventricular enlargement may also be identified. A systolic ejection click is often audible and increased prominence of the aortic closure sound noted. A characteristic rough systolic ejection murmur may be heard along the left sternal border and in the back, particularly over the area of the coarcted segment (Fig. 26). A continuous murmur may also be heard over the back and/or in the interscapular or subscapular areas indicating increased blood flow through collateral channels. In approximately 30% of individuals with aortic coarctation, a systolic murmur of an associated bicuspid aortic valve is audible at the base of the heart. A murmur of aortic insufficiency may also be detected in some of these individuals.



Schematic illustration of the principal murmurs heard in patients with coarctation of the aorta. The lower left side of the illustration shows the auscultatory events associated with a bicuspid aortic valve, namely, an aortic ejection sound (E), a short midsystolic murmur (SM), and an early diastolic murmur (EDM) of aortic incompetence. On the upper left are shown the continuous (CONT) and the delayed systolic murmurs (SM) that originate in the coarctation itself and are best heard posteriorly over the thoracic spine. On the right are shown collateral arterial murmurs that are crescendo-decrescendo in shape and delayed in onset and termination because of origin in vessels that are some distance from the heart. These collateral murmurs are best designated "delayed systolic murmurs." (Collateral murmurs are, as a rule, bilateral and are here shown on one side simply as a matter of convenience).

Figure 26

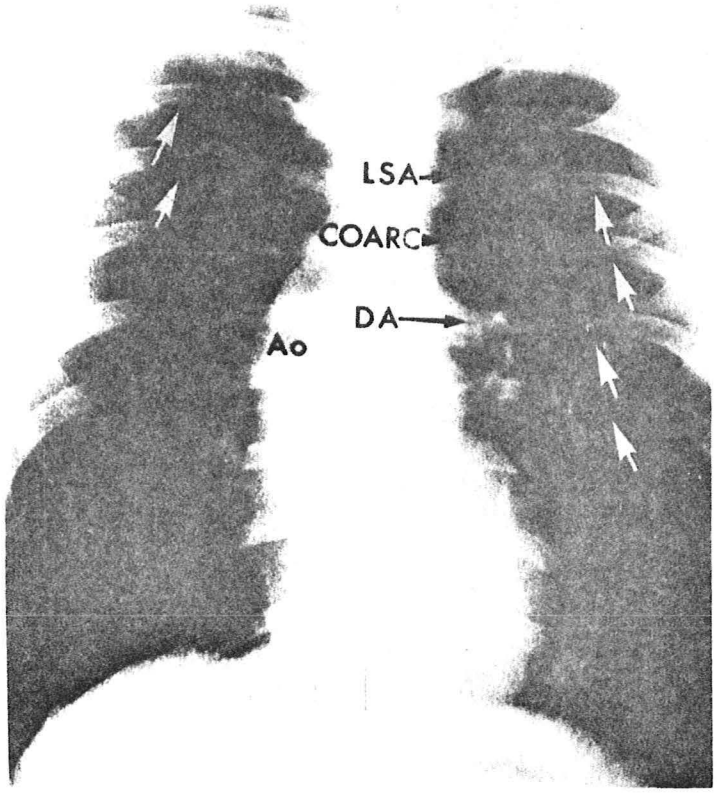
The electrocardiogram may demonstrate evidence of left ventricular hypertrophy. Heart size by chest x-ray may be normal although often left ventricular enlargement is noted. Notching of the ribs develops in young and older adults with coarctation of the thoracic aorta (Figs. 27 & 28). Rib notching develops due to increased collateral flow through the intercostal arteries. The rib notching develops along the posterior surface of the ribs between ribs three and eight; the notching may by symmetrical or rarely may be unilateral. The anterior ribs are spared because the anterior intercostal arteries do not run in costal grooves. Notching of the ribs rarely develops prior to age four. It should be further emphasized that when the zone of coarctation is located just distal to the left subclavian artery, rib notching is bilateral and confined to posterior ribs 3-8. However, collateral vessel development depends upon patency of the origins of the subclavian arteries. Therefore, when the coarct severely narrows the orifice of the left subclavian artery, collateral circulation fails to develop on the left side and unilateral notching develops in the right posterior ribs. When there is



X-ray from a 12 year old girl in whom coarctation had compromised the lumen of the left subclavian artery. There is unilateral notching of ribs (right hemithorax). The ascending aorta is prominent but the left subclavian artery is not dilated. There is slight rounding of the left ventricular silhouette.

Figure 27

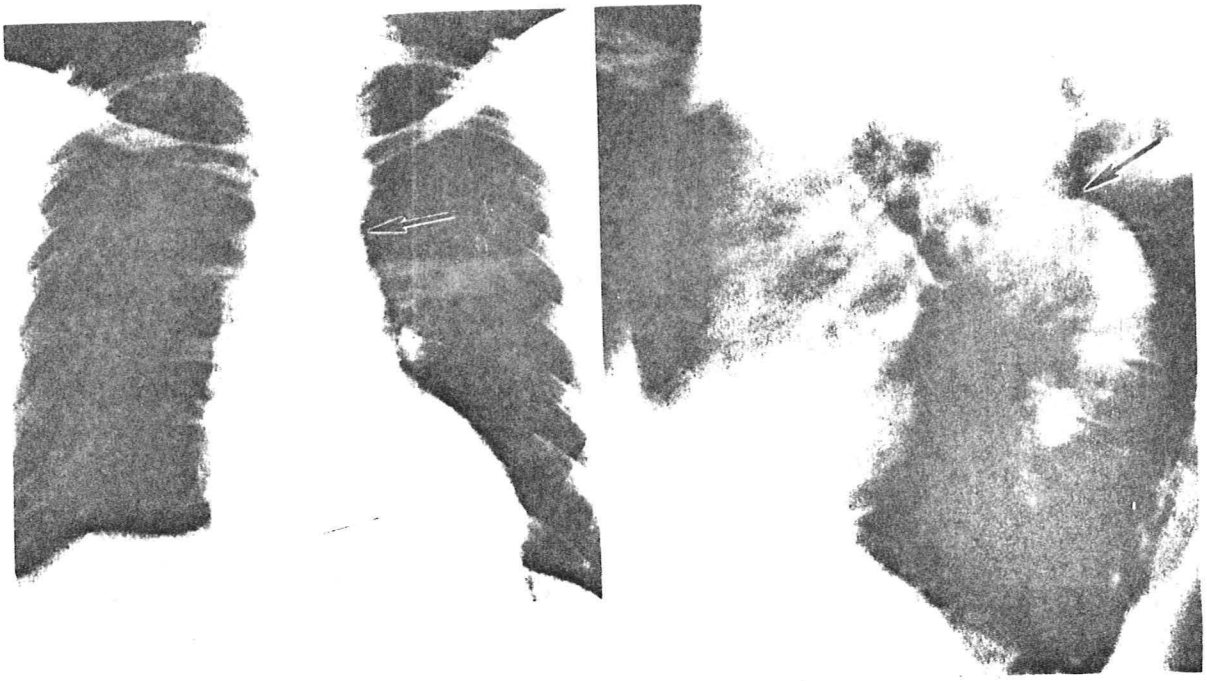
anomalous origin of the right subclavian artery distal to the coarctation, collaterals fail to develop in the right hemithorax and unilateral notching develops on the left. In those rare circumstances in which the coarct develops proximal to the innominate artery, rib notching does not develop. When the coarct involves the lower thoracic or abdominal aorta, rib notching is confined to the lower ribs.



X-ray from a 23 year old man with coarctation of the aorta. Arrows point to sites of notching on the undersurfaces of the posterior ribs. The ascending aorta (Ao) forms a rightward convexity. In addition, a dilated left subclavian artery (LSA) is seen above the coarctation and the dilated descending aorta (DA) is seen below, forming together the silhouette of a "figure 3."

Figure 28

The coarctation itself may be visible as an indentation in the aorta, a dilated left subclavian artery and pre and poststenoic dilatation of the aorta (Fig. 28). The classical radiologic sign is the E sign which represents impressions of the descending arch of the aorta on the barium filled esophagus. The first impression on the esophagus is from the aortic segment just above the coarctation; the second from the coarctation itself and the third from the poststenoic dilatation. Angiography, preferably an aortogram, will outline the site and extent of obstruction and the collateral circulation. One interesting pitfall in the radiographic identification of coarctation results from "pseudo-coarctation of the aorta" (Fig. 29). This term identifies an aortic



X-rays from two patients with "pseudocoarctation" characterized by buckling or kinking of the aorta (arrows) at or just beyond the site of the ligamentum arteriosum. There is no narrowing of the aortic lumen at the site of the external deformity. The posteroanterior film is from a 62 year old man. The lateral film is from a 68 year old woman. (Courtesy of Dr. Homer Twigg, Georgetown University Hospital, Washington, D. C.)

Figure 29

arch anomaly characterized by kinking at or just beyond the ligamentum arteriosum. This is a developmental abnormality in the aorta, but it differs from true coarctation by the absence of narrowing of the aortic lumen at the site of the localized external deformity.

Course and Prognosis

Patients with coarctation are at risk to develop cerebrovascular accidents, congestive heart failure and hypertensive encephalopathy. In addition, bacterial endartertis is a constant threat. Aortic dissection and/or rupture are also potential complications for individuals with this entity.

Medical treatment is directed toward blood pressure control and toward treatment of the complications should they develop. Coarctation that results in a significant gradient between the proximal and distal portions of the aorta and systemic arterial hypertension is ordinarily treated surgically with low operative mortality. The surgery, when possible, should be performed after five years of age to provide opportunity for collateral development and to allow the aorta to enlarge to a

point that it will be of adequate size at the anastomotic site when the patient is fully grown. Problems related to surgical correction include brain and/or spinal cord injury, renal damage, persistent hypertension, mesenteric arteritis (this is an extremely rare complication today in adults) and late aneurysm formation at the site of the anastomosis. Older patients have the additional technical hazards associated with an atherosclerotic aorta.

Malformations of the Coronary Arterial System

Malformations of the coronary arterial system may be divided into two types -- anomalous origin and anomalous communications of the coronary arteries.

Anomalous Origins of the Coronary Arteries

Five principal anomalous arrangements have been described.

1. The left coronary system originates from the pulmonary artery. This is the most common one.

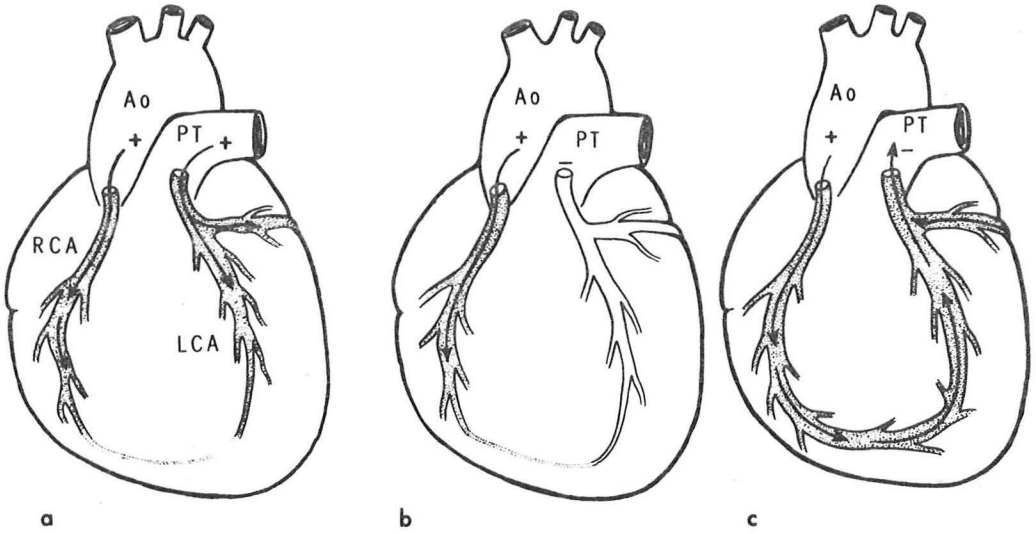
2. Both coronary arteries originate from the pulmonary artery. This is very rare and is frequently associated with other severe congenital cardiac lesions. Death occurs in the neonatal period.

3. The right coronary artery originates from the pulmonary artery. This also is quite rare and apparently not associated with significant other cardiac difficulties.

4. A single coronary ostia is present in the aorta from which one main coronary artery originates. This vessel may divide into a right and left coronary artery or may directly give off the usual branches of the coronary arterial system. In the absence of associated defects, this causes no hemodynamic problems.

5. Origin of the left circumflex artery from the right coronary artery is a relatively common malformation which by itself causes no functional derangement.

In this discussion we will concentrate on anomalous origin of the left coronary artery from the pulmonary artery as this lesion is the one of major clinical significance and the one that may be amenable to correction (Fig. 30).



Patterns of flow through the coronary bed in anomalous origin of the left coronary artery from the pulmonary artery. (Modified after Edwards, J. E.: Circulation 29:163, 1964.)

a, In the fetal and early neonatal periods the high pressure in the pulmonary trunk (PT) provides a perfusion gradient for flow *into* the anomalous left coronary artery (LCA). The aorta (Ao) perfuses the normally originating right coronary artery (RCA). Intercoronary anastomoses are not functional.

b, The subsequent fall in neonatal pulmonary arterial pressure is accompanied by a parallel decline in flow through the left coronary. Intercoronary anastomoses are still not functional.

c, When the pressure in the anomalous left coronary falls *below* that in the right, blood can then flow from *right* to *left* coronary artery through intercoronary anastomoses. The left coronary artery then drains *into* the pulmonary trunk rather than receives blood from it.

Figure 30

Myocardial ischemia is the most serious problem with this lesion. The cause of the ischemia is based chiefly upon the direction of flow through the coronary bed (Fig. 30). In the fetus and early neonate, pulmonary arterial hypertension provides an adequate perfusion gradient into the anomalous coronary. When this perfusion falls to below systemic levels, blood flow then occurs from right to left coronary artery through intercoronary anastomosis. These intercoronary anastomotic channels bypass the capillary bed, thus depriving the myocardium of oxygen and causing ischemia. The resultant myocardial ischemia may result in infarction with fibrosis, dysfunction of the mitral valve apparatus with mitral regurgitation, congestive heart failure, or even the development of a ventricular aneurysm. In addition, there is a small left to right shunt into the pulmonary artery from the aorta through these connections.

History

This anomaly occurs equally among males and females. Evidence of ischemic pain and cardiac failure usually become manifest after the first few months of life and may cause irritability, dyspnea, wheezing, cough, pallor and sweating. These findings are often precipitated or aggravated by feeding. In some individuals there are no other detectable signs until congestive heart failure develops. Sudden death is a frequent occurrence both in the infant and in the older patient with or without prior evidence of heart disease.

Clinical Features

The physical examination usually reveals an acutely and chronically ill underdeveloped child with evidence of congestive heart failure. The heart is usually enlarged and cardiac pulsations generally diminished. Pulsus alternans reflects left ventricular failure. Murmurs may be present; the murmur of mitral regurgitation is generally the one that develops. However, a continuous murmur may also be audible reflecting the intercoronary anastomoses that connect the right and left coronary arteries. Third heart sounds are ordinarily prominent.

The electrocardiogram is characteristic demonstrating evidence of myocardial ischemia or infarction. Often there are deep Q waves in leads I, AVL, V_5 and V_6. Left ventricular hypertrophy may also be evident in older children and young adults.

Generalized cardiac enlargement is usually present and particularly enlargement of the left ventricle.

At cardiac catheterization, a small left-to-right shunt at the pulmonary artery level will often be detected by oxygen analysis and/or by dye dilution curve. Endocardial fibroelastosis is sometimes associated with anomalous origin of the coronary artery from the pulmonary artery, but this is most likely to occur when both coronary arteries originate from the pulmonary artery. Abnormalities of the mitral and tricuspid valves and tetralogy of Fallot have been reported in association with anomalous origin of the coronary arteries.

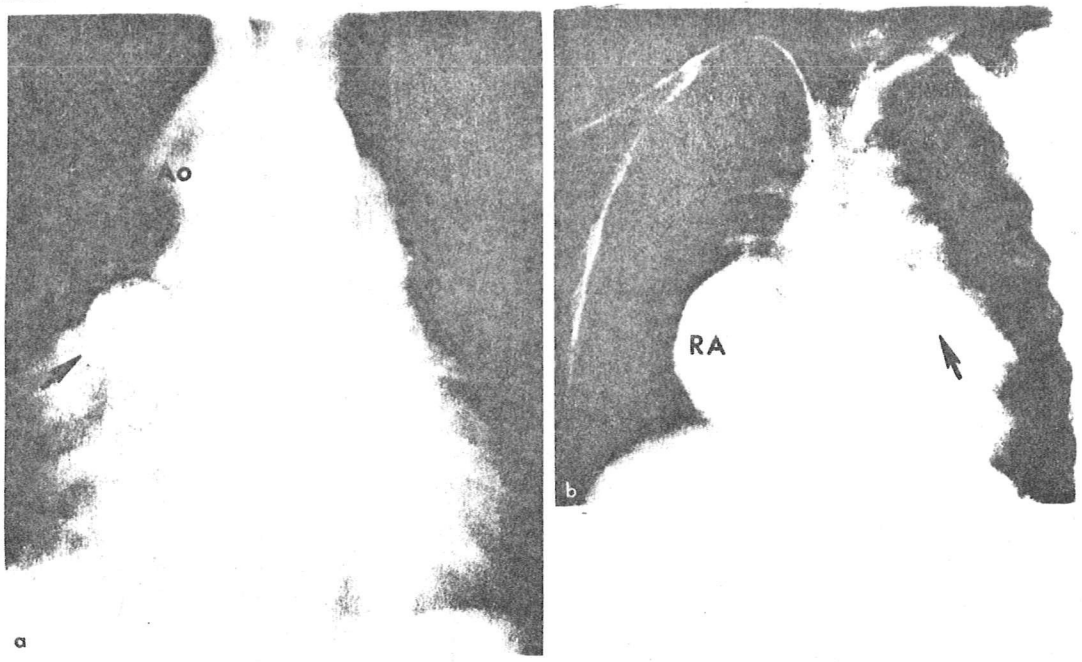
Course and Prognosis

The majority of patients die in the first year of life from congestive heart failure. Those who survive infancy may remain free of major symptoms during childhood only to experience sudden death probably secondary to an arrhythmia as young adults. Occasionally, ventricular aneurysms also develop; systemic embolic disease from an aneurysm may also occur.

Medical treatment is directed at the complications, i.e. congestive heart failure, myocardial infarction and ventricular arrhythmias. The treatment of choice is surgical and consists of either transplantation of the left coronary artery from its pulmonary origin to the aorta or a bypass graft from the aorta to the main left coronary artery. This procedure has been successful in older children, but the results have not been terribly good in very small infants. Among the palliative procedures that have been used, the most successful has been ligation of the main left coronary artery at its origin from the pulmonary artery. This procedure is applicable only in individuals where there is evidence of a sizable left to right shunt from the right coronary artery through the left coronary artery into the pulmonary artery.

Anomalous Communications of the Coronary Arteries

In this type of anomaly, the coronary arteries arise from the aorta, but one (or rarely both) communicate distally (without passing through a capillary bed) with the right atrium or ventricle, left atrium or ventricle, pulmonary artery, coronary sinus, or directly into a cardiac vein. Most often the communication is into the right heart or pulmonary artery (Fig. 31); only rarely does the fistula enter the left heart. Saccular aneurysms of the coronary arteries are frequently present. Coronary fistulae are almost always of congenital origin and occasional examples of posttraumatic acquired coronary fistulae have been reported.



a, Aortogram of a 23 year old woman with a coronary arteriovenous fistula from the right coronary artery to the right atrium. An arrow points to the dilated, tortuous, elongated right coronary artery. The left to right shunt was 1.4 to 1 (Ao = ascending aorta.)

b, Selective left coronary artery angiocardiogram in an eight year old boy. A large coronary arteriovenous fistula communicates with the coronary sinus. The dilated coronary artery (arrow) narrows as it joins the coronary sinus, which then drains into the opacified right atrium (RA).

Figure 31

These communications result in a shunt at the precapillary level which deprives the myocardium of its normal blood flow. Variations in the pathophysiology depend on the location of the fistula and the volume of the shunt. Factors of importance in regard to flow through the fistula are its size and the potential temporary occlusion or diminution in the lumen size during contraction of the receiving chamber. If the fistula opens into either the atrial cavity or pulmonary artery, the left to right shunt is greater during ventricular systole since the pressure gradient between the aorta and atria is greater during systole. This can be appreciated clinically by the prominence of the systolic murmur. If the fistula communicates with the right ventricle, greater flow occurs during diastole. This results from a reduction in the size of the fistula during ventricular contraction. In this situation, the diastolic murmur is the more pronounced. The shunt through the fistula is generally small in any event, the volume overload moderate, and the intracardiac pressure normal.

History

Males and females are involved equally with this abnormality. Most patients live asymptomatically for many years and survival into the eighth decade has been reported. Cardiac symptoms may, however, occur at any age and when present include angina, fatigue, dyspnea and sometimes evidence of heart failure. The physical examination ordinarily demonstrates a continuous murmur. It is loud and superficial and often accompanied by a thrill. The location of the murmur depends upon the chamber or vessel that receives the fistulous communication. In the rare circumstance in which the receiving chamber is the left ventricle, physical examination will reveal peripheral signs of aortic regurgitation, a wide pulse pressure and cardiac enlargement.

The electrocardiogram is not specific and in many instances is normal. Right or left ventricular hypertrophy may be noted depending on the site of the fistulous communication. Rarely, evidence of myocardial ischemia is present electrocardiographically.

The chest x-ray is similar to that noted in other forms of left-to-right shunts. If the shunt is small, the chest x-ray is normal.

At cardiac catheterization a small left to right shunt is evident by oxygen saturations if the fistulae enters the right heart or pulmonary artery. Angiography with selective injection into the aortic root or into the coronaries is usually diagnostic. It will show the location and course of the dilated coronary vessel and the chamber into which it empties.

Associated congenital cardiac anomalies are rare, although pulmonic stenosis, atrial septal defects and patent ductus arteriosus have all been described.

Course and Prognosis

These patients are often free of complications and the lesion is compatable with survival into adult life. Prognosis for the individual patient, however, is solely dependent on the volume of the shunt. Complications, as noted earlier, include myocardial ischemia, bacterial endarteritis, pulmonary hypertension, and congestive heart failure.

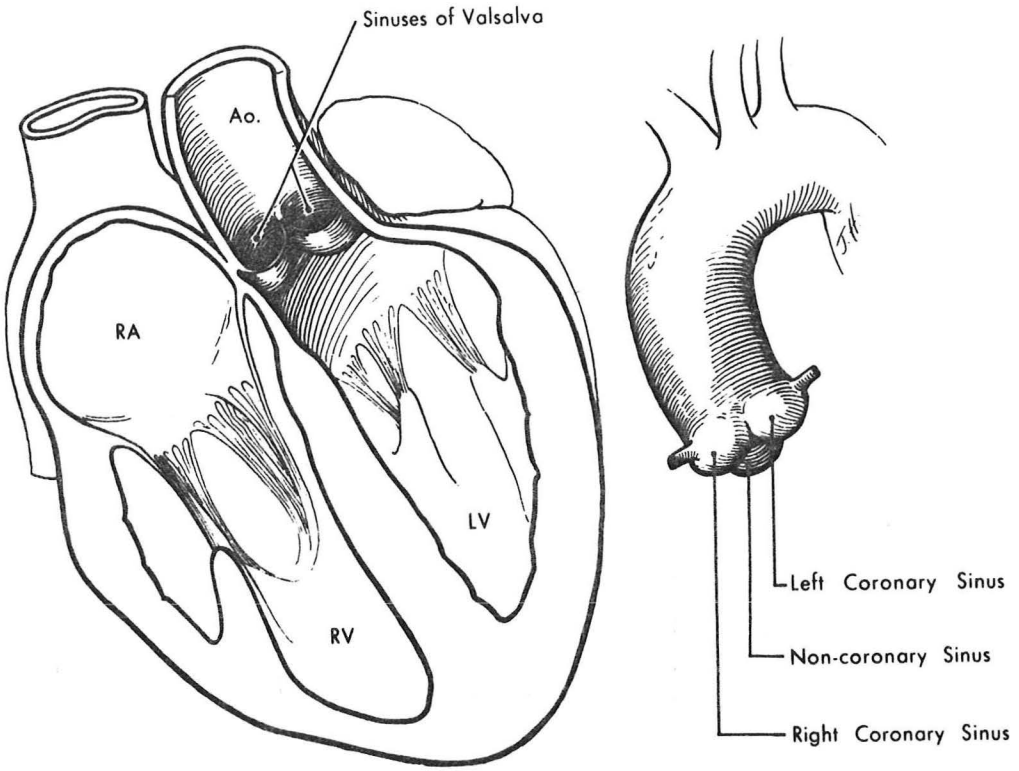
Medical treatment is directed toward complications including congestive heart failure and bacterial endarteritis. Medical measures to relieve anginal pain are also utilized. If there is a significant left to right shunt, surgical correction should be advised and the fistula should be closed so as to maintain the myocardial blood supply.

Sinus of Valsalva Aneurysms

Aneurysms of the sinuses of Valsalva may be either acquired or congenital. In this discussion we will deal with the congenital type although the physiologic consequences are equally applicable to the acquired form. Congenital aneurysm of the sinuses of Valsalva results from a weakness of the wall of the aorta at its base. In the congenital form, aneurysms usually involve the right coronary sinus and to a lesser extent the noncoronary sinus (Figs. 32,33 & 34). Only rarely is the left coronary sinus or more than one sinus involved. This is in contrast to the acquired form where there is equal predilection for involvement of all of the sinuses.

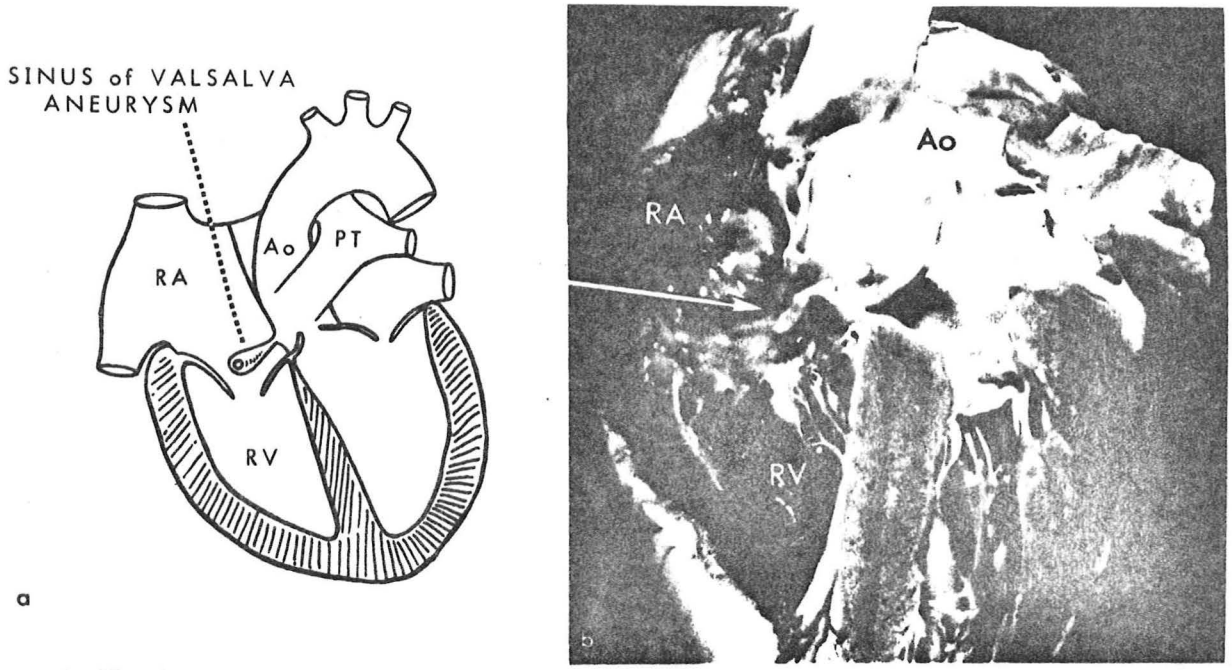
The congenital weakness in the aortic wall gradually becomes thin and distended due to the continuing stress of left ventricular ejection. Over a matter of time a true aneurysm develops, but its existence may go undected for many years due to the absence of signs or symptoms of cardiac embarrassment. The life threatening complication of this entity is rupture of the aneurysm into a cardiac chamber. This most commonly occurs after the second decade of life though it has been described during early childhood.

The congenital type of aneurysm usually ruptures into the right ventricle, less frequently into the right atrium, and only rarely into the left heart chambers. As a rule the fistula empties into only one chamber. Acquired aneurysms are usually larger and frequently rupture outside the cardiac chambers in contradistinction to the congenital ones which are almost always small and remain intracardiac. With rupture of a congenital aneurysm into a right heart chamber, there is formation of an arteriovenous fistula with resulting volume overload of the right heart. The magnitude of the shunt load is dependent primarily on the size of the fistulous communication. Pulmonary blood flow is increased and thereby results in a secondary volume overload for the left side of the heart. This often precipitates congestive heart failure which can be fatal.



Schematic illustration of the aortic root showing the location of the sinuses of Valsalva.

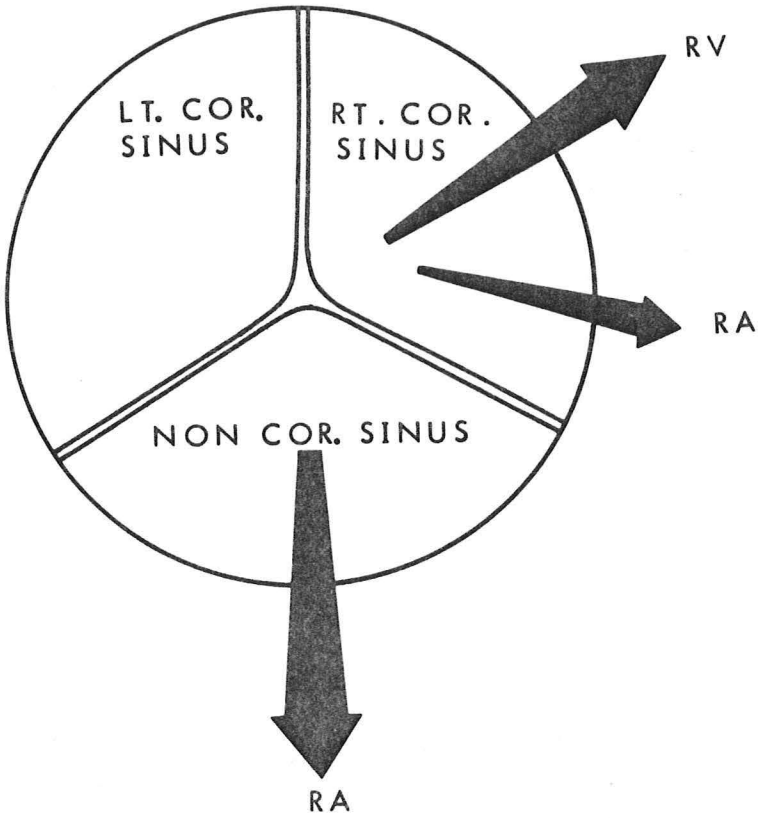
Figure 32



a, Schematic illustration of a sinus of Valsalva aneurysm projecting into the right atrium. The entire sinus is not dilated, but instead the aneurysm presents as a finger-like or nipple-like extension with a perforation at its tip. (Modified after Edwards, J. E., et al.: Congenital Heart Disease.)

b, Specimen from a 27 year old woman whose heart is cut to illustrate the essential anatomic features shown in the schematic illustration. The ruptured aortic sinus aneurysm (arrow) extends as a finger-like projection into the right atrium (RA).

Figure 33



Ninety to 95 per cent of congenital sinus of Valsalva aneurysms originate in the right or noncoronary sinus and rupture into the right ventricle or right atrium. This sketch shows the relative distribution of the sinus of origin and the chamber that receives the rupture. (Adapted from Sakakibara, S. et al.: Amer. Heart J. *63*:405, 1962.)

Figure 34

With rupture of the sinus of Valsalva aneurysm into the right heart there is a marked drop in systemic cardiac output and pressure; this form of aortic "runoff" results in a lowered systemic pressure and a widened pulse pressure. There is a sudden decrease in coronary blood flow which may approach the critical level and is probably responsible for the chest pain frequently noted following the rupture of the aneurysm. Occasionally, pain is absent and the rupture is associated with intense dyspnea. Rarely, rupture of a small aneurysm into the right heart may be associated with no significant hemodynamic alteration in cardiac function.

Clinical Features

This abnormality occurs in males in 75% of reported cases. Prior to the rupture of the aneurysm, patients are asymptomatic except in rare instances where the aneurysm is responsible for either heart block or angina pectoris due to mechanical coronary artery compression.

Most patients come to the hospital at the time of rupture with the rather typical syndrome of acute dyspnea with or without associated chest pain of sudden onset; frequently this is precipitated by exertion. Signs and symptoms of congestive heart failure may develop rapidly or there may be a latent period of a few weeks to a few months before progressive intractable congestive heart failure develops.

Individuals with a sizable left to right shunt develop bounding arterial pulses and a wide pulse pressure. Rupture into the right heart results in a large left to right shunt and right ventricular failure. There is almost always elevation of the venous pressure with prominence of both the A and the V waves in the jugular venous pulse. Palpation of the chest reveals a hyperdynamic right and sometimes left ventricular impulse with cardiomegaly. A thrill is often present along the left sternal border; this thrill may be continuous or more prominent in systole or diastole. The most striking auscultatory finding is a loud continuous superficial murmur which is similar to that of coronary arteriovenous fistula. This murmur is best heard along the lower left sternal border. Third heart sounds are ordinarily present and the pulmonary valve closure sound may also be accentuated.

The electrocardiogram may show evidence of left or right or combined ventricular hypertrophy. Intraventricular conduction distrubances and/or atrioventricular block are often seen probably due to compression of the atrioventricular node or junction by the aortic sinus aneurysm. The chest x-ray usually reveals generalized cardiomegaly and evidence of a left to right shunt. Disproportionate right atrial enlargement is seen with rupturing of an aneurysm into that chamber. Pulmonary arterial blood flow is increased depending on the volume of the left to right shunt.

Cardiac catheterization may help one establish the presence of a left to right shunt and ordinarily identifies modes elevation of pulmonary artery and right ventricular pressures. Right atrial hypertension is usual when the fistula communicates with that chamber. Retrograde aortography with injection into the aortic root is essential for establishing the diagnosis. This demonstrates dilatation of one or more aortic sinuses and a fistulous communication with one of the heart chambers. Occasionally, the catheter may pass across the fistula into a right heart chamber further verifying the diagnosis.

Associated abnormalities include ventricular or atrial septal defects and/or coarctation of the aorta. Aneurysm formation of one or more sinuses of Valsalva occurs with Marfan's syndrome.

Course and Prognosis

Aneurysms of the aortic sinus of Valsalva are a serious abnormality because of potential rupture, either spontaneously or as a result of bacterial endocarditis. Death is the rule within one year although some isolated instances of long term survival have been reported. Death is usually due to congestive heart failure.

Medical treatment is aimed at treating the congestive heart failure and/or associated infection. The preferred treatment for this lesion is prompt surgical intervention. This is best accomplished with cardiopulmonary bypass. Depending on the site of the fistulous communication, that chamber and the aorta are opened and the defect is repaired. Operative mortality figures are sparse due to the relatively small number of cases that have been performed, but successful correction of this otherwise fatal lesion is now possible.

Left Ventricular Outflow Obstruction

Obstruction to left ventricular outflow can be congenital or acquired and the location of this stenosis can be valvular, supravalvular or subvalvular. We will deal with congenital etiologies for left ventricular outflow obstruction in this discussion. The clinical assessment of left ventricular outflow obstruction should endeavor to identify the location of the obstruction as well as to estimate its severity.

A. Valvular aortic obstruction

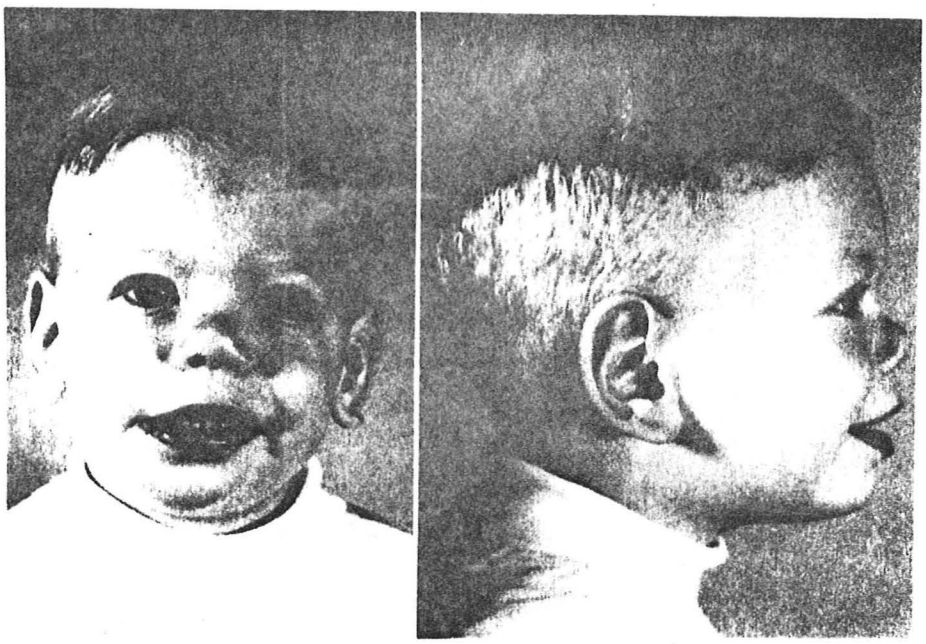
The most common type of congenital aortic stenosis is a result of narrowing of a bicuspid or a tricuspid valve. It has been estimated that one-third to one-half of patients with anatomically isolated aortic stenosis have congenitally bicuspid aortic valves. It has also been estimated that the incidence of congenitally bicuspid aortic valves may be 2% of the general population. If this assessment is correct, then bicuspid aortic valve may be the most common congenital malformation of the heart or great vessels. Bicuspid aortic valves may be functionally normal, stenotic or incompetent. A congenitally bicuspid aortic valve becomes stenotic only when its cusps are fibrotic and calcified which is usually not the condition of the valve at birth. It follows that delayed development of isolated congenital valvular aortic stenosis is likely to be due to a bicuspid valve, whereas congenital valvular aortic stenosis present at birth is likely to be due to a different abnormality namely congenital fusion of a tricuspid valve, congenital unicommissural valve and/or hypoplasia of the aortic annulus. For completeness, we should mention that congenital obstruction at the aortic valvular level may also be caused by myxoid dysplasia of the valve or stenosis of a quadracuspid valve.

B. Subvalvular aortic stenosis

Subvalvular aortic stenosis may be divided into two categories according to whether the obstruction is fixed (discrete anatomic stenosis) or variable (muscular or idiopathic hypertrophic subaortic stenosis). Fixed subvalvular stenosis is ordinarily caused by a localized circumferential fibrous collar that is present at birth and that encircles the left ventricular outflow tract and often attaches to both the ventricular septum and the anterior mitral leaflet. Rarely, discrete subaortic stenosis may be caused by hypoplasia of the left ventricular outflow tract and a so called "tunnel" deformity, by an accessory bicuspid valve or by tumor in the left ventricle. In muscular or hypertrophic subaortic stenois the obstruction is related to a hypertrophied septal muscle mass protruding into the outflow tract and laterally displacing the anterior leaflet of the mitral valve. With hypertrophic subaortic stenosis there is often an inconstant degree of obstruction. The severity of the obstruction may become greater during periods of relative volume depletion, fright, the administration of powerful inotropic pharmacologic agents, with the loss of the atrial contribution to cardiac output (i.e. atrial fibrillation) or apparently spontaneously. Idiopathic hypertrophic subaortic stenosis, IHSS, ASH (asymmetric septal hypertrophy) or DUST (disproportionate upper septal thickening) may be largely congenitally acquired although some suspicion remains that it is possible to acquire this abnormality secondary to the presence of other types of congenital heart disease.

C. Supravalvular aortic stenosis

Supravalvular aortic stenosis is usually due to a localized, segmental, hourglass-shaped narrowing immediately above the aortic sinuses. That portion of aorta immediately beyond the stenotic region is normal or reduced in size, but not dilated. Supravalvular obstruction may also be caused by a localized fibrous diaphragm or by uniform narrowing of the entire ascending aorta beginning near the origin of the coronary arteries. One interesting variation noted in some individuals with supravalvular aortic stenois is as follows. Some individuals are mentally retarded and their physical features resemble one another quite strikingly (Fig. 35). In these individuals, the chin is small (hypoplastic mandible), the mouth is large, the lips patulous, the nose blunt and upturned, the eyes wide set with occasional internal strabismus, the forehead is broad, the cheeks are baggy and the teeth are malformed. These individuals have friendly temperaments and a deep and somewhat metallic voice. Hypercalcemia may also be present. This variety of supravalvular aortic stenosis is ordinarily nonfamilial. The other variety of supravalvular aortic stenosis can be sporadic or familial, has a normal physical appearance and no hypercalcemia. It should be emphasized that supravalvular aortic stenosis occasionally occurs in patients with Marfan's syndrome and in this circumstance aortic valvular incompetence is expected. Supravalvular aortic stenosis may also coexist with branch stenosis of the pulmonary arteries and has been described with coarctation of the aorta and with the rubella syndrome.



Facial appearance of a two year old boy with supravalvular aortic stenosis, bilateral stenosis of the pulmonary arteries, mental retardation and infantile hypercalcemia. The chin is small, the mouth large, the lips patulous, the nose blunt and upturned, the eyes wide-set, the forehead broad, the cheeks baggy, and the teeth malformed. (See also Figure 6-8.) (Perloff, J. K.: Progr. Cardiov. Dis. *10*:323, 1968.)

Figure 35

History

Congenital aortic stenosis is more common in males that in females with a sex ratio of approximately 4 or 5:1. Ordinarily a murmur is detected during the first few days of life and the mother is likely to be told that her infant has congenital heart disease early on. It is important to keep in mind, however, that normally functioning, congenitally bicuspid aortic valves are rarely detected in childhood, but may subsequently generate prominent murmurs as they become stenotic later in life. The presence of coarctation of the aorta also raises concern that the same individual may also have a bicuspid aortic valve. Delayed development of a murmur is also common in patients with hypertrophic subaortic stenosis; the average age of detection for this abnormality is approximately 15 years after birth.

Individuals that develop significant left ventricular outflow obstruction often complain of angina, syncope, (either with effort or following effort, depending on the location of the obstruction), signs

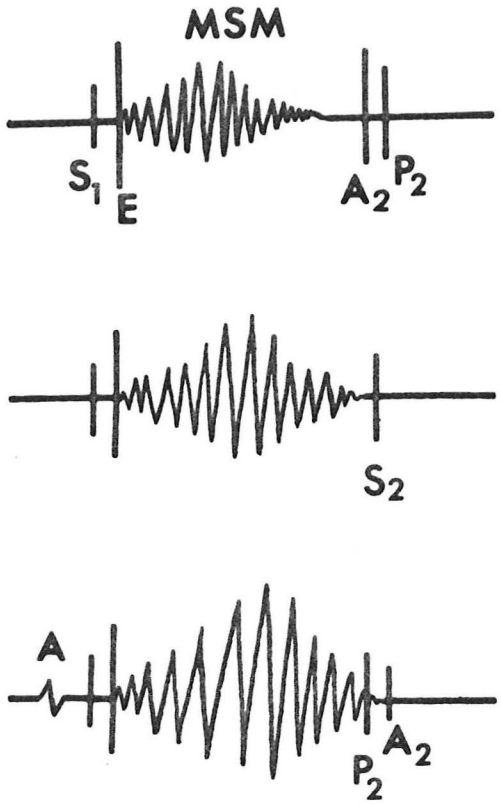
and symptoms of congestive heart failure; they also have systolic ejection murmurs. Death during vigorus exercise may occur, but is almost certainly less frequent than some of the older literature might indicate. *I wish to emphasize that cerebral symptoms can be very subtle and may consist of nothing more than mild light-headedness, particularly with effort.*

Physical Examination

A. Bicuspid aortic valves or obstruction at the level of the aortic valve -- one expects individuals with this entity to have a systolic ejection murmur (Fig. 36). The murmur may be associated with a thrill that is present at the base of the heart and radiates toward the right shoulder and right neck. This thrill may also be felt in the suprasternal space. A systolic ejection click is ordinarily present in patients with bicuspid aortic valves that are not heavily calcified. This click does not change its intensity or its relationship to the first heart sound with any recognized physiological or pharmacological maneuvers. The systolic ejection murmur typically radiates well into the carotid and may be heard in the back. A fourth heart sound is expected and a third heart sound may be present if left ventricular failure coexists. Diastolic decrescendo murmurs suggesting aortic regurgitation exist in some patients with bicuspid aortic valves either in combination with a systolic ejection murmur or sometimes alone. The carotid upstroke is reduced and the volume of the carotid is also reduced in patients with hemodynamically important valvular aortic stenosis once adulthood is reached. In addition, there may be a thrill and shudder over the carotid vessel. The second heart sound may split normally, may be paradoxically split or may be single (a very soft aortic component not being audible) in patients with hemodynamically important valvular aortic stenosis. One expects the intensity of left ventricular outflow obstruction murmurs to become louder in the beat following a ventricular premature beat, but this sign does not distinguish between obstruction located at the valvular, subvalvular or supravalvular levels.

B. Subvalvular aortic stenosis is ordinarily due to a bar, diaphragm or fixed collar. With this form of subvalvular obstruction aortic regurgitation commonly coexists and the typical murmur is noted. The same systolic ejection murmur described for valvular aortic stenosis is also present (Fig. 36), but with subvalvular aortic obstruction ejection clicks are generally not present. The other findings may be essentially the same as described above for valvular aortic obstruction.

Dynamic left ventricular outflow obstruction resulting from idiopathic hypertrophic subaortic stenosis also generates a systolic ejection murmur (Fig. 37). This murmur typically does not radiate well into the carotids, but may be heard well along the left sternal border and at the base of the heart. Individuals with IHSS also often have a murmur of mitral regurgitation and typically have third and fourth heart sounds.



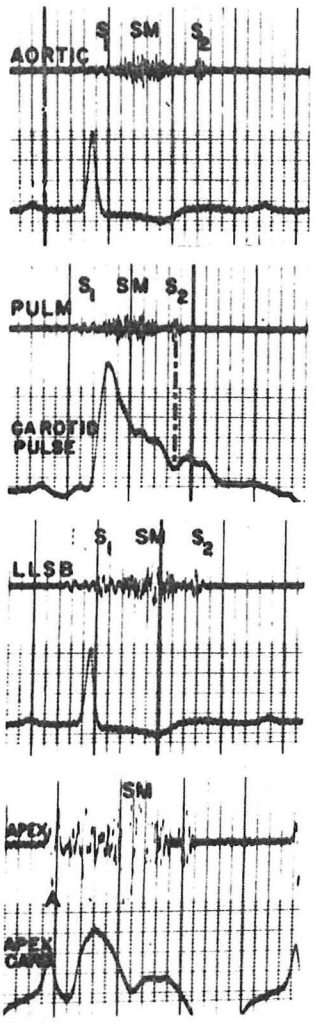
Schematic illustration of the auscultatory signs of mild, moderate and severe congenital valvular aortic stenosis.

Mild: An ejection sound introduces a rather short midsystolic murmur that peaks relatively early in systole. The second sound splits normally and aortic closure (A_2) is prominent.

Moderate: The ejection sound introduces a longer midsystolic murmur with a later systolic peak. The second sound (S_2) is single.

Severe: An atrial sound (A) is now present. The ejection sound introduces a long midsystolic murmur with a late systolic peak. The second heart sound is paradoxically split (A_2 follows P_2).

Figure 36



Tracings from a 21 year old man with hypertrophic subaortic stenosis. The midsystolic murmur (SM) is comparatively soft at the base, intermediate at the lower left sternal border (LLSB) and maximal at the apex. An atrial sound (A) is especially prominent over the left ventricle where the apex cardiogram records a prominent presystolic impulse. The second heart sound is single. The carotid pulse has a brisk upstroke and a single unsustained peak.

Figure 37

The second hear sound is often paradoxically split. Most patients with IHSS increase the intensity of their systolic ejection murmur with physiological or pharmacological maneuvers that result in 1) increased force of cardiac contractility or 2) decreases in ventricular volume. Typically, sitting, standing, the administration of nitroglycerin or amyl nitrite and the Valsalva maneuver increase the intensity of the systolic ejection murmur and squatting decreases the intensity of the murmur. However, in some individuals the expected change in the murmur

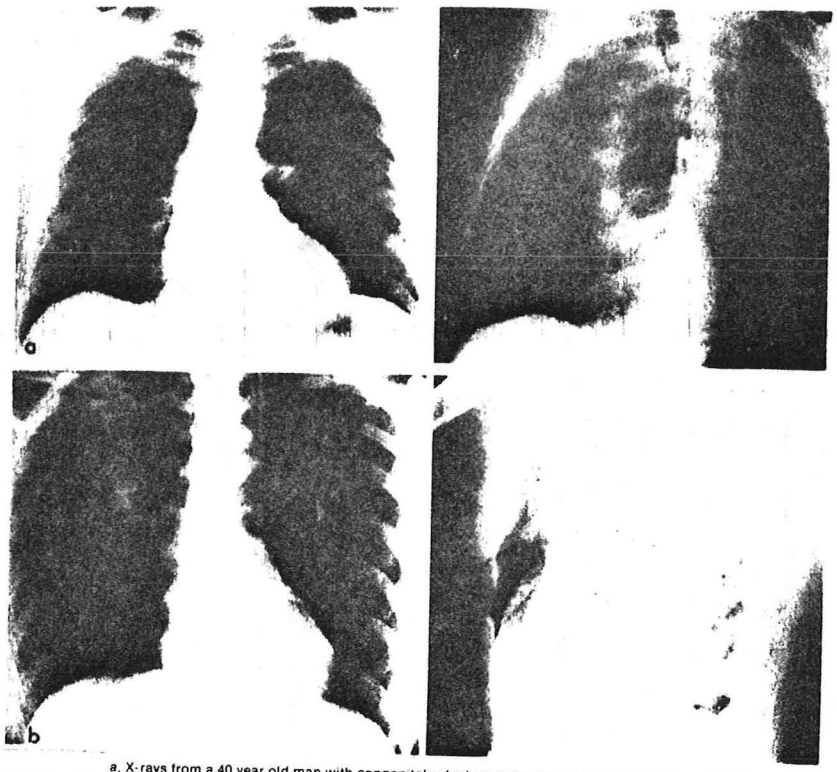
is not noted and rather it remains at the same intensity despite these maneuvers. This does not exclude the possibility of IHSS. The carotid upstroke is important in the recognition of patients with IHSS and differs from the carotid upstroke of patients with other types of subvalvular and those with valvular aortic obstruction in the sense that it is rapid rather than delayed. In addition, it may be bisferiens (but combined severe valvular aortic stenosis and aortic insufficiency may also result in a bisferiens carotid pulse).

C. Supravalvular aortic stenosis results in the same systolic ejection murmur described in patients with valvular aortic stenosis and the murmur tends to radiate into the carotid vessels. Third and fourth heart sounds are common with this entity and the splitting of the second heart sound may be normal, paradoxical or single. Systolic ejection clicks are ordinarily not heard in individuals with supravalvular aortic stenosis; the presence of aortic regurgitation is also rare. A typical finding in individuals with supravalvular aortic stenosis is a lower systolic blood pressure in the left arm than in the right; this discrepancy may be 15 mmHg or greater. The cause of the blood pressure asymmetry between the upper extremities is not entirely clear, but seems to be related to the presence of a high velocity jet upstream from the supravalvular obstruction with greater release of energy into the innominate than into the left subclavian artery.

Electrocardiographic criteria do not distinguish supravalvular from valvular or discrete subvalvular obstruction, but certain features favor the diagnosis of hypertrophic subaortic stenosis. Individuals with hypertrophic subaortic stenosis almost always have ECG evidence of left ventricular hypertrophy with strain, but this pattern may be absent in patients with important valvular, supravalvular and discrete subvalvular aortic stenosis. The echocardiogram may also be helpful in detecting the presence of IHSS by demonstrating 1) systolic anterior motion of the anterior leaflet of the mitral valve and 2) apposition of the anterior leaflet of the mitral valve and the ventricular septum. Sometimes these abnormalities are not present at rest, but can be produced with inotropic influences such as isoproterenol or the administration of agents that decrese left ventricular volume, i.e. nitroglycerin. The echocardiogram is of less help in individuals with other types of left ventricular outflow obstruction although occasionally the presence of valvular aortic stenosis can be identified, a region of marked narrowing in the proximal aorta may be detected or a discrete area of obstruction beneath the aortic valve can be suspected. Some patients with IHSS have "pseudo-infarct" patterns on their electrocardiogram consisting of significant Q waves in either the inferior or anterior leads. This pseudoinfarct pattern develops because of localized fibrosis and the extreme hypertrophy of the septal muscle mass.

The chest x-ray may or may not demonstrate evidence of left ventricular enlargement in individuals with left ventricular outflow obstruction. Some enlargement of the heart is ordinarily present in patients with IHSS, but obvious cardiac enlargement may be missing in individuals with

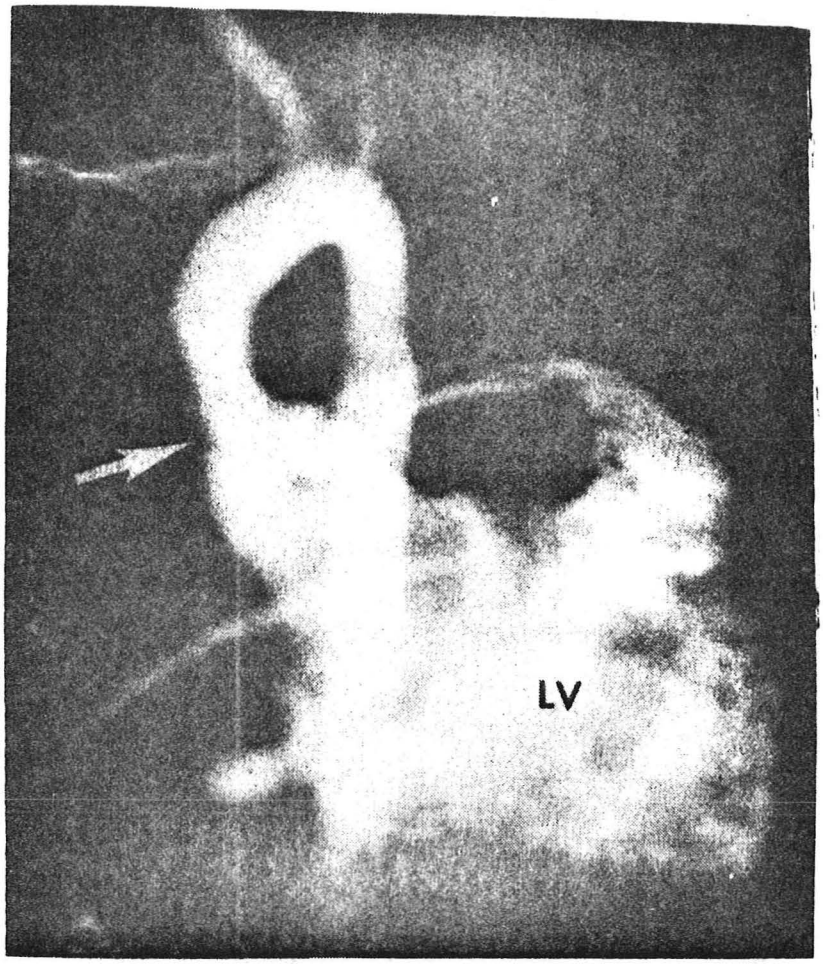
other types of left ventricular outflow obstruction. Fluoroscopy may help to identify the presence of calcification in the aortic valve or this may be obvious on the plain chest film. In general, the more severe the aortic valvular calcification, the more likely important obstruction to be found. Individuals with all of the different types of left ventricular outflow obstruction may demonstrate generalized cardiac enlargement and particularly those with IHSS may show important enlargement of the left ventricle. Poststenotic dilatation of the aorta is expected in individuals with significant valvular aortic obstruction, but ordinarily does not occur with the other types of left ventricular outflow obstruction. Identification of the presence of coarctation of the aorta from the plain chest film would also lead to suspicion of the possible presence of a bicuspid aortic valve. Figures 38,39 and 40 demonstrate additional radiographic or angiographic manifestations of the various types of left ventricular outflow obstruction.



a. X-rays from a 40 year old man with congenital valvular aortic stenosis (gradient, 95 mm. Hg). Poststenotic dilatation of the aorta is evident in both views. In the posteroanterior projection the left ventricular silhouette appears convex and elongated; in the left anterior oblique view the convex left ventricle projects over the vertebral column.

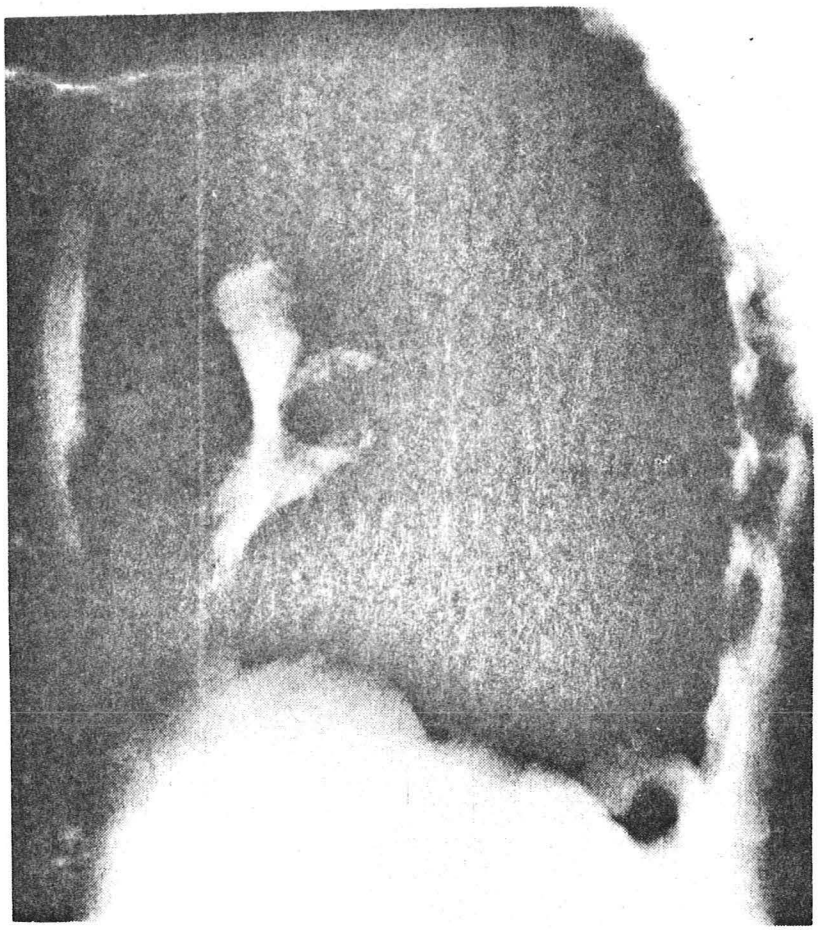
b. X-rays from a 20 year old man with congenital valvular aortic stenosis (gradient, 40 mm. Hg). In the posteroanterior projection the cardiac silhouette is entirely normal except for poststenotic dilatation of the aorta. In the lateral view poststenotic dilatation is again apparent, and the left ventricular silhouette is somewhat convex and globular.

Figure 38



Left ventricular angiocardiogram from a six month old girl with supravalvular aortic stenosis. There is an area of segmental narrowing (arrow) just above the dilated aortic sinuses. The infant also had bilateral pulmonary artery stenosis. (Courtesy of Dr. Stephen Epstein, National Heart Institute, Bethesda, Maryland.)

Figure 39



Lateral angiocardiogram from a 23 year old man with idiopathic hypertrophic subaortic stenosis. There is a striking decrease in left ventricular cavity size, with funnel-shaped narrowing of the outflow tract. The aortic valve is normal.

Figure 40

Course and Prognosis

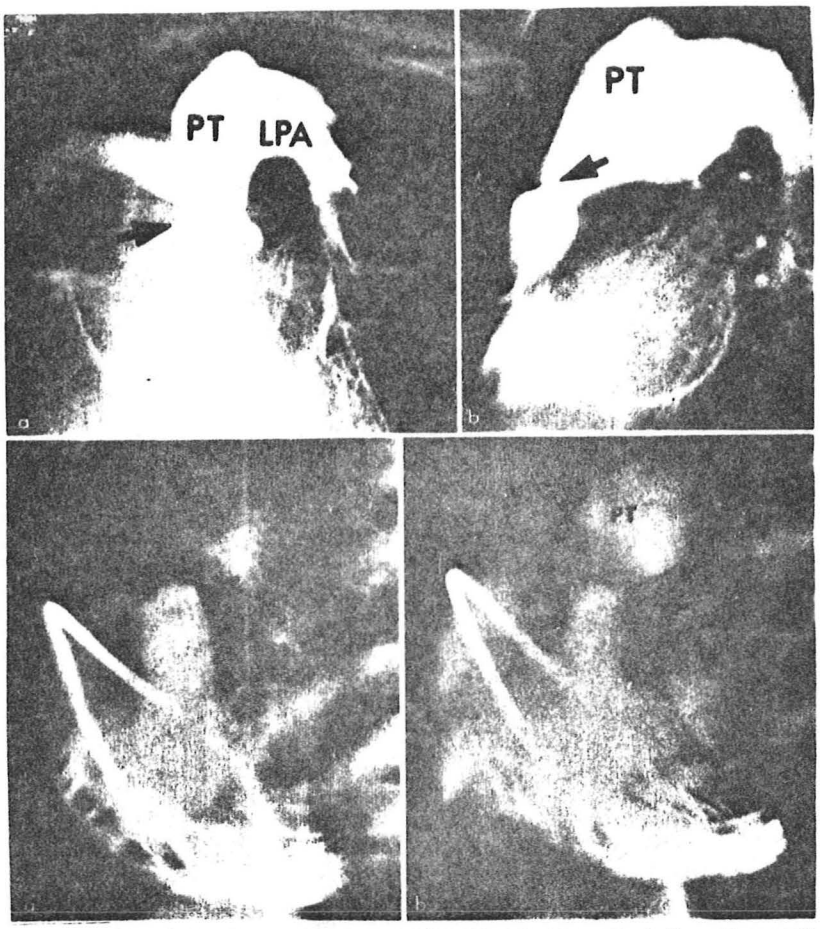
Once syncope, angina or congestive heart failure develop in individuals with left ventricular outflow obstruction the prognosis is subsequently guarded. One utilizes medical management in patients including digitalis and diuretics in the treatment of patients with valvular, supravalvular and discrete subvalvular obstruction, but once syncope, angina or severe congestive heart failure develop then surgical correction is necessary for prolonged survival. Individuals with IHSS are, of course, treated with beta adrenergic blockade (i.e. propranolol) at the maximal doses they will tolerate. This form of therapy reduces the severity of the left venticular outflow obstruction in some patients and is the preferred method of treatment. Those with severe mitral regurgitation and IHSS may require mitral valve replacement and those that do poorly on a good medical regimen of propanolol may require subsequent surgery to relieve the subvalvular obstruction; the surgical therapy is resection of the hypertrophed asymmetric septal muscle mass.

Congenital Pulmonic Stenosis

Congenital obstruction to right ventricular outflow may be valvular, subvalvular or supravalvular. Valvular pulmonic stenosis is characterized by a dome-shaped zone with a narrow outlet at its apex (Fig. 41). Three raphes extend from the small central opening into the wall of the pulmonary artery although separate leaflets may not be identified. Poststenotic dilatation of the pulmonary trunk is the rule and is often accompanied by dilatation of the left pulmonary artery. Valvular pulmonic stenosis may occur as an isolated, uncomplicated anomaly. Occasionally, the obstructive mechanism is related to markedly thickened immobile cusps characterized by disorganized myxomatous tissue.

Subvalvular pulmonic stenosis may be either infundibular or less often, subinfundibular. Poststenotic dilatation of the pulmonary artery does not occur with obstruction at subvalvular levels. Discrete infundibular stenosis is generally the result of narrowing that is localized to the entrance of the outflow tract beyond which the infundibulum may be somewhat dilated. Hypertrophy of normal infundibular muscle may also cause obstruction. Subinfundibular stenosis is a relatively rare variety of right ventricular outflow obstruction and it may be caused by hypertropy of either abnormal muscle groups or normal bulbar muscle. Subvalvular pulmonic stenosis is generally associated with ventricular septal defect, but occasionally occurs as an isolated anomaly.

Supravalvular pulmonic stenosis results from narrowing of either the pulmonary trunk, its bifurcation or its primary or peripheral branches. Occasionally membranous obstruction occurs immediately above the valve. Stenosis of the pulmonary artery and its branches may be unilateral or bilateral; such obstruction may involve a localized site or it may involve varying lengths of a branch. Supravalvular pulmonic stenosis commonly coexists with other abnormalities, especially with



Upper: Angiocardiograms with contrast material injected into the right ventricle of a 47 year old woman with severe valvular pulmonic stenosis (gradient, 106 mm. Hg; confirmed at surgery). *a*, In the posteroanterior projection the dilatation of the pulmonary trunk (PT) is not especially evident, but the left branch (LPA) is conspicuously dilated. Arrow points to the level of the stenotic valve. *b*, In the lateral projection the dome-shaped stenotic pulmonary valve (arrow) is well seen, and poststenotic dilatation of the pulmonary trunk is readily apparent.

Lower: Angiocardiograms (left anterior oblique projection) with contrast material injected into the right ventricle in a 10 month old girl with severe valvular pulmonic stenosis (right ventricular pressure well above systemic). *a*, The right ventricle and infundibulum are filled and a wisp of dye is entering the pulmonary trunk. *b*, The dome-shaped stenotic valve (arrow) is well seen, together with poststenotic dilatation of the pulmonary trunk (PT).

Figure 41

valvular pulmonic stenosis, atrial septal defect, ventricular septal defect, patent ductus arteriosus and Fallot's tetralogy. As noted earlier, occasional individuals have a combination of supravalvular aortic stenosis and bilateral branch stenosis of the pulmonary arteries. These patients may also have peculiar facies, abnormal dentition, mental retardation and infantile hypercalcemia.

The functional consequences of the various forms of pulmonic stenosis depend upon the severity of the right ventricular outflow obstruction. A systolic gradient exists across the zone of stenosis so that proximal

pressures are elevated and distal pressures are normal or reduced. Stenosis of the pulmonary artery and its branches produces hypertension in the pulmonary trunk whereas valvular or subvalvular obstruction produces hypertension in the right ventricle.

History and Clinical Signs

Valvular pulmonic stenosis occurs with equal frequency in males and females. The murmur is generally heard while the infant is in the newborn nursery. Survival into the sixth, seventh and eighth decades have been recorded. It should be emphasized, however, that with advancing age fibrous thickening and even calcium deposition may reduce valve mobility and increase the degree of obstruction. The average age at death in anatomically proved cases of important pulmonic valvular stenosis was 26 years in one series and the majority of Wood's patients were 30 years or less, but one was 67. In Perloff's series, 5 patients were 42-49 years of age at the time of death. The natural history of pulmonary artery stenosis is less well known. Familial occurrence is not a feature in most patients with valvular or infundibular pulmonic stenosis, but a family history may be found in individuals with stenosis of the pulmonary artery and its branches.

Symptoms are rare in infants and unsual in children with right ventricular outflow obstruction. Objective complaints tend to increase with age; mild pulmonic stenosis usually causes no symptoms and an appreciable number of patients with moderate to severe stenosis are also essentially asymptomatic. Even some patients with right ventricular systolic pressures as high as 180 mmHg are essentially asymptomatic. Perloff has reported a group of patients with right ventricular systolic pressures of 50-100 mmHg including a New Zealand long distance swimmer, a female athlete, an English hockey captain and a "first class" long-distance runner. These favorable reports, however, should not obscure the fact that asymptomatic individuals may occasionally deteriorate rapidly.

Once symptoms develop, dyspnea and fatigue are the most common ones. Orthopnea is ordinarily absent since pulmonary venous congestion does not occur. Initially, dyspnea and fatigue are provoked only by exercise, but as time goes by they may occur at rest. Patients with valvular or subvalvular pulmonic stenosis sometimes notice light-headedness or syncope with effort. A relatively low and fixed cardiac output appears to be responsible for these complaints. In contrast to aortic stenosis, syncope with death is very rare. Some adults with valvular or infundibular stenosis have chest pain resembling myocardial ischemia. A three and a half year old child with severe valvular pulmonic stenosis that died during an episode of apparent angina at autopsy had infarction of the hypertrophied right ventricle and interventricular septum; the coronary arteries were normal. With pulmonary artery stenosis, chest pain may also occasionally occur, but with this entity it may be due to thromboses of peripheral pulmonary arteries and does not necessarily

reflect myocardial ischemia. Recurrent hemoptysis should direct one's attention to the possiblity of pulmonary artery stenosis when right ventricular outflow obstruction is considered to be present. Post-stenotic dilatation of peripheral pulmonary artery branches may result in thin-walled aneurysms that rupture. Hemoptysis may be mild or it can be brisk in this circumstance. Patients with right ventricular outflow obstruction that is of importance, especially those with valvular and subvalvular obstruction, may be aware of markedly increased pulsations in their neck. These pulsations are large jugular venous A waves which may be particularly noticeable during effort or excitement. There is a risk of developing bacterial endocarditis in individuals with both valvular and infundibular pulmonic stenosis.

Physical Examination and Clinical Signs

Children with isolated valvular pulmonic stenosis generally appear remarkably healthy. However, it should be kept in mind that occasional individuals with severe right ventricular outflow obstruction have retarded growth and development, peripheral vasoconstriction and relative coolness of the extremities as a consequence of a markedly reduced cardiac output. Since valvular pulmonic stenosis may occur in association with Turner's syndrome one should also be aware of the fact that occasional individuals will present with the physical features of Turner's syndrome, namely small stature, shield chest and web neck (although coarctation of the aorta is a more likely cardiovascular accompaniment of Turner's syndrome). The rubella syndrome can result in physical and mental underdevelopment, cataracts and deafness which may also accompany stenosis of the pulmonic valve or the pulmonary artery and its branches (although patent ductus arteriosus is more commonly associated). Finally, valvular stenosis caused by myxomatous dysplasia may also be associated with delayed body growth and abnormal facies (triangular face with hypertelorism, low set ears and ptosis of the eyelids).

With important outflow obstruction to right ventricular ejection the jugular venous pulse is ordinarily elevated and the A wave is prominent and becomes progressively larger as the degree of right venticular outflow obstruction increases (assuming the presence of an intact ventricular septum). Exercise and excitment augment these A waves. V waves will also be noted if the tricuspid valve is incompetent and important tricuspid regurgitation develops.

One expects to feel a right ventricular impulse with important right ventricular outflow obstruction. A thrill may also be detected along the lower left sternal border. In valvular pulmonic stenosis, the thrill is maximal in the second or third left intercostal spaces with radiation upward and to the left. Infundibular pulmonic stenosis may be suspected if the thrill is maximal along the lower left sternal border, i.e. in the fourth and fifth left intercostal spaces. With stenosis of the pulmonary artery and its branches thrills are 1) less common and 2) more variable in location, but when present are often found in the

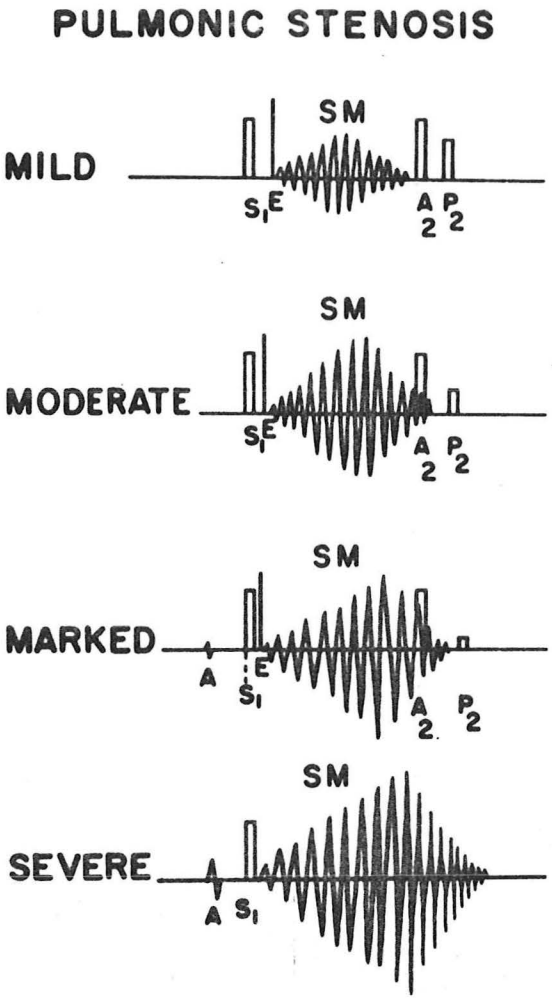
second left intercostal space. A systolic impulse over the pulmonary trunk is ordinarily absent irrespective of the level of obstruction when important right ventricular outflow obstruction exists. With subvalvular or supravalvular stenosis the main pulmonary artery is not dilated. In valvular pulmonic stenosis there is poststenotic dilatation of the pulmonary trunk, but obstruction acts to retard the rate of ejection from the right ventricle so there is little systolic expansion of the pulmonary trunk.

The important auscultatory finding with right ventricular outflow obstruction is the presence of a systolic ejection murmur audible in the same regions in which thrills are detected. A systolic ejection click is present in patients with valvular pulmonic stenosis and classically this click becomes softer or disappears altogether during inspiration. Uncomplicated subvalvular pulmonic stenosis and stenosis of the pulmonary artery and its branches are rarely associated with ejection sounds. An ejection sound is ordinarily absent in pulmonic valvular stenosis caused by myxomatous dysplasia of the valve. The sounds produced by valvular pulmonic stenosis are shown in Figure 42. With valvular pulmonic stenosis the timing of the ejection sound is of some help in estimating severity; the interval between the first heart sound and the pulmonic ejection click tends to vary inversely with the degree of obstruction. The timing and duration of the systolic ejection murmur is also of help in estimating the severity of valvular pulmonic stenosis since with increasing severity the systolic ejection murmur becomes longer and peaks later in systole.

Additional Signs

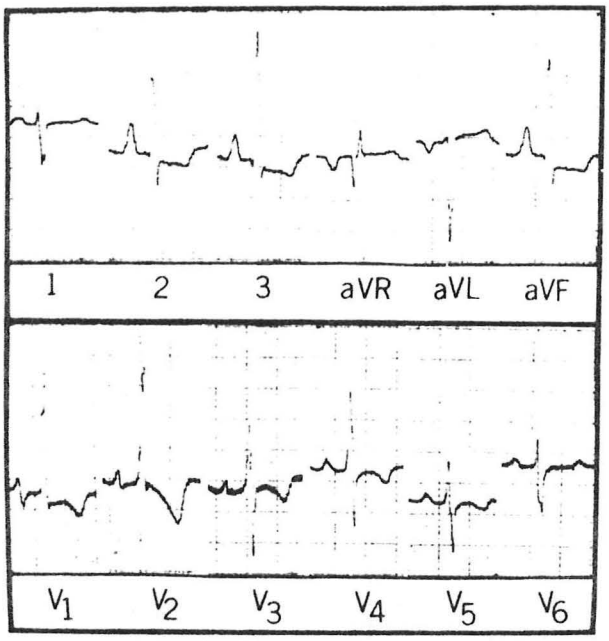
The electrocardiogram is helpful in estimating severity of the right ventricular outflow obstruction. Normal electrocardiograms are found in patients with mild pulmonic stenosis. Few individuals with right ventricular pressures exceeding 100 mmHg still have normal electrocardiograms. In general, important right ventricular outflow obstruction is characterized by the presence of right axis deviation, right ventricular hypertrophy and sometimes right atrial enlargement on the electrocardiogram (Fig. 43). Sinus rhythm is the rule although occasionally atrial fibrillation is present.

The chest x-ray may demonstrate helpful signs that allow one to localize the level of right ventricular valve flow obstruction. The pulmonary vascular pattern is normal even in the presence of severe valvular or infundibular stenosis until the output of the right ventricle falls. When severe obstruction is accompanied by right ventricular failure, pulmonary blood flow declines and the pulmonary vascular markings become correspondingly reduced. Classical pulmonic valvular obstruction is associated with poststenotic dilatation of the main pulmonary trunk (Fig. 44). Stenosis of the pulmonary artery and its branches may be associated with zones of segmental hypovascularity which correspond to the location of the areas of arterial obstruction. In addition, multiple



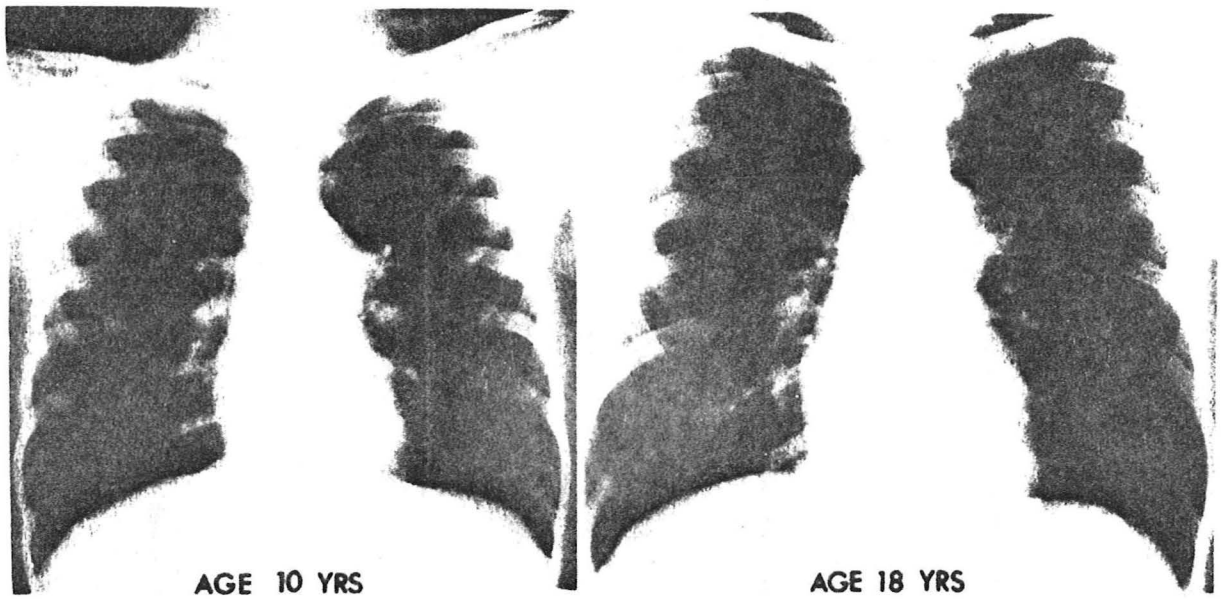
Schematic illustration of phonocardiograms in mild to severe valvular pulmonic stenosis. S_1 = first heart sound; E = ejection sound; SM = systolic murmur; A_2 = aortic component of the second heart sound; P_2 = pulmonic component of the second heart sound; A = atrial sound. (Modified from Vogelpoel, L., and Schrire, L.: Circulation *22*:55, 1960, and Perloff, J. K.: *In* Segal, B. L. (ed.): The Theory and Practice of Auscultation. Philadelphia. F. A. Davis Co., 1964.)

Figure 42



Electrocardiogram from a 43 year old woman with severe valvular pulmonic stenosis (gradient, 135 mm. Hg). There are tall peaked P waves of right atrial enlargement, especially in leads 2, 3 and aVF. Right ventricular hypertrophy is manifested by right axis deviation, tall monophasic R waves in right precordial leads and deep S waves in left precordial leads. In lead V_1 the height of the R wave alone is evidence that the right ventricular systolic pressure exceeds systemic. The T wave axis has shifted to the left, superior and posterior so T wave inversions appear in leads 2, 3 and aVF and extend beyond V_2. (Diagnosis confirmed at surgery.)

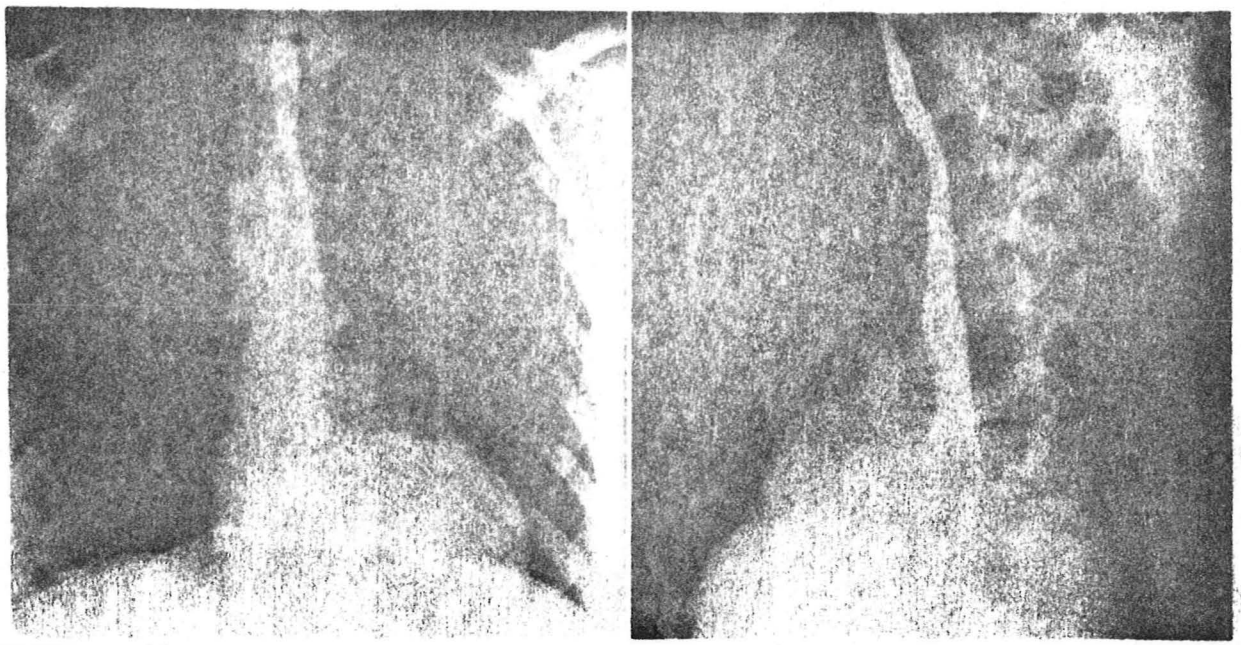
Figure 43



Virtually identical x-rays at ages 10 and 18 years in a boy with mild valvular pulmonic stenosis (gradient, 20 to 25 mm. Hg at both ages). The lung fields show normal pulmonary vascular markings. There is conspicuous dilatation of the pulmonary trunk and its left branch.

Figure 44

areas of poststenotic dilatation of the peripheral pulmonary arteries can sometimes be identified in the lung fields and are useful signs of peripheral pulmonary artery stenosis. As noted earlier, poststenotic dilatation of the main pulmonary trunk does not ordinarily occur with infundibular obstruction (Fig. 45) nor does it occur with stenosis of the pulmonary artery or its peripheral branches. Calcification of a congenitally stenosed pulmonary valve is unusual although occasional examples have been reported. A right aortic arch coexisting with right ventricular outflow obstruction often means that the obstruction is infundibular; in valvular pulmonic stenosis the arch is ordinarily left sided with few exceptions. Right ventricular enlargement and sometimes right atrial enlargement with evidence of systemic venous engorgement may also be detected in patients with right ventricular outflow obstruction.



X-rays from a 22 year old woman with isolated infundibular pulmonic stenosis (gradient, 38 mm. Hg). Of special interest is the right aortic arch which, in the frontal view, indents the barium-filled esophagus on the right and descends along the right side of the vertebral column. In the left anterior oblique projection the right arch causes a localized posterior displacement of the esophagus.

Figure 45

Medical treatment is utilized in those individuals with important right ventricular outflow obstruction and evidence of right ventricular failure. However, the development of major symptoms related to severe right ventricular outflow obstruction identifies the need for valvulotomy or resection of the infundibular obstruction. Generally speaking, this operative procedure is curative and can be accomplished with minimal risk.

Eisenmenger's Syndrome

The term "Eisenmenger's syndrome" refers to that situation in which there is a large communication between the right and left sides of the heart with pulmonary arterial hypertension at systemic levels. The marked increase in pulmonary artery pressure develops as a consequence of severely increased pulmonary vascular resistance which itself is equal to or greater than 75% of that present in the systemic arterial bed. This physiologic state results in a reversal of a previous left-to-right shunt through the communication so that it now becomes right to left (although it should also be admitted that occasionally bidirectional shunting or apparently delicately balanced shunting occurs).

The development of obliterative pulmonary vascular disease in patients with large left to right shunts (ordinarily through a ventricular septal defect or aorticopulmonary communication or as reviewed earlier less commonly through an atrial septal defect) is responsible for this phenomenon. The development of significant pulmonary vascular obstruction is related to the size of the communication and the reactivity of the pulmonary vascular bed to increased blood flow and pressure. Progression of the pulmonary vascular obstructive disease may be a consequence of a longstanding large left-to-right shunt although considerable evidence suggests that the pulmonary arteriolar changes may also develop in infancy or early childhood. Characteristic changes in the pulmonary arteriolar bed include medial hypertrophy, intimal thickening and prominence of the elastic membrane; these changes result in marked luminal narrowing and eventual obliteration with thrombosis *in situ*. As stressed previously, the development of Eisenmenger's syndrome is a rare occurrence in infants and children with large defects in the atrial septum, but not rare in those with ventricular septal defects or aortocopulmonary communications. It is not entirely clear why this difference should exist, but it has been suggested that the lesser compliance of the thicker walled right ventricle and the subsequent resulting resistance to diastolic filling may help to control the magnitude of the left to right shunt for similar size defects.

History

Females predominate over males with this syndrome. Symptoms consist of easy fatiguability, effort breathlessness, syncope, hemoptysis and congestive heart failure. Some also notice chest pain that sounds as if it is angina.

Physical Examination

On physical examination, central cyanosis often associated with clubbing is present. The presence of differential cyanosis, i.e. blue, clubbed toenails with pink lips and fingernails is characteristic of a patent ductus arteriosus and severe pulmonary hypertension.

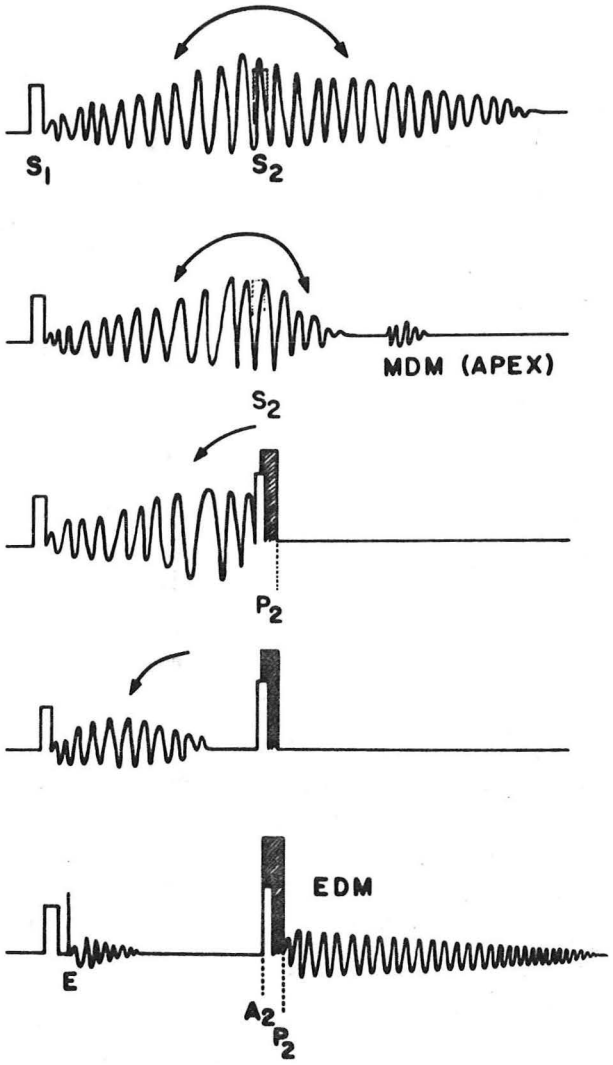
Jugular venous pressure is ordinarily elevated in patients with Eisenmenger's syndrome and a prominent A wave is noted in those with atrial septal defects, but not in those with large ventricular septal defects. A right ventricular impulse and a palpable pulmonary artery impulse are expected and the pulmonic valvular closure sound is markedly accentuated. The second heart sound usually ssems to be single and loud (very narrow split with marked pulmonic closure accentuation). Persistent narrow splitting of the second heart sound with pulmonic valve closure accentuation has been noted in some patients with Eisenmenger's syndrome and atrial septal defects and some with patent ductus arteriosus.

A systolic ejection click is often heard; this sound does not ordinarily diminish significantly with inspiration; the sound originates within the dilated main pulmonary artery trunk. The third heart sound is frequently audible and the fourth sound may be present; in many instances these sounds clearly increase with inspiration suggesting a right ventricular etiology for them. A systolic murmur along the left sternal border that is holosystolic and increases with inspiration allows one to identify tricuspid regurgitation itself a consequence of right ventricular failure. A high-pitched decrescendo diastolic murmur representing pulmonary valvular incompetence is heard in approximately 50% of patients with this entity (Fig. 46).

Laboratory Studies

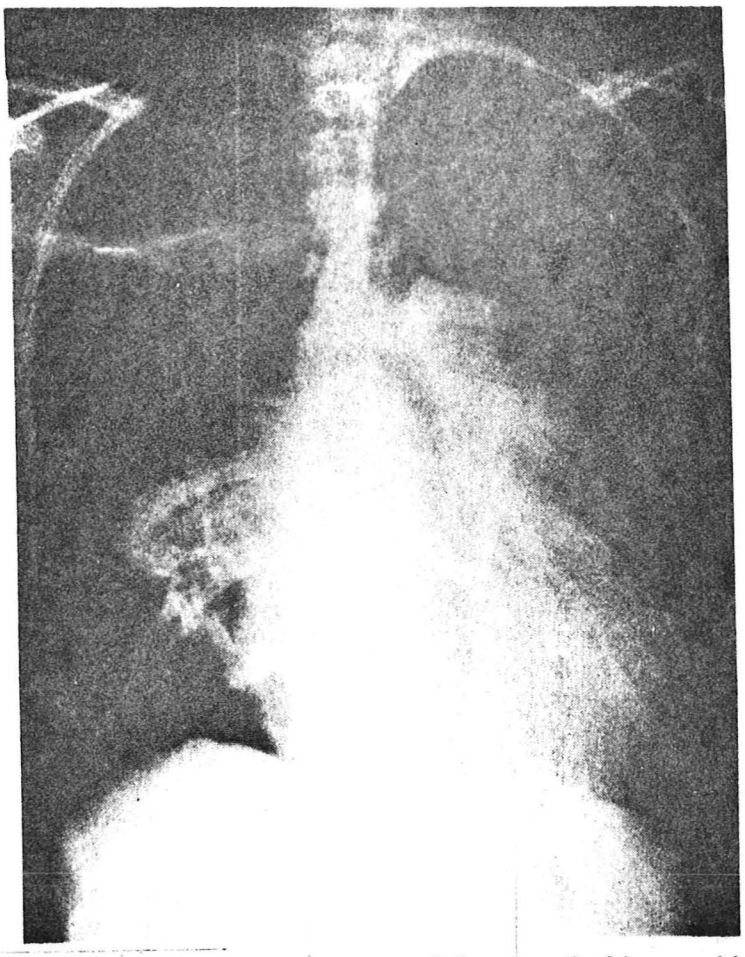
The electrocardiogram ordinarily demonstrates evidence of right axis deviation, right ventricular hypertrophy and right atrial enlargement. The rhythm may be sinus or supraventricular rhythm disturbances including importantly atrial flutter, atrial fibrillation or atrial tachycardia may be present.

The chest x-ray should provide additional important information regarding the presence of Eisenmenger's syndrome. The x-ray ordinarily demonstrates the presence of prominent enlargement of the main pulmonary trunk and of the proximal right and left pulmonary arteries (Figs. 47&48). Enlargement of the right atrium and right ventricle are also commonly present. Typically, there is rapid tapering of the peripheral pulmonary arteries so that the marked proximal enlargement stands in sharp contrast to the tapered peripheral pulmonary arteries and apparent relative oligemia of peripheral lung field arterial supply.



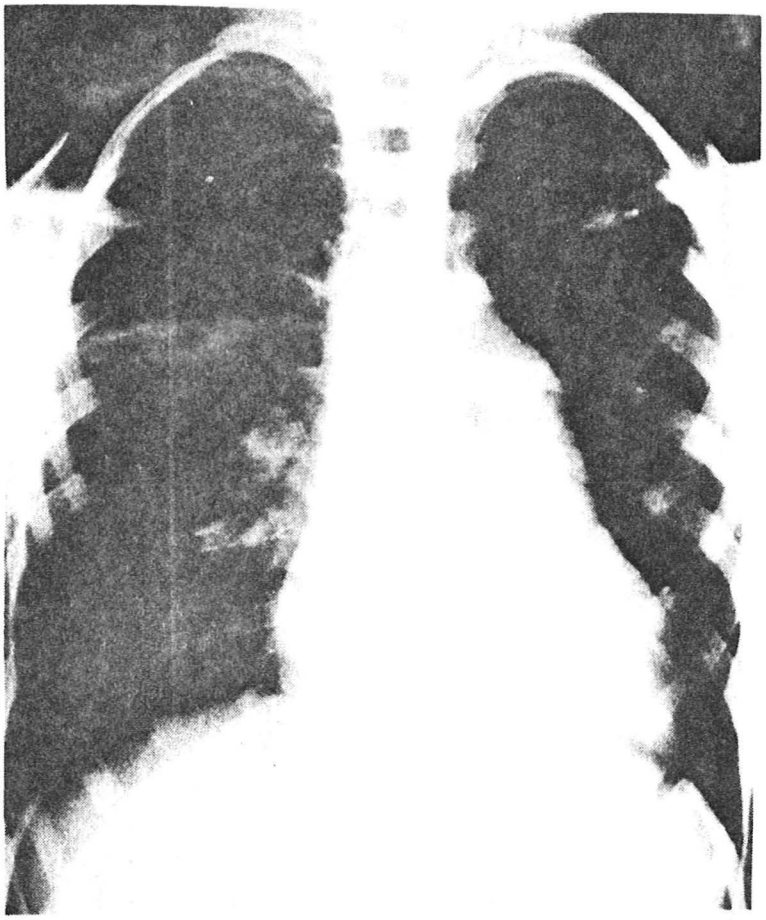
Schematic illustration of the sequential modifications of the ductus murmur from nonpulmonary hypertensive left to right shunt (above) to pulmonary hypertensive right to left shunt (below). MDM = middiastolic murmur across the mitral valve; EDM = early diastolic Graham Steell murmur. (Perloff, J. K.: Progr. Cardiov. Dis. 9:303, 1967.)

Figure 46



X-ray from a mildly cyanotic 64 year old woman with a pulmonary hypertensive ostium secundum atrial septal defect and a small persistent left to right shunt. The lung fields are clear although the film is overpenetrated. The pulmonary trunk and its right branch are aneurysmal and the right branch contains calcium. In addition, the large left branch can be seen behind the pulmonary trunk. The ascending aorta is inconspicuous. A right atrial shadow is seen to the right of the vertebral column and the dilated right ventricle occupies the cardiac apex.

Figure 47



X-ray from a 14 year old boy with a relatively large ventricular septal defect, a high pulmonary vascular resistance and a persistent left to right shunt (1.6 to 1). The pulmonary arterial pressure was 82/25 mm. Hg. There is a decrease in the prominence of peripheral intrapulmonary arteries ("pruning effect") so that the middle and outer thirds of the lung fields are clear. The pulmonary trunk and hilar vascularity remain prominent, and a dilated left ventricle occupies the cardiac apex. The ascending aorta is inconspicuous.

Figure 48

It should be emphasized that once severe pulmonary hypertension develops, the characteristic murmur or murmurs that previously identified a left to right shunt and allowed one to localize the site are no longer present (Fig. 46). As a large left to right shunt becomes bidirectional or balanced or predominantly right to left the characteristic murmur disappears. In fact, as pulmonary hypertension becomes progressively more severe there is a corresponding decrease in the length of the systolic murmur which identifies the presence of a left to

right shunt so that it becomes much shorter and less impressive. This murmur, as reviewed earlier, may ultimately be replaced by a murmur of functional tricuspid regurgitation and/or by the mumur of pulmonary valvular incompetence. A short systolic ejection murmur is to be expected in individuals that continue to shunt left to right with prominent pulmonary hypertension and it results from ejection into the dilated pulmonary trunk.

At cardiac catheterization the diagnosis is made by documenting marked increases in pulmonary artery pressure and vascular resistance to levels of 75% or more of systemic values. One also expects to find systemic arterial unsaturation and polycythemia. Left atrial pressure or pulmonary capillary wedge pressure is ordinarily normal. Shunt calculations should demonstrate a right to left or bidirectional shunt and it may also be possible to pass a catheter through a patent ductus arteriosus or through an atrial septal defect further defining the presence of these lesions. It is more difficult to pass a catheter through a ventricular septal defect in this circumstance. Selective angiography will also demonstrate the site of the defect as well as the characteristic changes in the pulmonary arterial vascular system.

Course and Prognosis

Many children survive to reach young adulthood with this syndrome. However, life expectancy with Eisenmenger's syndrome is severely shortened and most patients do not survive past the fourth decade of life. Causes of death include congestive heart failure, brain abscess, pulmonary infection, pulmonary thromboses and infarction, bacterial endocarditis, sudden death related to a ventricular arrhythmia and/or severe hemoptysis with death resulting from exsanguination. Pregnancy carries a high risk of mortality in patients with the Eisenmenger syndrome. Death following syncopal attacks with diminished cardiac output and coronary blood flow may occur about the time of delivery or during the puerperium in females with Eisenmenger's syndrome. Even patients that were previously relatively asymptomatic prior to pregnancy with this syndrome are at severe risk.

Medical therapy is directed toward prevention and treatment of the complications. Digitalis, diuretics and bed rest should be utilized and phlebotomy with appropriate volume replacement is utilized to treat significant polycythemia. Pregnancy should be avoided. Surgery is contraindicated to correct the shunt once pulmonary artery hypertension and pulmonary vascular resistance equal 75% or more of systemic vascular resistance. Attempts at surgical repair of a septal defect or aorticopulmonary communication in this setting carry a prohibitive mortality and in the rare instance of survival there has been no significant diminution in the degree of pulmonary vascular obstructive disease.

Differential Diagnosis

At least three other syndromes need to be briefly mentioned in conjunction with the differential diagnosis of apparent Eisenmenger's syndrome. These are:

1. Primary pulmonary hypertension

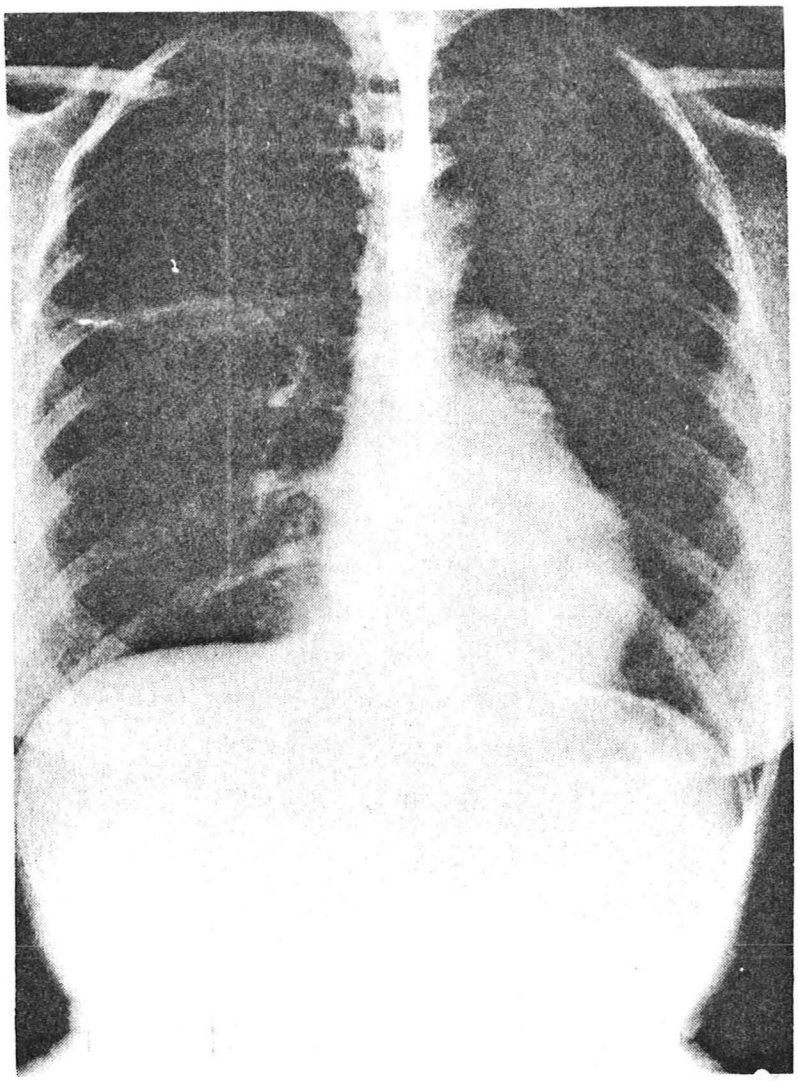
Primary pulmonary vascular disease is responsible for this entity. The term "primary" is used to designate a distinctive syndrome resulting from intrinsic, idiopathic obstructive disease in the small terminal arteries and arterioles of the pulmonary vascular bed. The etiology of primary pulmonary hypertension is unknown and opinion differs regarding potential mechanisms. The fundamental defect orginates in terminal precapillary vessels - the arterioles and smallest muscular arteries. The microscopic abnormalities that have been noted include medial hypertrophy, cellular intimal proliferation, cellular fibrous vascular occlusion, dilatation lesions and necrotizing arteritis. Microscopic in situ thrombi also occur. Irrespective of how pulmonary hypertension is initiated, it tends to reinforce itself by resulting in a sequence of changes that progressively increase resistance to flow through the lungs and thus, "pulmonary hypertension begets further pulmonary hypertension". As is true in Eisenmenger's syndrome, pulmonary arterial pressure and vascular resistance can rise to levels equal to or above those noted in the systemic circuit.

The consequences are essentially the same as those of Eisenmenger's syndrome. The disease process typically expresses itself in young women during early adulthood or middle age. The female to male ratio is approximately 5:1. Typical clinical features include: 1) a healthy young acyanotic woman, 2) effort syncope, 3) chest pain resembling angina, 4) dyspnea, weakness and fatigue especially with exertion and 5) *no previous history of having had a cardiac murmur* (i.e. no previous suspicion of having a left to right shunt).

There is presently no effective treatment of this entity and its etiology remains obscure. Recurrent small peripheral pulmonary emboli can be clinically indistinguishable from primary pulmonary hypertension and even at post mortem examination the distinction may still be difficult. However, this is one of the considerations in a patient that presents with evidence of severe pulmonary hypertension.

2. Idiopathic dilatation of the pulmonary artery

This entity is characterized by congenital dilatation of the pulmonary trunk and occasionally of its main branches in the absence of accompanying anatomic or physiologic cause (Fig. 49). Dilatation is probably the result of a developmental defect in pulmonary arterial elastic tissue. Idiopathic dilatation of the pulmonary artery must be distinguished from other causes of marked proximal enlargement of the pulmonary artery and its main branches including Eisenmenger's syndrome, recurrent pulmonary emboli and primary pulmonary hypertension. This is generally not difficult to do since there is no evidence on physical examination or electrocardiogram of the presence of pulmonary hypertension and it is only the radiographic appearance of the main pulmonary artery and its proximal branches that causes concern. The pulmonary artery impulse may be palpable on physical examination, a systolic



X-ray of a 21 year old woman with idiopathic dilatation of the pulmonary artery (hemodynamic and angiographic confirmation). The dilatation is confined to the pulmonary trunk. The film is otherwise normal.

Figure 49

ejection click may be noted, but the pulmonary valvular closure sound is normal, the right ventricle is not enlarged and the other important problems related to severe pulmonary hypertension do not exist. It may also be difficult to precisely distinguish between idiopathic dilatation of the pulmonary artery, mild valvular pulmonic stenosis and a small atrial septal defect. However, the absence of the expected murmurs and other classical abnormalities of these other two entities should ordinarily help one to distinguish between these abnormalities.

3. Recurrent pulmonary emboli

Reccurent pulmonary emboli over a period of time may result in marked enlargement of the main pulmonary trunk and its proximal branches. In addition, physical examination, electrocardiographic and roentenographic evidence of pulmonary hypertension may also be present. If there are other clinical manifestations to identify the presence of peripheral venous thrombosus and thus suggest recurrent pulmonary emboli, the diagnosis is not difficult, but these are often either subtle or totally absent. Thus, the differential diagnosis of pulmonary hypertension per se and of radiographic evidence of marked pulmonary hypertension includes this entity. Recurrent small pulmonary emboli may be difficult to exclude with certainty in any individual patient and as noted earlier, even at the time of post mortem examination it may be difficult to distinguish between "primary pulmonary hypertension" and recurrent small pulmonary emboli. Angiographically (pulmonary arteriography) the distinction may also be difficult and sometimes impossible.

Truncus Arteriosus

Truncus arteriosus is a congenital anomaly in which a single great vessel leaves the base of the heart through a single semilunar valve. The truncus is situated just above a ventricular septal defect and receives blood from both ventricles. The truncus gives rise to the coronary arteries and to the pulmonary and systemic circulation. No remnant of a second semilunar valve is present. There are four recognized types of truncus arteriosus (Fig. 50):

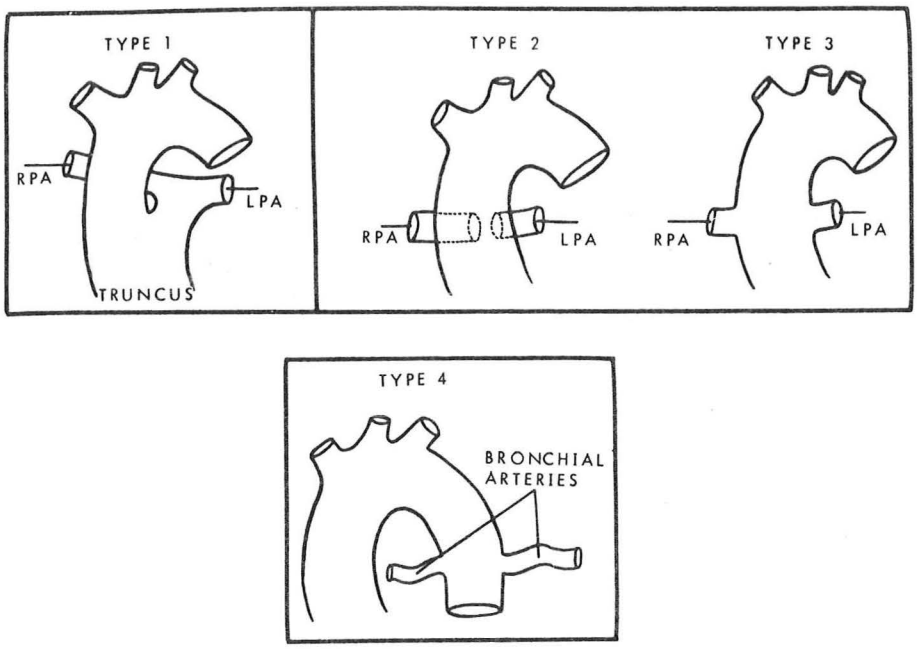
Type 1: A short pulmonary trunk emerges from the truncus arteriosus and gives rise to the right and left pulmonary arteries.

Type 2: The right and left pulmonary arteries arise directly from the posterior wall of the truncus.

Type 3: The right and left pulmonary arteries originate from the lateral walls of the truncus.

Type 4: The pulmonary arteries are absent altogether and the arterial supply to the lungs is through bronchial arteries.

There is some difference of opinion as to whether truncus arteriosus with absent pulmonary arteries is properly designated. This malformation closely resembles pulmonary valvular atresia with ventricular septal defect which is itself called "pseudotruncus". Although these two anomalies can sometimes be distinguished by selective angiography, their functional and clinical manifestations are the same.



Schematic illustrations of the various types of truncus arteriosus. (Adapted from Collett, R. W., and Edwards, J. E.: Surg. Clin. N. Amer. 29:1245, 1949.)

Type 1: A short main pulmonary artery arises directly from the truncus arteriosus and gives rise to the right and left pulmonary arterial branches.

Type 2 and Type 3: Both pulmonary arterial branches arise directly from the posterior or lateral wall of the truncus.

Type 4: The pulmonary arteries are entirely absent and the arterial supply to the lungs is through bronchial arteries.

Figure 50

The truncus is a large vessel since it accepts the entire output from both ventricles. Approximately one quarter of the time the truncus passes over the right bronchus as a right aortic arch. The valve of the truncus is positioned so that it overrides the ventricular septal defect; the number of cusps varies from two to six, but in the majority of instances there are three cusps analogous to a normal aortic valve. A ventricular septal defect is virtually always present and part of the syndrome of truncus arteriosus.

The physiologic consequences of truncus arteriosus depend chiefly upon the presence and size of the pulmonary arteries and upon the resistance to flow through the lungs. Right and left ventricular pressures are essentially the same since both ventricles communicate directly with the truncus. *Cyanosis occurs in some patients with truncus arteriosus with increased pulmonary blood flow; this is a useful combination diagnostically and an unusual one.* If large pulmonary arteries exist with a relatively low pulmonary vascular resistance, blood flow through the lungs is excessive. Under these circumstances the systemic arterial oxygen saturation is relatively high and may

approach normal, but this advantage exists at the consequence of marked volume overload of the left heart. With prolonged survival, the pulmonary vascular resistance gradually rises, pulmonary blood flow falls, cyanosis increases and volume overload of the left heart is partially relieved. When pulmonary vascular resistance rises to very high levels, flow to the lungs is markedly reduced and systemic arterial oxygen saturation is low. *Cyanosis then exists with diminished pulmonary blood flow.*

Approximately 90% of patients with truncus arteriosus have pulmonary arterial branches that arise either from a short main pulmonary artery or directly from the truncus itself. Truncus arteriosus generally occurs as an isolated anomaly.

History

Truncus arteriosus occurs with equal frequency in males and females. Family history is of little importance although isolated examples of truncus have been described in siblings. When truncus arteriosus occurs with absent pulmonary arteries, cyanosis is one of the first clinical features noted. However, when pulmonary arteries exist and pulmonary blood flow is normal or excessive patients are not initially cyanotic.

Survival to adulthood with this entity depends on there being blood flow to the lungs and also a delicate balance between flow patterns with advancing increases in pulmonary vascular resistance to regulate the volume of pulmonary blood flow and adequate systemic arterial oxygen saturation to sustain life. Infants with truncus arteriosus and large pulmonary arteries usually do not survive the first year of life; the majority die in the first six months from severe congestive heart failure. *The relatively few individuals that survive infancy and reach childhood and the first few years of adult life are those with favorable regulation of pulmonary blood flow.*

Physical Signs

Physical underdevelopment and cyanosis are the two important features in the physical appearance of patients with truncus arteriosus. Children with marked pulmonary plethora may appear acyanotic until they cry or exert effort and then cyanosis is detected.

A precordial bulge due to cardiac enlargement is common when truncus arteriosus is associated with large pulmonary arteries. The dynamic left ventricular impulse reflects the hypertropy and dilatation of volume overload. Since the right ventricle ejects at systemic pressures, a right ventricular impulse is virtually always noted. When separate pulmonary arteries arise directly from the truncus, a systolic impulse in the second left intercostal space is conspicuous by its absence. However, when the truncus gives rise to a short main pulmonary artery, that vessel may dilate and produce a second left intercostal impulse.

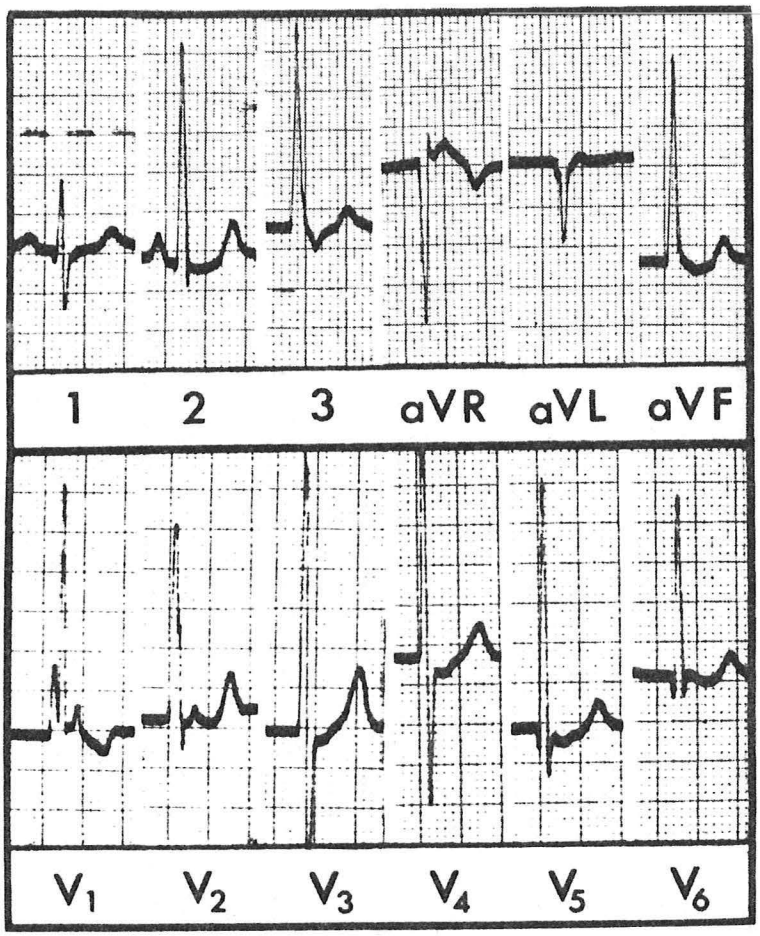
In individuals with truncus arteriosus with pulmonary arteries that are enlarged, the first heart sound is normal and is ordinarily followed by a systolic ejection click originating within the dilated solitary trunk. Systolic murmurs are common and as a rule are ejection in timing. The murmur pattern is influenced by the size of the ventricular septal defect and the volume of blood ejected by the left ventricle. When the ventricular septal defect is moderate to large in size and the left ventricular output large, a conspicuous murmur is generated. The murmur shortens and becomes softer when a rising pulmonary vascular resistance causes pulmonary blood flow and left ventricular stroke volume to diminish. Occasionally no murmur is detected particularly in individuals with increased pulmonary blood flow and very large ventricular septal defects. In this circumstance flow from the left ventricle into the truncus presumably occurs with relatively little turbulence so that the interventricular communication is silent. *It should be emphasized that when a murmur is present it is often preferentially transmitted to the second right intercostal space.* The systolic murmur radiates upward and to the right because it is transmitted directly into the truncus which basically takes the course of an aorta. The second heart sound is important both because it is loud and it lacks splitting. The second sound is loud because the large solitary truncus brings its valve close to the chest wall and the cusps close at systemic pressure. True splitting is theoretically impossible since the truncus is equipped with only one valve. However, when the cusps are structurally abnormal and when they exceed three in number, the closure sound may be "reduplicated" and an apparent split second sound generated.

Diastolic murmurs may originate from two sources. Apical mid-diastolic murmurs result from increased mitral valve flow in patients with large pulmonary arteries and increased pulmonary blood flow. These sounds are often introduced by prominent third heart sounds and exist as an "S_3 rumble complex". High frequency early diastolic murmurs maximal along the left sternal border sometimes occur because of incompetence of the truncal valve.

Laboratory Studies

The electrocardiogram ordinarily demonstrates volume overload of the left heart and pressure overload of the right (Fig. 51). Sinus rhythm is the rule. Evidence of right atrial enlargement may be apparent as may biventricular enlargement. The mean QRS axis is ordinarily directed inferiorly and often to the right. A rightward shift in mean QRS axis is expected when pulmonary blood flow is reduced and a leftward shift is likely when pulmonary blood flow is large, but marked axis deviation to either the right or left is unusual.

As noted earlier, when the truncus arteriosus exists with large pulmonary arteries and increased pulmonary blood flow the pulmonary vascular pattern appears plethoric. The combination of increased pulmonary blood flow with cyanosis is an important step in the clinical

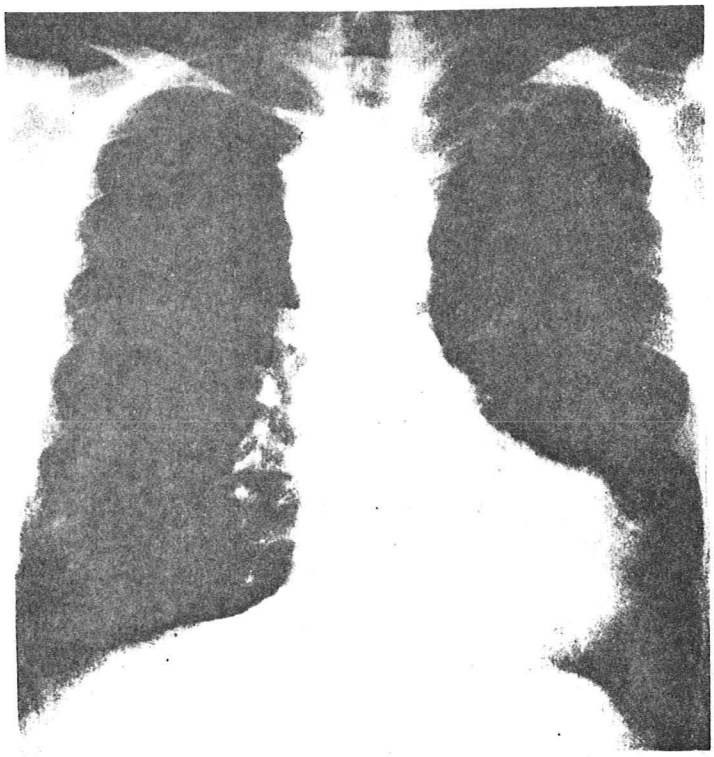


Electrocardiogram from a 15 year old cyanotic boy believed to have truncus arteriosus with absent pulmonary arteries. Flow to the lungs was through large bronchial collaterals. There is A-V dissociation, with peaked right atrial P waves in leads 2 and V_1. The QRS electrical axis is directly inferior. The rR' in lead V_1 reflects right ventricular hypertrophy; in leads V_{5-6} the prominent R waves and distinct q waves indicate that the left ventricle received adequate flow.

Figure 51

recognition of this variety of truncus arteriosus. The pulmonary artery segment is flat or concave when separate pulmonary arterial branches arise directly from the truncus. This concavity is especially apparent in the right anterior oblique projection. Occasionally, a large left pulmonary artery assumes a distinctive appearance with a relatively high shadow as it emerges from the cardiac silhouette and curves upward and to the left; this sign has been referred to as a "left hilar comma". A short, dilated, main pulmonary artery may originate from the truncus and

form a convex shadow. The truncus itself may produce a conspicuous rightward shadow in young adults and older children; the ascending portion may sweep to the right forming a convex upper border that resembles a dilated ascending aorta. The sweep may continue upward so that the level of the arch appears quite high. As pulmonary vascular resistance increases, pulmonary blood flow falls and the radiologic evidence of plethora diminishes and in fact the peripheral pulmonary lung fields may appear oligemic. When truncus arteriosus exists with absent pulmonary arteries, the x-ray is indistinguishable from that seen with pulmonary atresia with a ventricular septal defect (Fig. 52).



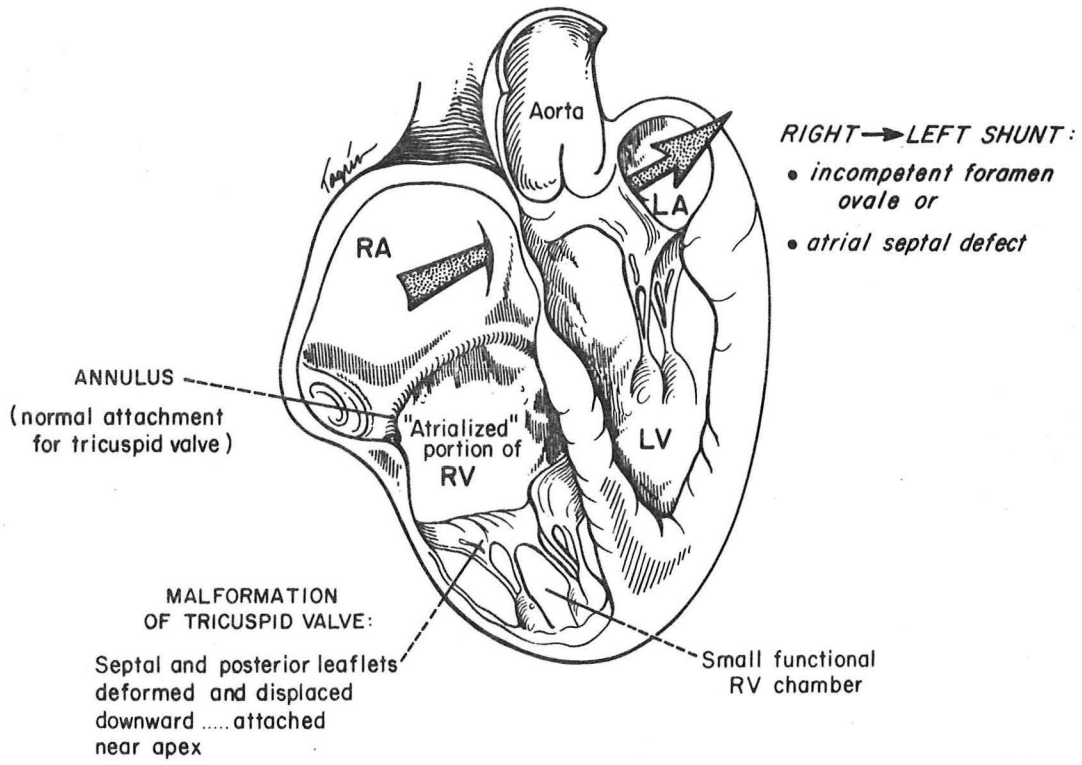
X-ray from the 15 year old cyanotic boy referred to in Figure 28-5. The radiologic picture resembles that of pulmonary atresia with ventricular septal defect. The lung fields exhibit the lacy, reticular pattern of bronchial collateral circulation. The aortic arch is large and right-sided. The pulmonary artery segment is concave and the cardiac apex boot-shaped.

Figure 52

(Modified from Perloff, J.K. The Clinical Recognition of Congenital Heart Disease, W.B. Saunders Company, Philadelphia, 1970.)

Ebstein's Anomaly

This anomaly of the tricuspid valve is a congenital defect in which the septal and posterior leaflets of the tricuspid valve are deformed, shortened, displaced downward and attached to the right ventricular wall near the apex (Fig. 53). The anterior leaflet arises from the normal position at the annulus fibrosus. As a result, the functional right ventricular chamber is significantly reduced. The right atrium is dilated and consists of normal atrium plus the "atrialized" portion of the right ventricle. An intact atrial septum is rare as most patients have an atrial septal defect or at the very least an incompetent foramen ovale.



Schematic illustration of Ebstein's anomaly of the tricuspid valve.

Figure 53

The basic abnormality results in tricuspid insufficiency due to the malformed leaflets. The reduced pumping ability of the right ventricle results in diminished pulmonary blood flow and contributes to the ineffective emptying of the right atrium. Thus, right to left shunting develops through stretching of the foramen ovale or through a true atrial septal defect. If there is no adequate communication between the atria, severe right heart failure ensues and death occurs early.

Clinical Features

This abnormality affects males and females equally. Symptoms may be absent, but when present consist of dyspnea on exertion, fatigue and weakness. Cyanosis is inevitably present at some point in the natural history of the defect and rhythm disturbances, i.e. especially recurrent supraventricular tachyarrhythmias are common. Right heart failure occurs as individuals age or as noted above sometimes in early infancy, particularly when an inadequate intraatrial communication exists.

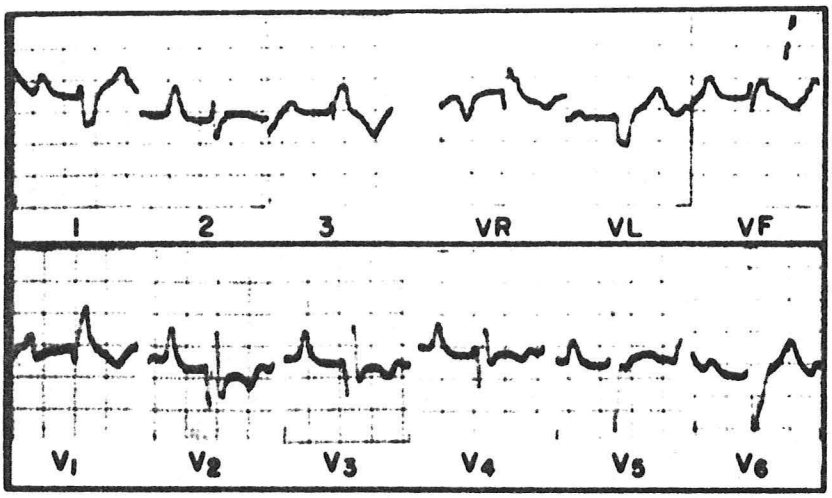
Physical Examination

Cyanosis of a mild to moderate degree is present sometime during life, but it may not be present until adult years. In some instances cyanois is not present at rest, but can be brought out with effort or exercise. The jugular venous pulse may reveal a prominent V wave or it may be absent since the velocity of the regurgitant jet across the tricuspid valve is markedly reduced. There is no systolic impulse over the body of the right ventricle. A systolic thrill is rarely palpable at the lower left sternal border. Auscultation reveals the presence of third and fourth heart sounds. The pulmonic valvular closure sound is often decreased. A holosystolic murmur is usually audible along the left sternal border, but it may not change with respiration in this setting.

Laboratory Studies

The electrocardiogram is quite helpful in identifying this entity. Right axis deviation, a prolonged P-R interval, right atrial enlargement and right ventricular conduction disturbances are common (Fig. 54). One does not detect, however, evidence of right ventricular enlargement. Arrhythmias, especially supraventricular, are common as is the occurrence of the Type B Wolff-Parkinson-White syndrome.

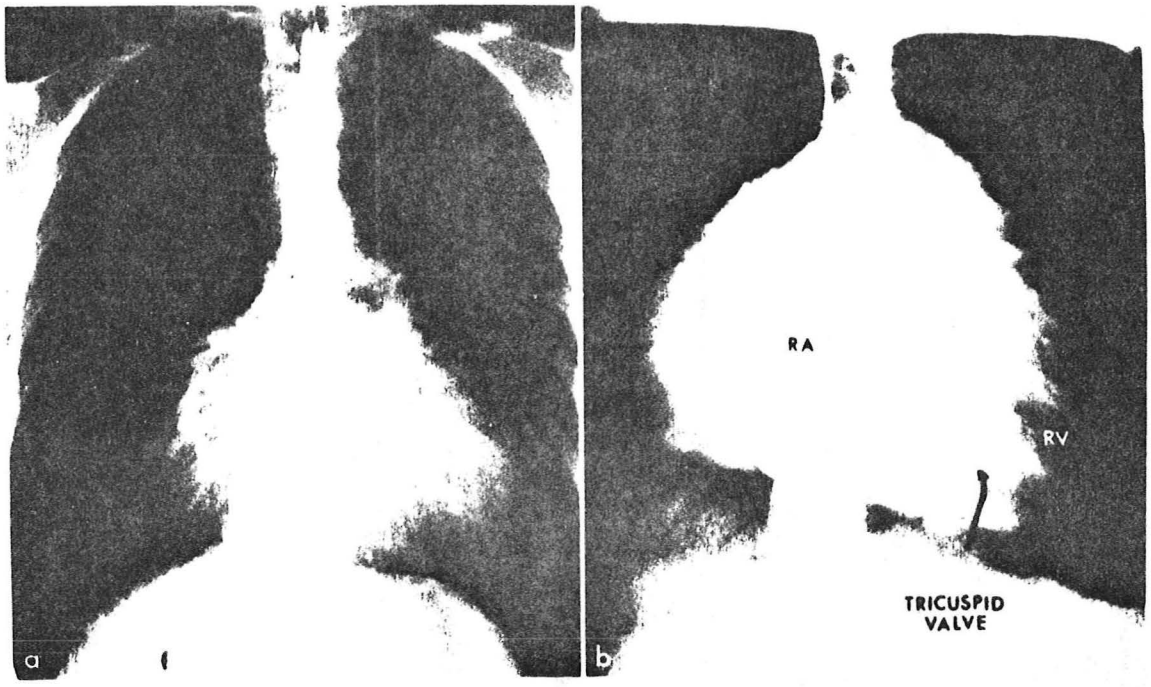
Classical chest x-ray demonstrates an enlarged heart; this enlargement is principally the right atrium (Fig. 55). Pulmonary vascularity often appears slightly decreased and the aorta small.



Electrocardiogram from the 18 year old boy referred to in Figure 13-3. There are tall, peaked, right atrial P waves, PR interval prolongation and complete right bundle branch block. The QR pattern with inverted T waves in leads V_1 is highly suggestive of Ebstein's anomaly.

Figure 54

(Modified from Perloff, J.K. The Clinical Recognition of Congenital Heart Disease, W. B. Saunders Company, Philadelphia, 1970.)

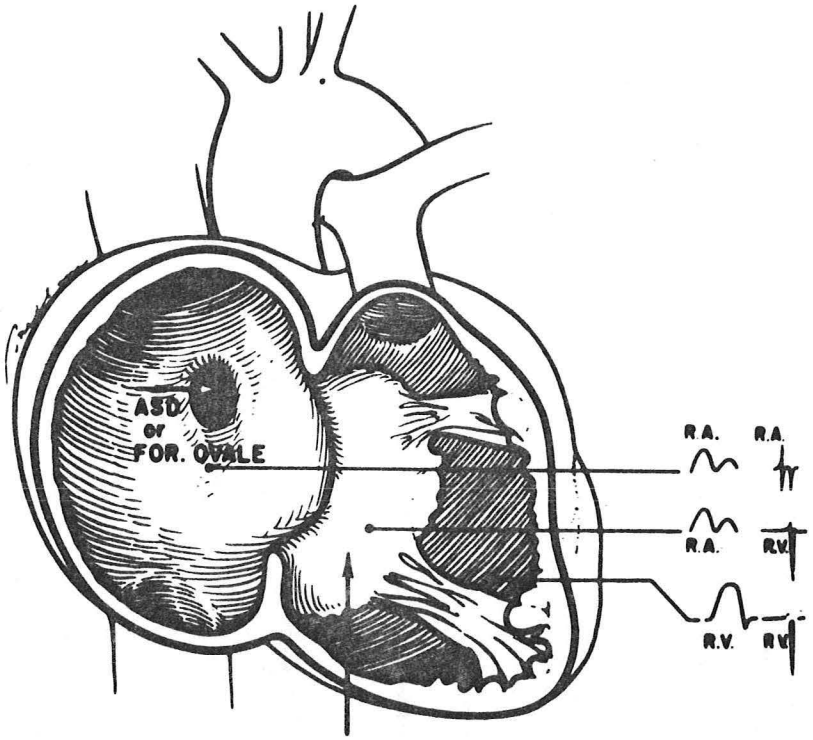


X-ray and angiocardiogram from an acyanotic 32 year old woman with Ebstein's anomaly. *a*. The lung fields are remarkably clear even when the degree of penetration is taken into account. The ascending aorta is small, but the left pulmonary artery forms a convexity along the upper left cardiac border. The enlarged right atrium causes a striking shadow along the lower right border of the heart. *b*, The angiocardiogram outlines the huge right atrium and the displaced tricuspid valve. (Courtesy of Dr. James Crockett, Charlottesville, Virginia.)

Figure 55

(Modifed from Perloff, J.K. The Clinical Recognition of Congenital Heart Disease, W. B. Saunders Company, Philadelphia, 1970.)

At cardiac catheterization the right to left shunt at the atrial level can ordinarily be identified. Systemic arterial unsaturation is also expected. Pressure measurements in the right atrium reveal an elevated right atrial mean pressure with prominence of the V wave. The diagnosis is made with certainty when one establishes that right atrial pressures exist in a region of the right heart that has right ventricular electrical potentials (Fig. 56). Selective angiography demonstrates a markedly enlarged right atrium, a small and poorly functioning right ventricular chamber and the presence of tricuspid regurgitation.



Schematic illustration of the essential anatomic, mechanical and electrophysiologic abnormalities in Ebstein's anomaly. In the right atrium (RA) proper both the pressure pulse and intracardiac electrocardiogram are right atrial; in the right ventricle (RV) proper both the pressure pulse and intracardiac electrocardiogram are right ventricular; however, over the displaced tricuspid valve, the pressure pulse is atrial, whereas the intracardiac electrocardiogram is ventricular ("atrialized" portion of the right ventricle). (Perloff, J. K.: Med. Ann. D. C. *31*:342, 1962.)

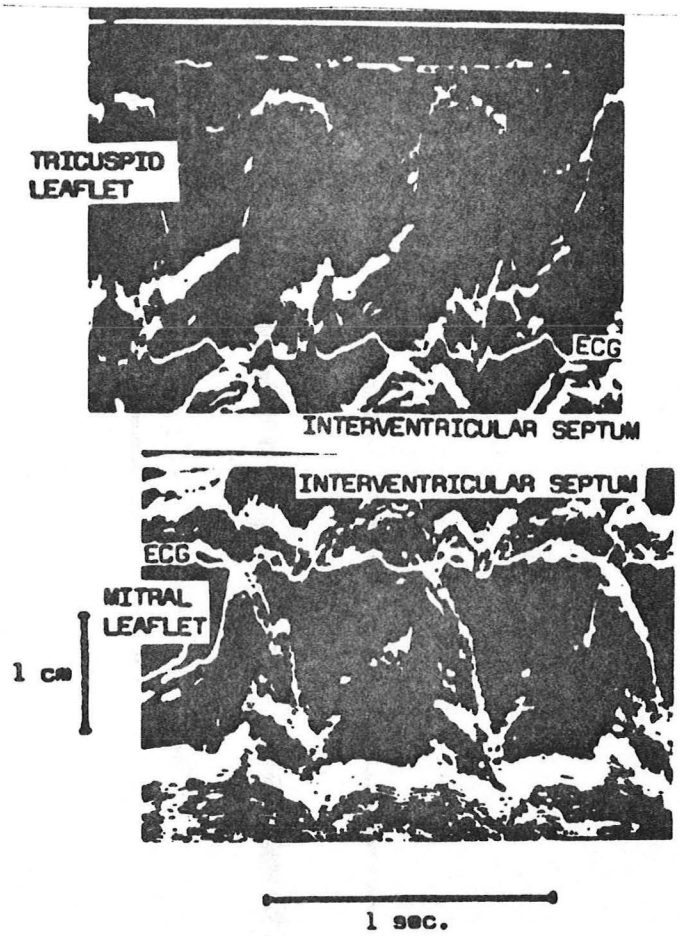
Figure 56

Course and Prognosis

The life span of these patients is varied. Many die by the second decade of life, but some have been reported to live much longer into adult life. The major causes of death are congestive heart failure and arrhythmias. Bacterial endocarditis is unusual with this entity.

Medical treatment is directed toward the complications, i.e. suppression of arrhythmias and control of the congestive heart failure. Occasionally tricuspid valve replacement is performed, but the results have been far from dramatic in terms of improving the patients functionally. A Glenn procedure (superior vena cava-right pulmonary artery anastomosis) has been performed in some patients in order to improve pulmonary blood flow but again the results are less than outstanding.

The echocardiogram has been alleged to be helpful in the recognition of patients with this entity (Fig. 57).



Echocardiograms from patient with Ebstein's disease. The tricuspid echo has a large excursion with slow diastolic closure. The tricuspid valve was recorded to the left of the usual location. (From Lundstrom and Edler: Ultrasoundcardiography in infants and children. Acta Paediat. Scand., 60, 117, 1971.)

Figure 57

Congenital Mitral Stenosis

Congenital obstruction to left atrial flow can be caused by one of a number of malformations that originate at or proximal to the mitral valve. The various types are listed in Table IV.

TABLE IV

Congenital Osbtruction to Left Atrial Emptying

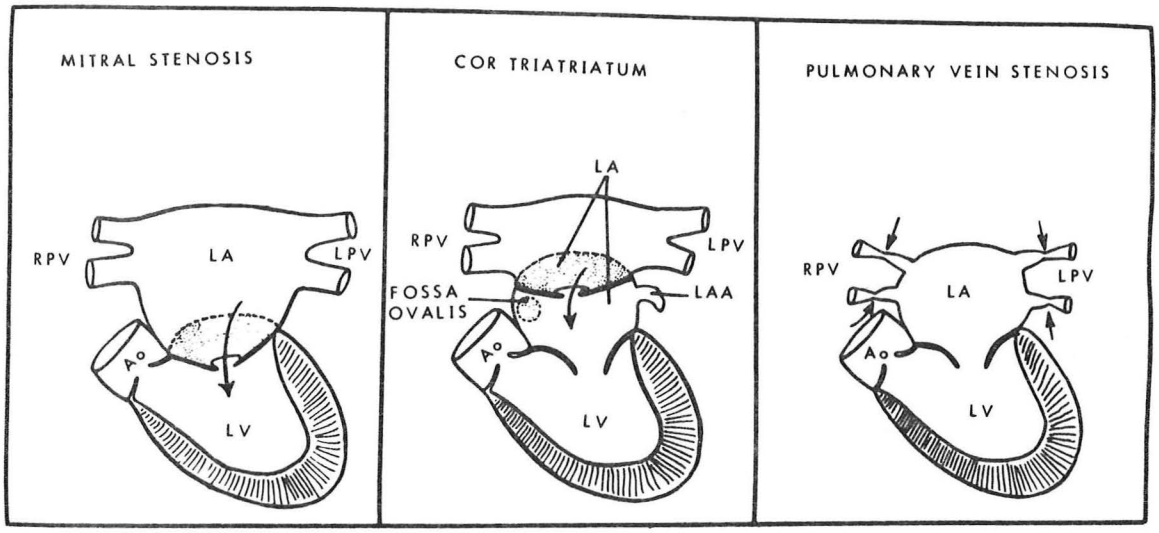
(from Perloff J.K. The Clinical Recognition of Congenital Heart Disease W.B. Saunders Company, Philadelphia, 1970)

1. Congenital mitral stenosis
   a. Obstruction at valvular level caused by congenitally abnormal leaflets and chordae
   b. Parachute mitral valve
   c. Supravalvular stenosing ring
2. Cor triatriatum
3. Congenital pulmonary vein stenosis

The physiologic consequences of this relatively rare list of disorders is that they result in increases in pulmonary venous pressure with pulmonary arterial and right ventricular hypertension. These lesions should be considered acyanotic malformations originating in the left heart and occurring without a shunt.

The abnormalities listed in Table IV are also shown in Figure 58.

In general, mitral stenosis may be produced as a consequence of the mitral leaflets being thickened, nodular or fibrotic. Commissures in this instance may be rudimentary and the chordae tendineae are shortened and fused and the papillary muscles fibrosed. The valve is transformed into a funnel-shaped structure. A second type of congenital mitral stenosis consists of a "parachute" deformity of the valve. Normal leaflets and commissures are in close apposition with one another by shortened chordae that converge and insert into a single large papillary muscle. The interchordal slit-like spaces provide the only source of access to the left ventricle. An additional form of congenital mitral stenosis is the result of a circumferential supravalvular ridge of connective tissue that arises at the base of the atrial aspect of the mitral leaflets. In this entity the mitral valve and its supporting structures are normal. With this abnormality the ridge produced may occur as an isolated stenosing defect and partition the left atrium into a proximal compartment receiving the pulmonary veins and a distal compartment containing the fossa ovalis, left atrial appendage and a low



a, Schematic illustration of the usual type of congenital mitral stenosis. The obstruction is at valvular level; the leaflets consist of a funnel-shaped, flat or diaphragm-like structure. (RPV, LPV = right and left pulmonary veins; LA = left atrium; LV = left ventricle; Ao = aorta.)

b, Schematic illustration of the usual variety of cor triatriatum. A fibrous or fibromuscular diaphragm partitions the left atrium. The proximal compartment receives the pulmonary veins and is a high pressure zone; the distal compartment contains the fossa ovalis and left atrial appendage (LAA) and is a low pressure zone.

c, Schematic illustration of one of the main types of pulmonary vein stenosis. The veins are locally narrowed near their junctions with the left atrium.

Figure 58

pressure zone. This latter complex may also exist as part of a developmental abnormality which combines four obstructive lesions, i.e. supravalvular ring of the left atrium, parachute mitral valve, subaortic stenosis and coarctation of the aorta. Finally, there may be obstruction that involves one or more pulmonary veins as they empty into the left atrium producing a reduction in left atrial flow.

The functional consequences of congenital mitral stenosis are essentially the same as those of acquired mitral obstruction. There is an elevation in the left atrial pressure and pulmonary venous pressure which is accompanied by hypertension in the pulmonary artery and the right ventricle.

History

Interestingly, there is no sex predilection with congenital mitral stenosis. Cardiac murmur is usually discovered within the first few months following birth. Obstruction to left atrial flow results in

orthopnea and cough, dyspnea and paroxysmal nocturnal dyspnea. Pulmonary arterial hypertension results in right ventricular failure, respiratory infections are frequent, recurrent pulmonary edema may occur. Individuals with this entity rarely have hemoptysis in contrast to those with acquired mitral stenosis.

Physical Examination

One expects ordinarily to hear an apical diastolic rumble in individuals with congenital mitral stenosis. The first heart sound may be increased, but it is often not as loud as that noted with acquired mitral stenosis. Opening snaps rarely occur with congenital mitral stenosis; absence of the opening snap appears to be related to the poor leaflet mobility that characterizes the congenitally stenosed mitral valve. An enlarged right ventricle can often be identified on physical examination and the second sound reflects the presence of pulmonary hypertension.

Laboratory Findings

Sinus rhythm is the rule with this entity. Electrocardiographic evidence of left and right atrial enlargement and of right ventricular enlargement may be documented. The mean QRS axis is often oriented downward and to the right. Electrocardiographic evidence of left ventricular hypertrophy is not present when this abnormality exists as an isolated finding.

The chest x-ray demonstrates evidence of pulmonary venous congestion and an enlarged main pulmonary trunk. Left atrial enlargement is ordinarily present. Calcification of the mitral valve is not seen on the x-ray. Evidence of right atrial and right ventricular enlargement may also coexist.

Cor Triatriatum

Partitioning of the left atrium into two compartments is known as "cor triatriatum". In this abnormality, the pulmonary veins drain into an accessory left atrial chamber that lies proximal to the true left atrium. The accessory chamber is believed to represent the dilated common pulmonary vein of the embryo. The distal compartment communicates with the mitral valve and contains the left atrial appendage and usually the fossa ovalis. The fibrous or fibromuscular diaphragm that partitions the left atrium possesses one or more openings, the size of these openings determines the degree of left atrial obstruction.

The functional consequences of this entity are essentially the same as those of mitral stenosis, i.e. pulmonary venous pressure is elevated and pulmonary hypertension exists. However, in this entity pressure in

the true left atrium is normal and the stenotic orifice remains open throughout the cardiac cycle. *Accordingly, cor triatriatum allows blood to flow across the obstructing partition throughout the cardiac cycle whereas in mitral stenosis forward flow is possible only in diastole.*

The severity of the symptomatic complaints depend on the severity of the obstruction. Survival into adulthood and to advanced ages is possible when the obstruction is not severe.

The physical findings with this entity may be similar to those found with congenital mitral stenosis or in some patients may be quite different. Cor triatriatum can occur with no murmur at all although systolic, diastolic or even continuous murmurs may be present. Evidence of pulmonary hypertension is expected on physical exam, but the first sound is generally unimpressive. Left atrial stenosing membrane is responsible for the different types of murmurs generated. The murmurs may be middiastolic, presystolic or even continuous. The continuous murmur has been ascribed to flow across the obstructing partition throughout the cardiac cycle.

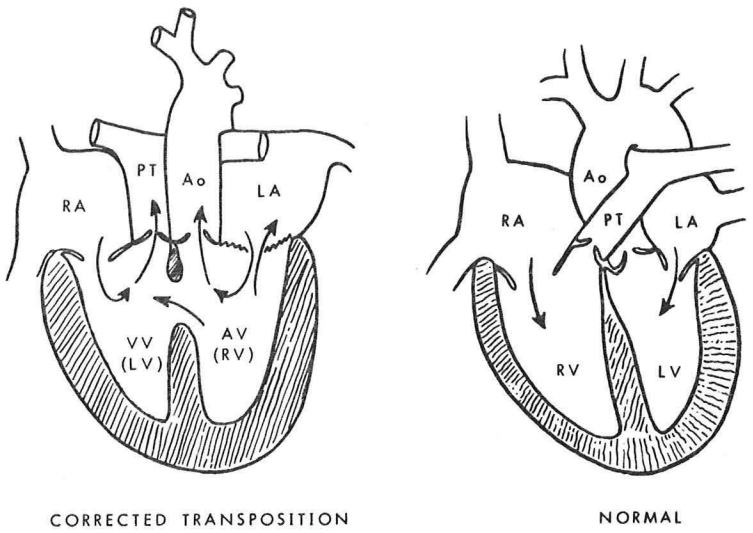
The findings electrocardiographically and on chest film are essentially the same as those with congenital mitral stenosis.

Treatment

Treatment consists of efforts aimed at relieving congestive heart failure using diuretics and salt restriction. Arrhythmias are controlled as they ordinarily are. Once prominent symptoms develop and are associated with evidence of pulmonary venous and pulmonary artery hypertension and a significant gradient is documented between the left atrium or a portion of the left atrium and the left ventricle, then surgical correction is indicated.

Corrected Transposition of the Great Arteries

Congenitally corrected transposition of the great arteries is characterized by the transposed portion of the great arteries being physioloically corrected by the abnormal position of the cardiac chambers (Fig. 59). In this entity the aorta and the pulmonary artery are transposed so that the aorta lies anteriorly to the pulmonary artery, but the abnormal position of the cardiac chambers results in arterial blood being directed into the aorta and venous blood into the pulmonary artery. The most common type of corrected transposition is the "bulboventricular". The major features of this entity are reversal of the right-left positional relation of the aorta and pulmonary trunk and inversion of the bulboventricular portion of the heart. The pulmonary trunk, therefore, arises posteriorly and to the right of the aorta from



Schematic illustrations comparing congenitally corrected transposition of the great vessels with the normal heart. In corrected transposition the aortic root is to the left of and anterior to the pulmonary trunk; the two great vessels arise parallel to each other and do not cross as in the normal heart; the arrangement, therefore, resembles complete transposition. However, in corrected transposition, right atrial blood flows across an a-v valve that is morphologically mitral into a venous ventricle (VV) that is morphologically left ventricle (LV) and then into the pulmonary trunk (PT). Left atrial blood flows across an a-v valve that is morphologically tricuspid into an arterial ventricle (AV) that is morphologically right ventricle (RV) and then into the aorta (Ao). The left a-v valve is often malformed and incompetent (reversed arrow), and there is often a defect in the ventricular septum. (Modified after Schiebler, G. L., et al.: Pediatrics *27*:851, 1961; and Edwards, J. E., et al.: Congenital Heart Disease.)

Figure 59

a rightsided ventricle with the anatomic features of a left ventricle. The aorta arise from a leftsided ventricle with the anatomic features of the right ventricle and is anterior and to the left of the pulmonary trunk. In the otherwise uncomplicated situation the course of the circulation is normal. Venous blood from both cavae returns to the right atrium and flows across an atrioventricular valve that is anatomically mitral into a ventricle that is anatomically left and then into a pulmonary trunk. Pulmonary venous blood enters the left atrium and flows across the atrioventricular valve that is anatomically tricuspid into a ventricle that is anatomically right and thence into an ascending aorta. Therefore, systemic venous blood is delivered into the pulmonary artery and pulmonary venous blood into the aorta. Thus, despite the transposed relationship of the great vessels the circulation is "corrected". A number of associated intracardiac defects create hemodynamic alterations that may result in problems. In the absence of associated complications and in the absence of associated intracardiac abnormalities survival for a normal lifespan is expected.

History

Males are afflicted more commonly than females. The presence of symptoms is dependent upon the presence or absence of associated defects; patients with uncomplicated corrected transposition are asymptomatic.

On physical examination in the uncomplicated form there are no abnormalities other than a slightly decreased first heart sound which may be the result of P-R prolongation on the electrocardiogram. The only other apparent abnormality is that the second sound is generally single as a consequence of the relatively posteriorly positioned pulmonary trunk which makes the pulmonary valve closure sound soft and often inaudible.

Laboratory Studies

The electrocardiogram identifies the presence of atrioventricular conduction disturbances in approximately 75% of patients and these include P-R interval prolongation, second degree heart block and sometimes complete heart block. Paroxysmal tachycardias and the presence of the Wolff-Parkinson-White syndrome may also be present. *However, the characteristic electrocardiographic finding is reversal of the initial forces of ventricular activation which results in an absence of Q waves in lead I and in the left precordial leads and the presence of Q waves in the right precordial leads.*

In corrected transposition, the "waist" of the heart may be quite narrow since the pulmonary artery is located behind the aorta.

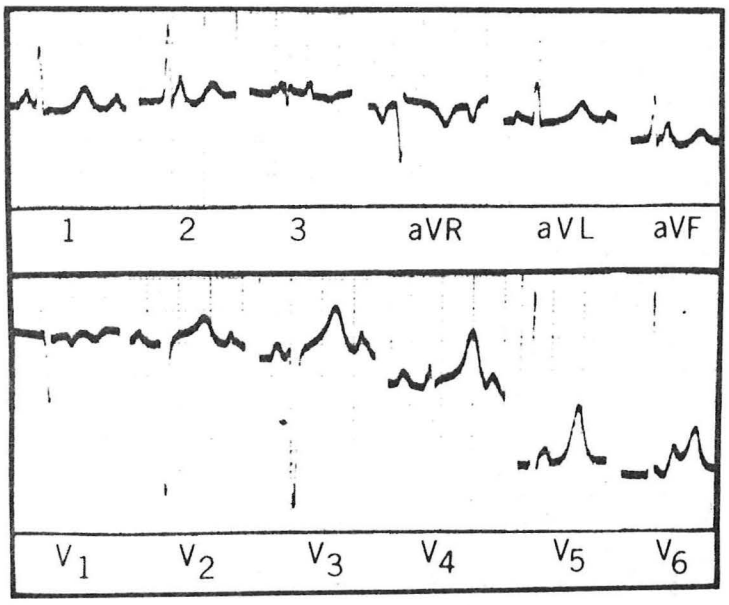
At cardiac catheterization, angiographic studies help to outline the abnormal position of the pulmonary artery which is medial and to the right of the ascending aorta in the frontal projection and posterior to it in the lateral projection. Angiography may also demonstrate the inversion of the ventricles.

Course and Prognosis

In the uncomplicated form of corrected transpostion there is no circulatory derangement and thus life expectancy is normal and no treatment required. If heart block complicates corrected transposition and particularly if the heart block is of the Mobitz 2 type or is complete, then pacemaker insertion is indicated. Antiarrhythmic agents may be necessary to treat recurrent supraventricular tachyarrhythmias and if coexisting intracardiac defects are present, then congestive heart failure may occur as a consequence of any of these and of particular concern is a large ventricular septal defect, gross atrioventricular valve insufficiency or other shunt lesions.

Congenital Complete Heart Block

This entity was probably first recognized around 1900 with several reports describing children with Stokes-Adams attacks with death in childhood. The abnormality is characterized by a slow heart rate and typically with a junctional or sometimes ventricular pacemaker (Fig. 60).



Electrocardiogram of a 25 year old man. The P waves are independent of the QRS complexes. The QRS is "supraventricular" and its axis normal. There are tall R waves and relatively tall peaked T waves in leads V_5 and V_6.

Figure 60

The rate of the junctional pacemaker is most commonly 50-60 beats/min. Often this is adequate to provide normal perfusion of important organs including the brain. The basis for the development of this abnormality may be congenital absence of the atrioventricular node, replacement of AV junctional tissue by fibrotic scar, or separation of the His bundle from the AV junction by fibrous bands that disrupt continuity. Some evidence that occasionally this abnormality is due to in utero infection and in some of these circumstances the congenital complete heart block may resolve and return to sinus rhythm has been documented.

In most individuals with this abnormality, the myocardium is basically normal and the ventricular rate rarely falls below 40 beats/min. This means that the abnormality is ordinarily well tolerated at rest and initially temporary or permanent pacemaker insertion is often not necessary. In many individuals there is also a modest heart rate increase with exercise as well as an increase in cardiac output further emphasizing the relatively benign aspect of this abnormality during the early years of life. It does need to be emphasized, however, that occasionally slower ventricular responses are present, a marked decline in heart rate may develop or frank asystole may occur resulting in severe decreases in cerebral blood flow and Stokes-Adams attacks. This, of course, requires permanent pacemaker insertion.

The physical findings, the electrocardiogram and the chest x-ray, are those that one would expect for the abnormality described above. There is no sex predilection for the abnormality. Familial congenital heart block is uncommon, but definitely recognized.

Cardiac Malposition

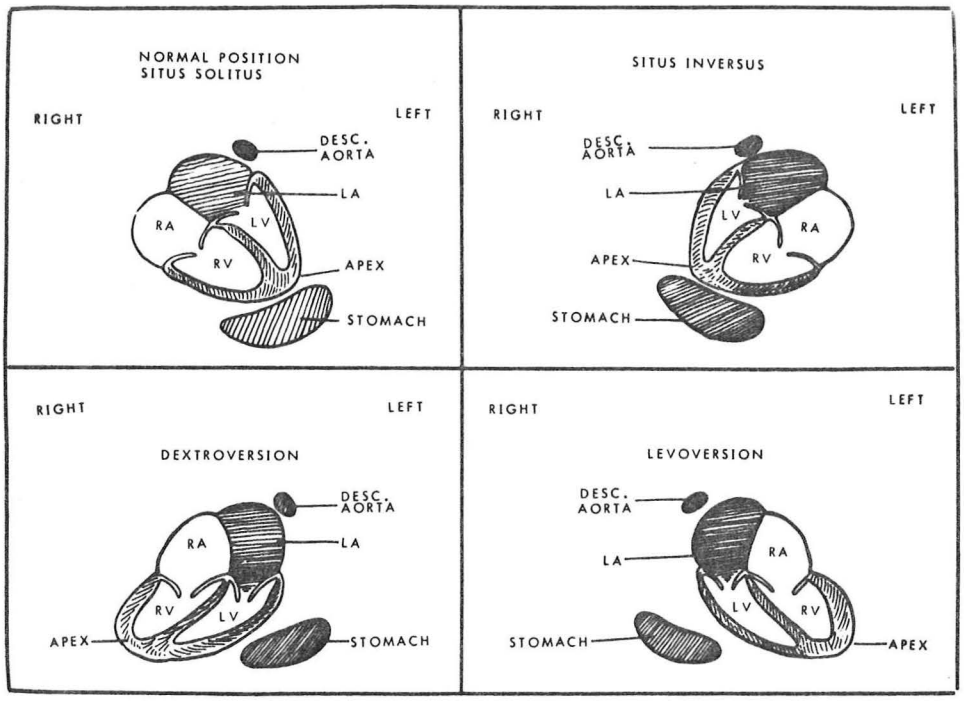
Figures 61 and 62 identify the important cardiac malpositions. Table V identifies additional terms that are used and provides a description of the associated cardiac position.

TABLE V

(from Perloff J.K. The Clinical Recognition of Congenital Heart Disease, W.B. Saunders Company, Philadelphia, 1970)

1. *Situs Solitus* (Normal Position) = *Left* Descending Aorta + *Left* Apex
2. *Situs Inversus* = *Right* Descending Aorta + *Right* Stomach + *Right* Apex
3. *Dextroversion* = *Left* Descending Aorta + *Left* Stomach + *Right* Apex
4. *Levoversion* = *Right* Descending Aorta + *Right* Stomach + *Left* Apex

Two basic positions of the heart and abdominal viscera are described. First "situs solitus". Situs means "site" or "position" and solitus means "usual". Situs solitus means normal position. The second term that is utilized is "situs inversus". Inversus means "turned about" and implies an organ arrangement that is opposite or the reverse of normal. The term "mirror image dextrocardia" has also been applied to this abnormality. With situs solitus (normally positioned) heart, the aortic



Schematic illustration of the anatomic relationship of the descending aorta, left atrium, apex and stomach in the four basic cardiac positions.

In situs solitus (normal position) the descending aorta, left atrium, apex and stomach are all on the left.

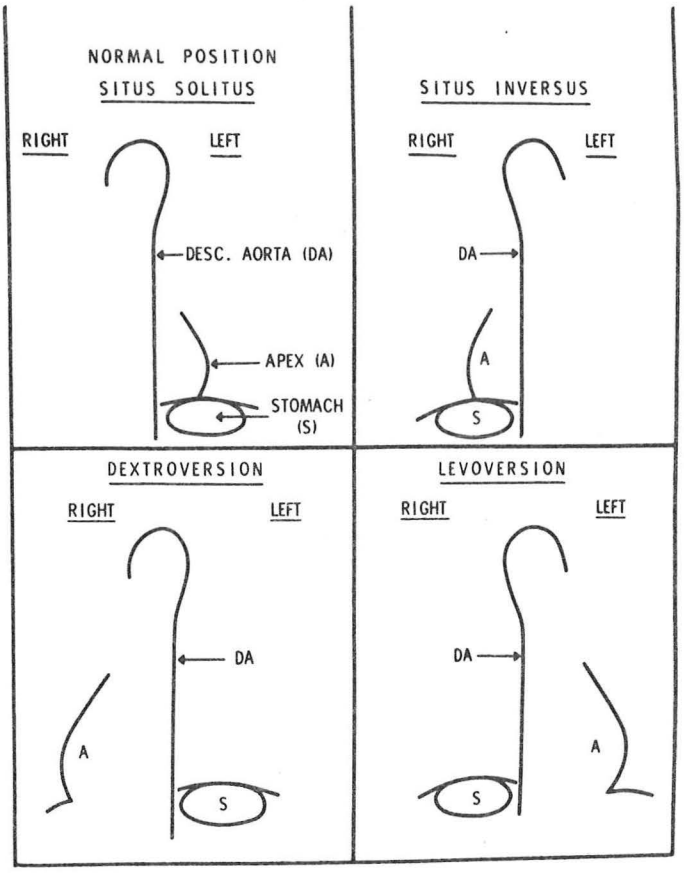
In situs inversus (mirror image dextrocardia) the descending aorta, left atrium, apex and stomach are all on the *right*.

In dextroversion the descending aorta, left atrium and stomach are on the *left* (as in the normal) but the apex is on the *right*.

In levoversion the descending aorta, left atrium and stomach are on the right (as in situs inversus) but the apex is on the *left*.

(Modified after Elliott, L. P., et al.: Invest. Radiol. *1*:17, 1966.)

Figure 61



Schematic illustration of the alignment of descending aorta, apex and stomach in the four basic cardiac positions. The line drawings are shown as they would be projected in the frontal view of a chest x-ray.

In situs solitus (normal position) the descending aorta, apex and stomach are all on the *left*.

In situs inversus (mirror image dextrocardia) the descending aorta, apex and stomach are all on the *right*.

In dextroversion the descending aorta and stomach are on the *left* (as in the normal), but the apex is on the right.

In levoversion the descending aorta and stomach are on the right (as in situs inversus), but the apex is on the *left*. (Modified after Elliott, L. P., et al.: Invest. Radiol. *1*:17, 1966.)

Figure 62

arch, left atrium, cardiac apex and stomach are all on the left side. In the situs inversus (mirror image) abnormality, the aortic arch, left atrium, cardiac apex and stomach are all on the right side (Figs. 61 and 62, Table V).

This basic terminology is completed with the addition of two additional terms -- "dextroversion" and "levoversion". Dextroversion exists when the cardiac apex is on the right while the aortic arch, left atrium and stomach remain in their normal leftsided positions. Levoversion exists when the cardiac apex is on the left while the aortic arch, left atrium and stomach are on the right side. Accordingly, there are two types of "left-thoracic hearts"; the normal position and levoversion. There are two types of "right-thoracic hearts"; situs inversus or mirror image dextrocardia and dextroversion.

In the early fetus the cardiac apex is on the side opposite from that which it will ultimately occupy in later fetal life. In the normal position, the fetal apex moves from right to left. In situs inversus

the fetal apex moves from left to right. Many basic variations of this pattern can occur: 1) the apex may remain on the right (dextroversion of the ventricles) while the aortic arch, left atrium and stomach assume their normal leftsided position or 2) the apex may remain on the left (levoversion of the ventricles) while the aortic arch, left atrium and stomach assume the situs inversus position.

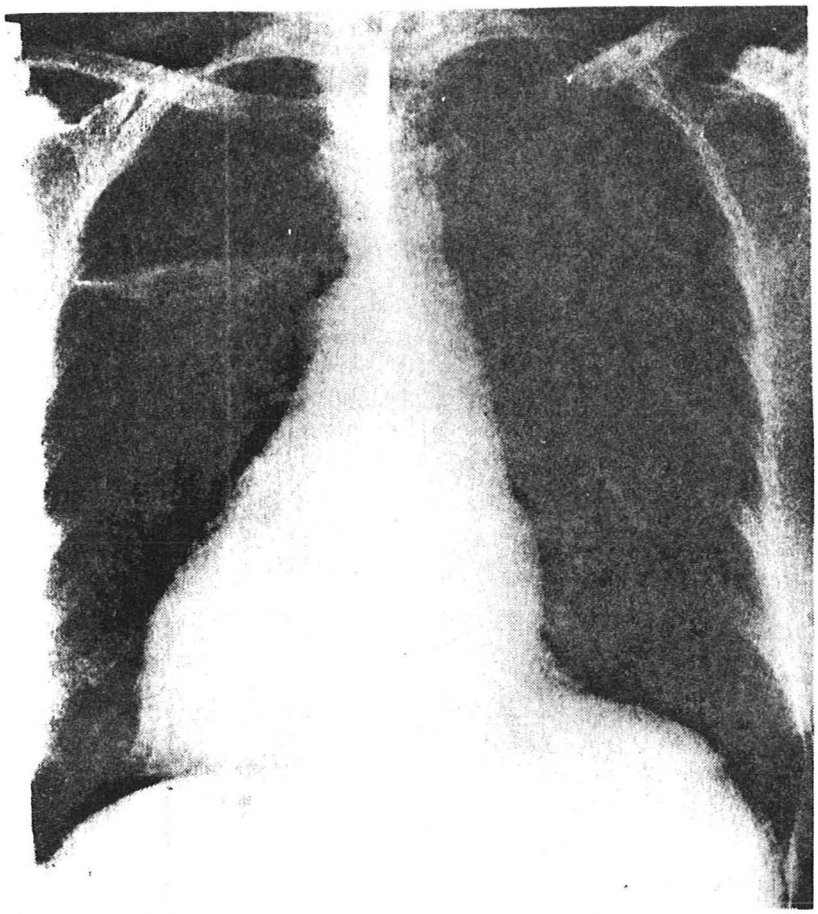
In situs inversus with mirror image dextrocardia, about 90% of the patients have hearts that are otherwise normal. Further, in large numbers of patients with known congenital heart disease, the incidence of situs inversus is about 1%, a frequency that is higher than situs inversus in the general population. If a congenital defect does exist, it is most likely to be corrected transposition of the great vessels.

In individuals with dextroversion of the heart, patients almost invariably have additional congenital cardiac malformations, although occasional examples of otherwise normal hearts have been described. The commonest associated anomalies are 1) corrected transposition of the great vessels, 2) pulmonic stenosis, and 3) ventricular or atrial septal defects. These lesions may occur singly or in combination. The acyanotic patient with dextroversion is most likely to have either pulmonic stenosis or a left to right shunt through a ventricular or atrial septal defect; cyanosis generally implies the presence of pulmonic stenosis with reversed interventricular or interatrial shunt.

Levoversion with an otherwise normal heart is so rare that it is believed to be virtually nonexistent. It is generally agreed that coexisting congenital cardiac defects are almost always present with this abnormality.

Mirror image dextrocardia is consistently associated with the stomach on the right side (Fig. 63). A normally positioned heart has on a few occasions been reported with the stomach on the right, although these cases represent examples of isolated inversion of the stomach without any other alterations.

Situs inversus with mirror image dextrocardia generally occurs in hearts that are otherwise normal so this abnormality is likely to be discovered accidentally during a routine physical examination or following an x-ray of the chest. It is interesting that left-handedness is alleged to occur in about 40% of these individuals. These individuals have normal longevity. *The only important fact is that symptoms that may develop in later life as a consequence of coronary artery disease, etc may have a rather unusual radiation and be located in the opposite chest or quadrant of the abdomen from the position in which they would ordinarily be expected.* The interesting association between sinusitis, bronchiectasis and situs inversus is referred to "Kartagener's syndrome". Respiratory symptoms are significant parts of the history and may prompt an investigation of the respiratory tract. The electrocardiogram is helpful in identifying mirror image dextrocardia since the P wave is generally negatively oriented in standard lead I of the electrocardiogram.



X-ray from a 65 year old man with mirror image dextrocardia and no coexisting congenital heart disease. Anginal pain was retrosternal and right precordial. Note the "R" in the upper right corner, indicating that the film is being viewed properly. The aortic arch, stomach and cardiac apex are all clearly on the right; the diaphragm is lower on the side of the apex (right).

Figure 63

Dextroversion and levoversion are generally discovered because of the various manifestations of the accompanying congenital cardiac defects. Longevity is determined by the type of associated anomaly and not by the cardiac malposition.

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