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Ventricular outflow tract anomalies

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The ventricular outflow tract is the most common site of congenital cardiac defect. The anomalies involving the ventricular outflow tract can be categorized into two groups: those occurring as an isolated lesion in an otherwise normally formed heart, and those occurring as a complex malformation with an abnormal ventriculoarterial connection and an abnormal great arterial relationship. The latter group includes tetralogy of Fallot, complete and corrected transpositions of the great arteries, double-outlet right and left ventricles, and truncus arteriosus. These lesions are also known as 'conotruncal or truncoconal anomalies', because they are considered to result from an errant development of the conotruncal region of the embryonic heart.[1]

Conotruncal anomalies account for 25–30% of all congenital heart diseases in infants (Table 19.1).[2–4] Tetralogy of Fallot and complete transposition are the two most common conotruncal anomalies, each occurring in approximately 8–12% of infants with congenital heart disease. Double-outlet ventricle, truncus arteriosus and corrected transposition are less common. The conotruncal anomalies are reported to be less prevalent in the fetal series than in the infant series. They accounted for only 10% of the fetal cases analyzed by Allan et al.[5] This discrepancy is considered to be mainly due to the low detection rate of conotruncal anomalies at fetal cardiac screening, which often depends on the four-chamber view alone.[5–7] On the other hand, conotruncal anomalies frequently require urgent treatment immediately after birth. Therefore, it is important to detect conotruncal anomalies prenatally by incorporating the ventricular outflow tract evaluation in the fetal screening protocol.

In this chapter, we discuss normal ventricular outflow tract anatomy, the fetal echocardiographic technique for ventricular outflow tract evaluation, the pathology and fetal echocardiographic findings of conotruncal anomalies, and the postnatal outcome.

Normal anatomy of the ventricular outflow tracts

The right and left ventricular outflow tracts exhibit significant morphological differences (Figure 19.1). The

Table 19.1. Relative distribution (%) of the conotruncal anomalies

Diagnosis	*NER*[2] *1969–1974 n = 2381*	*Brompton*[3] *1973–1982 n = 1653*	*NGP*[4] *1982–1987 n = 4735*	*Fetal diagnosis*[5] *1980–1992 n = 1006*
Tetralogy of Fallot	8.9	9.9	11.5	3
Complete transposition	9.9	10.4	10.1	2
Corrected transposition	0.7	0.8	0.9	< 1
Double-outlet right ventricle	1.5	3.0	2.0	3
Truncus arteriosus	1.4	2.1	2.0	1.5

NER, New England Infant Cardiac Program; NGP, Northern Great Plains Regional Cardiac Program.

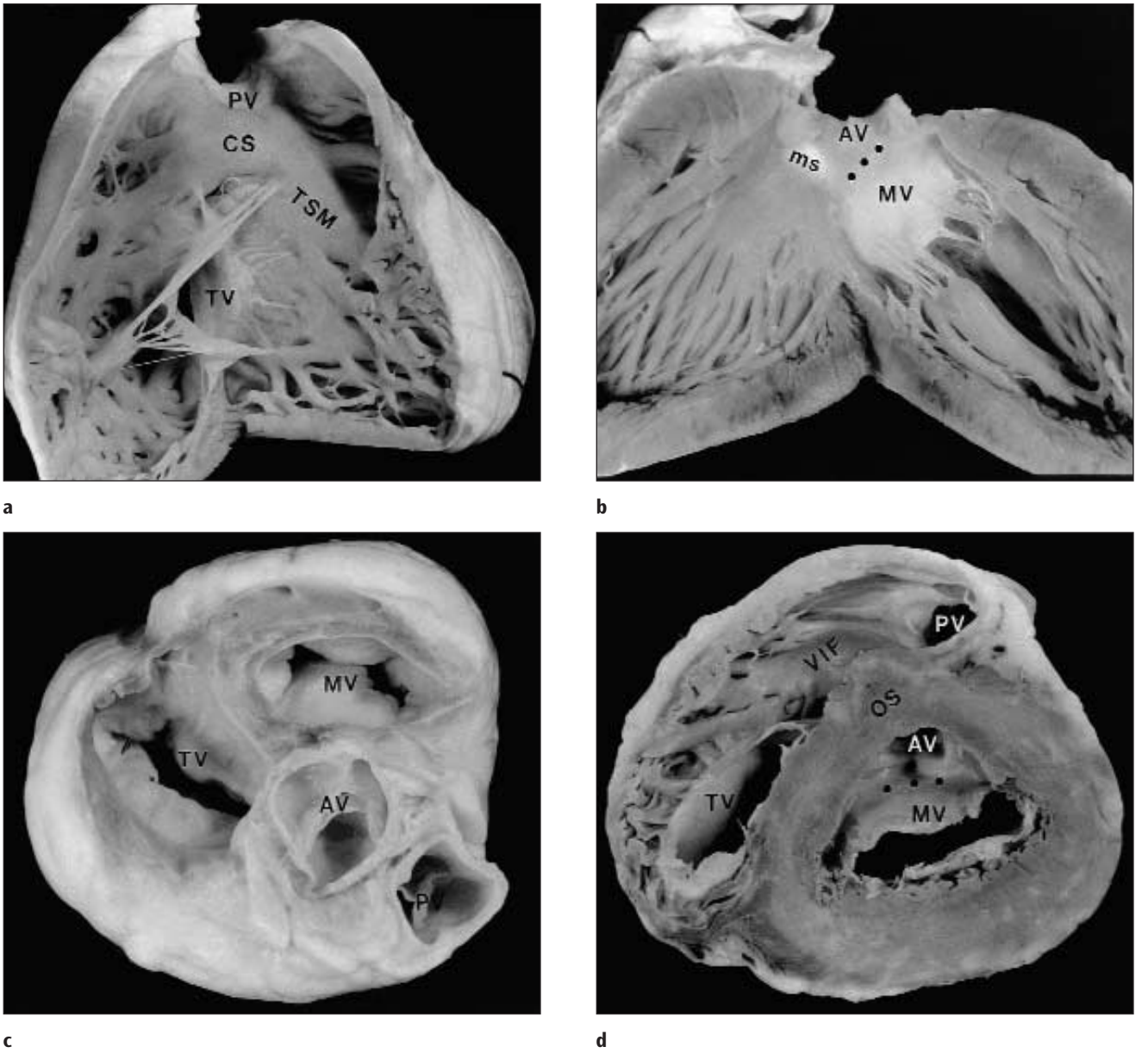


Figure 19.1
Photographs of a normal cardiac specimen. Opened right (**a**) and left (**b**) ventricles, and the base of the ventricles seen from above (**c**) and below (**d**). As the crista supraventricularis (CS) intervenes between the tricuspid (TV) and pulmonary (PV) valves, the right ventricular outflow tract is a completely muscular funnel. The crista supraventricularis consists of parietal and septal parts: the ventriculoinfundibular fold (VIF) and outlet septum (OS), respectively. The left ventricular outflow tract is partly devoid of muscular wall because of the fibrous continuity (dots in (**b**) and (**d**)) between the mitral (MV) and aortic (AV) valves. Note the deeply wedged position of the aortic valve between the tricuspid and mitral valves in (**c**). ms, membranous septum; TSM, trabecula septomarginalis.

major difference is the existence of the crista supraventricularis in the right ventricle. The crista supraventricularis is the muscular crest that separates the pulmonary and tricuspid valves in the normal right ventricle.[8,9] The major part of the crista is a parietal structure, which is called the ventriculoinfundibular fold.[9] Only a small part of the crista is a septal structure, which is called the outlet or infundibular septum. The outlet septum is cradled between the two limbs of the trabecula septomarginalis. The crista supraventricularis completes the muscular funnel of the right ventricular outflow tract. In the left ventricle, on the other hand, the aortic

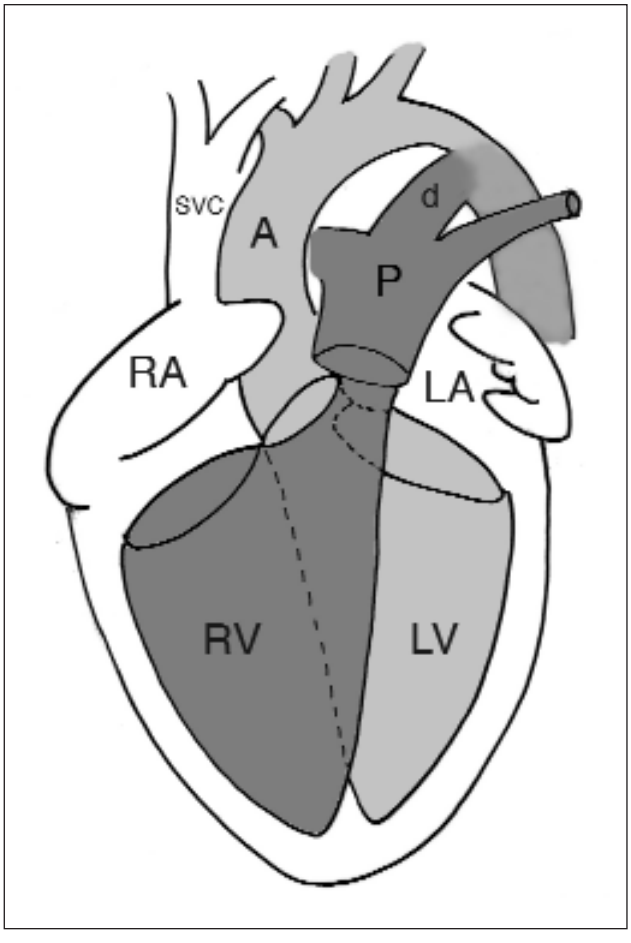


Figure 19.2
Diagram showing the long-axis view of the normal right (RV) and left (LV) ventricles. The two outflow tracts cross each other. A, aorta; d, ductus arteriosus; LA, left atrium; P, pulmonary artery; RA, right atrium; SVC, superior vena cava.

and mitral valves are in fibrous continuity and there is no muscular crest between the two valves. Therefore, the left ventricular outflow tract is not completely muscular, and is less conspicuous than the right ventricular outflow tract. The two ventricular outflow tracts cross each other (Figure 19.2). The right ventricular outflow tract is anterior to the left ventricular outflow tract. The right ventricular outflow tract faces leftward, and the left ventricular outflow tract faces rightward, as they course upward toward the arterial trunks. The aortic valve is deeply wedged between the tricuspid and mitral valves (Figure 19.1c and d). This unique position allows a fibrous continuity to be maintained between the aortic and mitral valves. Being supported by a muscular infundibulum that faces leftward, the pulmonary valve is located to the left of and anterior to the aortic valve.

Echocardiographic technique for fetal ventricular outflow tract evaluation

There are three facets of the make-up of the heart: the morphologies, the connections, and the relations. For the evaluation of the ventricular outflow tracts, the ventricles and great arteries should be identified according to the morphological criteria as discussed in the chapter on the sequential segmental approach. Color Doppler mapping facilitates identification of the great arteries. After morphological identification, the ventriculoarterial connections and great arterial relationship should be evaluated by using the following echocardiographic views (Figures 19.3 and 19.4):[7,10–21]

1. Left ventricular outflow tract view
2. Right ventricular outflow tract view
3. Basal short-axis view
4. Three-vessel view

The left and right ventricular outflow tract views are necessary for determination of the type of ventriculoarterial connection.[11,14–18,21] To obtain these two views, we start from a transducer position for a four-chamber view (Figure 19.5). The transducer is then moved radially around the

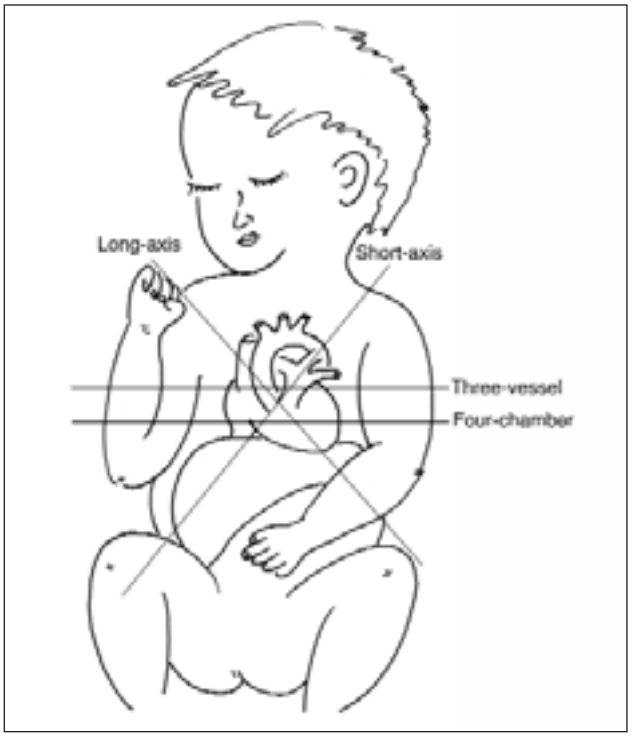
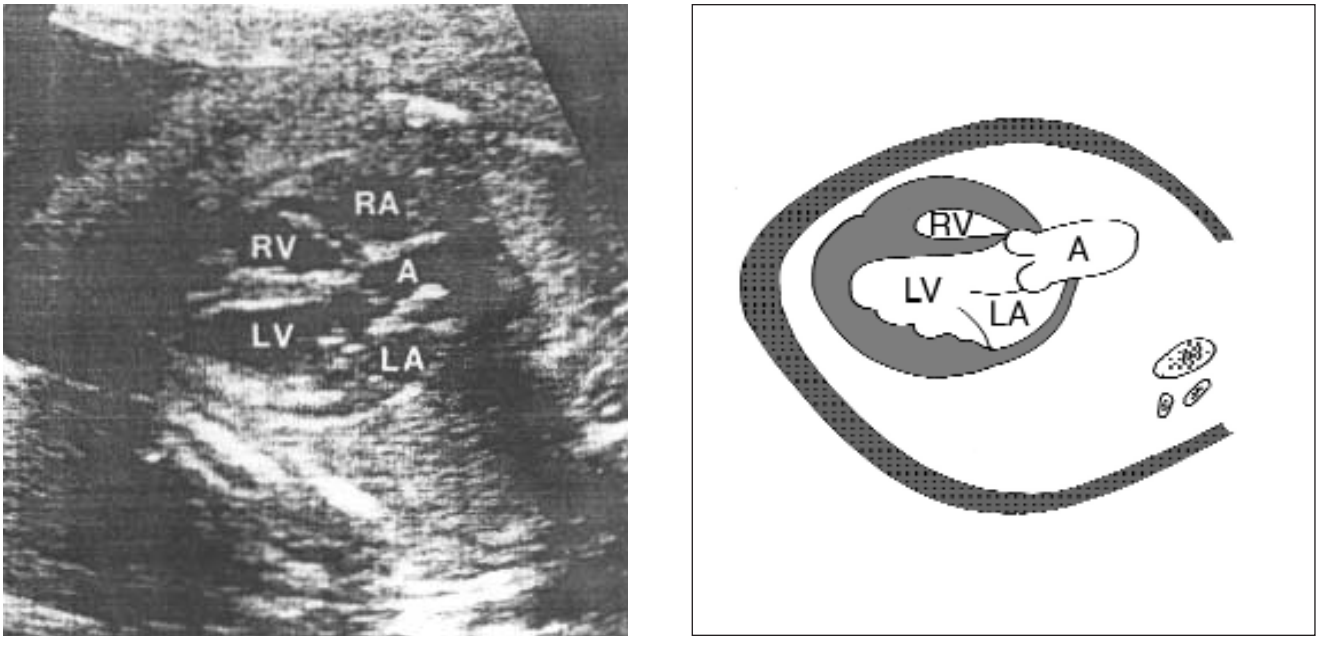


Figure 19.3
Diagram showing the imaging planes for echocardiographic views.



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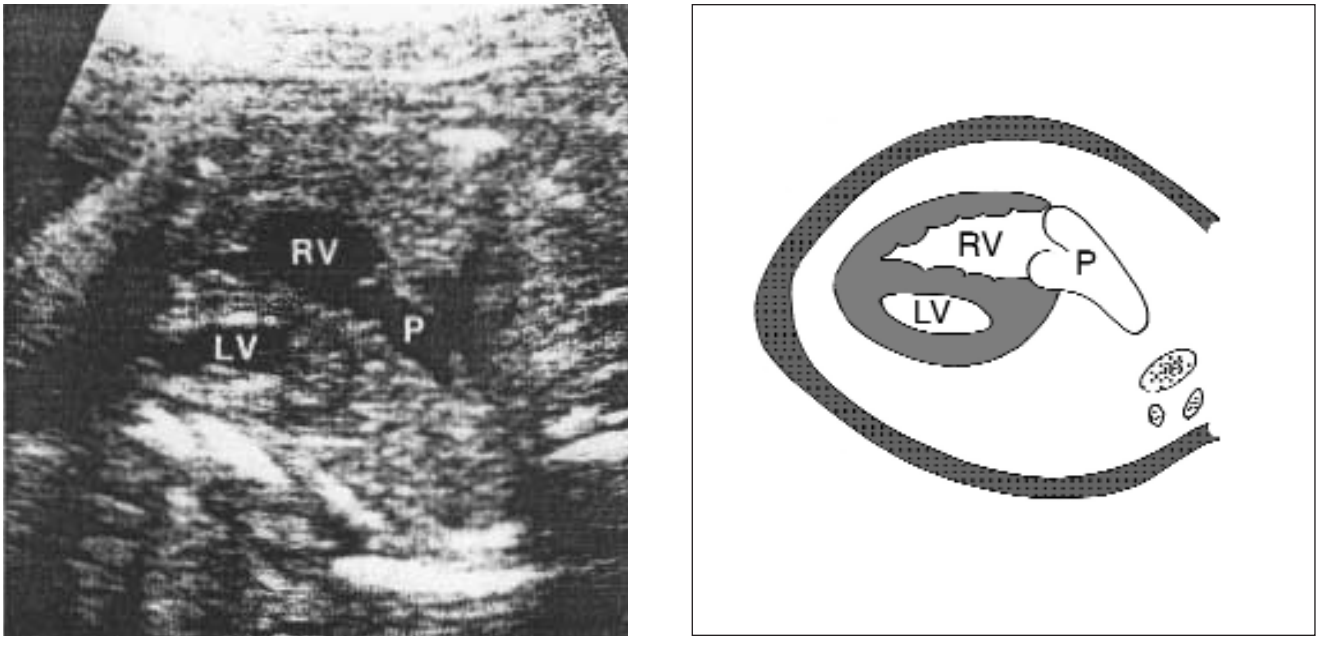
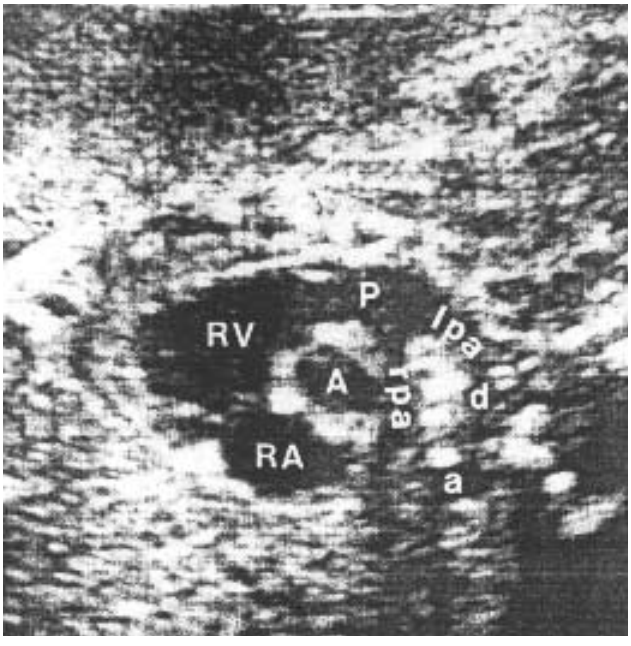
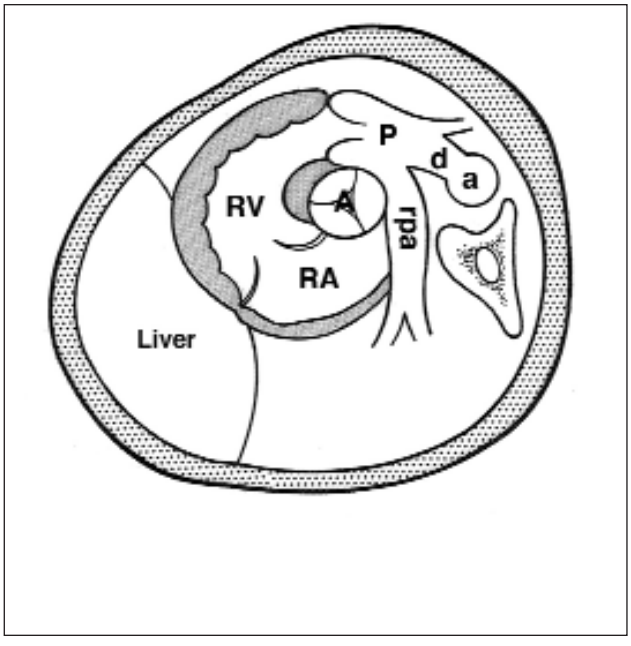


b

Figure 19.4
Echocardiograms and corresponding diagrams for ventricular outflow tract examination. (**a**) Left ventricular outflow tract view. (**b**) Right ventricular outflow tract view.

maternal abdomen, keeping the four-chamber plane in view, until the ventricular septum is aligned perpendicular to the sonographic beam axis. From this particular transducer position, the left ventricular outflow tract view can be obtained simply by rotating the transducer through 40–50° clockwise or counterclockwise toward the cardiac apex. The transducer is then moved slightly upward along the fetal thorax until the right ventricular outflow tract view is visualized. It should be remembered that the normal right and left ventricular outflow tracts cross each other and that they can not be visualized in a single imaging plane (Figures 19.2 and 19.4a,b).





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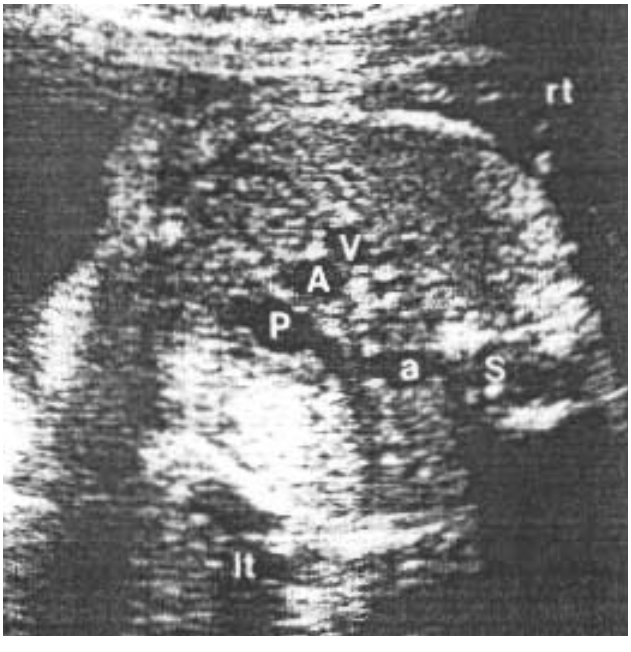
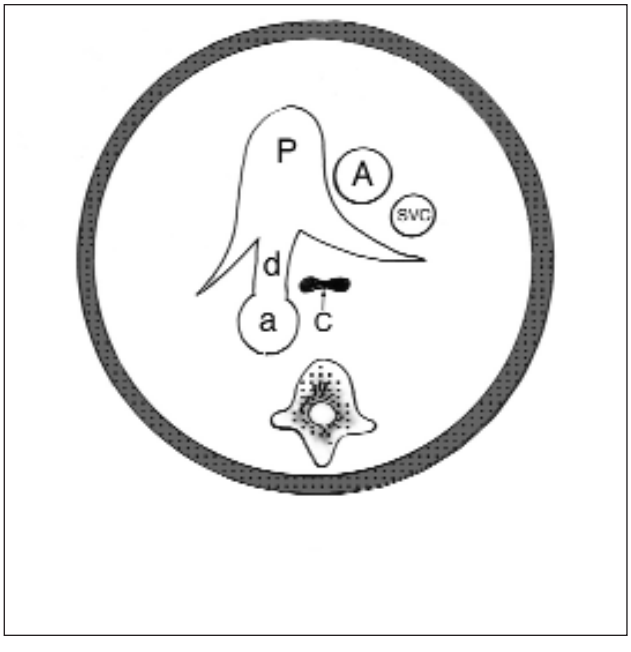




d

(**c**) Short-axis view of the base of the ventricles. (**d**) Three-vessel view. A, ascending aorta; a, descending aorta; C, carina; d, ductus arteriosus; LA, left atrium; lpa, left pulmonary artery; lt, left; LV, left ventricle; P, main pulmonary artery; RA, right atrium; rpa, right pulmonary artery; rt, right; RV, right ventricle; svc, superior vena cava.

The basal short-axis view is used for evaluation of the morphology of the outlet septum and the ventricular outflow tract dimension.[14,18] The short-axis plane of the heart can be found by placing the transducer to connect the right lobe of the liver and the left shoulder (Figure 19.3). The transducer is moved upward or downward along the fetal thorax with some cranial or caudal tilt until the aortic valve is located in the center of the cardiovascular section that visualizes the right atrium, right ventricle, main pulmonary artery, and right pulmonary artery. An alternative view is the modified short-axis view that is equivalent to the angiographic

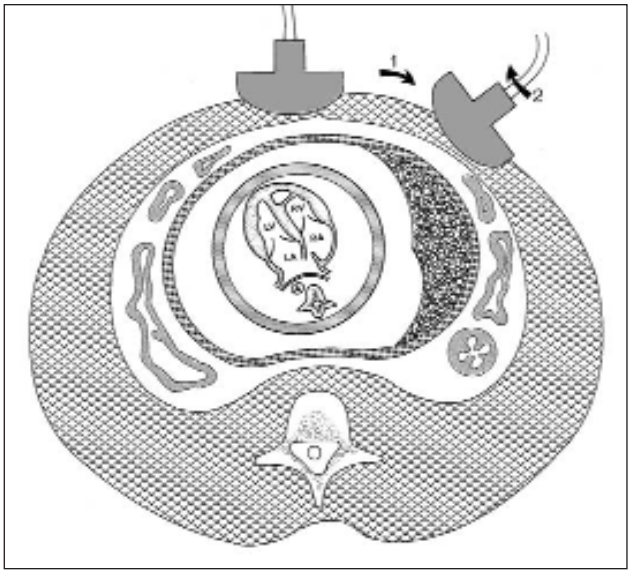
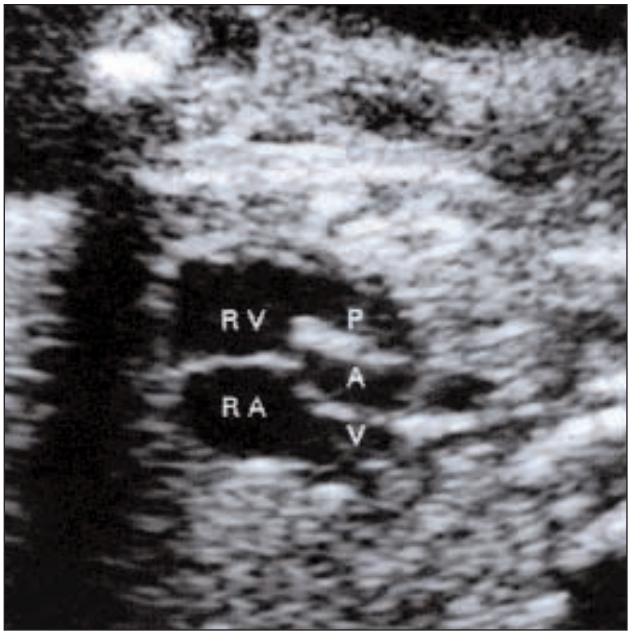


Figure 19.5
Diagram showing the maneuver for scanning the ventricular outflow tracts in a fetus in cephalic presentation and supine position. The examination starts from the position providing the four-chamber view. The transducer is moved radially around the maternal abdomen (arrow 1) until the ventricular septum is aligned perpendicular to the sonographic beam axis. Then the left ventricular outflow tract is obtained by rotating the transducer toward the cardiac apex (arrow 2). By sliding the transducer upward along the fetal thorax, the right ventricular outflow tract view is obtained. A, descending aorta; LA, left atrium; LV, left ventricle; RA, right atrium; RV, right ventricle. (Reprinted with permission from reference 21).



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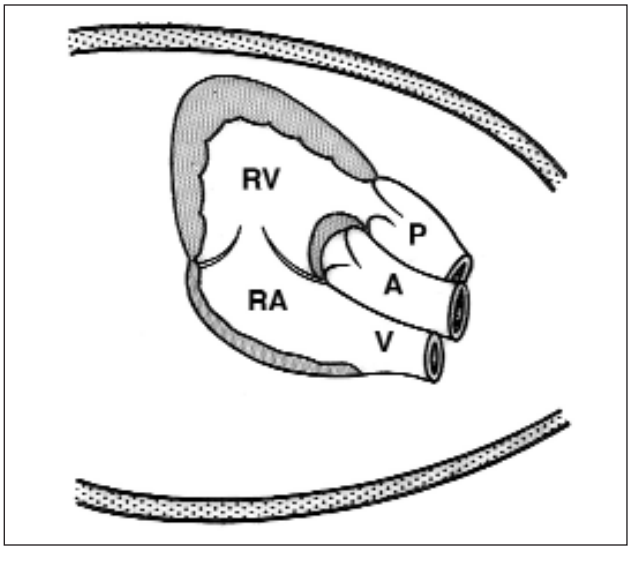

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Figure 19.6
Modified short-axis view and corresponding diagram. This semicoronal view is equivalent to the angiographic right anterior oblique view. The long-axis cuts of the three vessels are aligned from the left anterior to the right posterior aspects of the fetal thorax. A, aorta; P, pulmonary artery; RA, right atrium; RV, right ventricle; V, superior vena cava.

right anterior oblique view (Figure 19.6). This view can be obtained from the coronal view by displacing the transducer to the left anterior or right posterior part of the chest wall.

The three-vessel view is an orthogonal transverse view of the upper mediastinum, where normally the oblique section of the main pulmonary artery and cross-sections of the ascending aorta and superior vena cava are arranged in a straight line with a decreasing order of their size (Figure 19.4c).[19,20] This view is particularly helpful in the evaluation of the spatial relationship and size of the great arteries, which is abnormal in almost all cases with a ventricular outflow tract anomaly. The three-vessel view can be obtained simply by sliding the transducer upward from the four-chamber plane toward the fetal upper mediastinum.

Doppler interrogation is an important adjunct to two-dimensional imaging. It demonstrates the direction and velocity of the flow through the outflow tracts. It also demonstrates the flow through the ductus arteriosus, which is important for recognition of so-called 'ductus-dependent' lesions. Power and color Doppler mapping facilitates identification of the vessels.

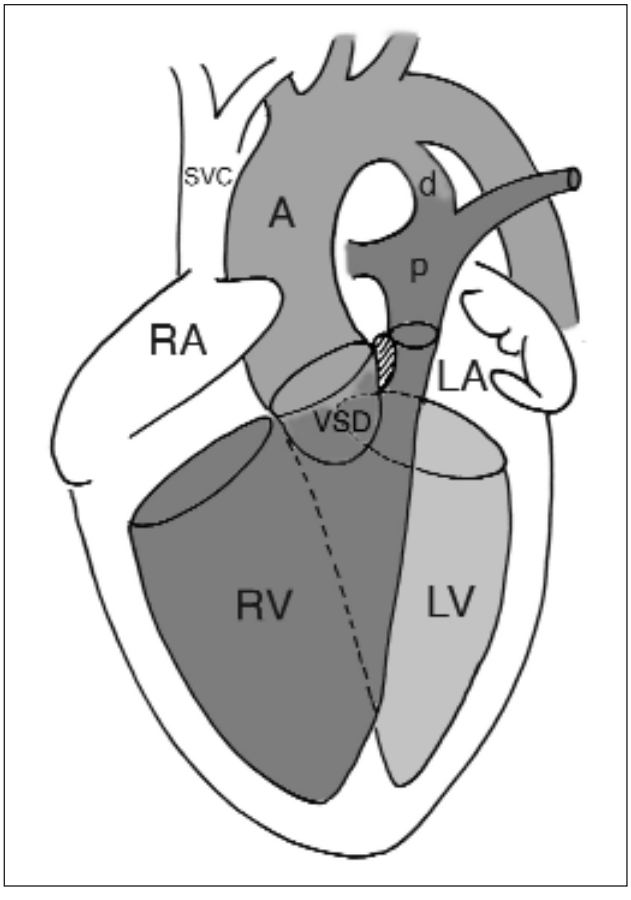


Figure 19.7
Diagram showing the pathology of tetralogy of Fallot. A, aorta; d, ductus arteriosus; LA, left atrium; LV, left ventricle; P, pulmonary artery; RA, right atrium; RV, right ventricle; SVC, superior vena cava; VSD, ventricular septal defect.

Individual lesions

Tetralogy of Fallot

The four anatomical features of tetralogy of Fallot are a ventricular septal defect, subpulmonary stenosis, over-riding aorta and right ventricular hypertrophy (Figure 19.7).[22] The essence of tetralogy is anterior, superior, and leftward deviation or malalignment of the outlet septum relative to the rest of the ventricular septum (Figure 19.8). The deviation of the outlet septum results in a large ventricular septal defect of the anterior malalignment type, and variable degrees of subpulmonary stenosis and aortic over-riding. Right ventricular hypertrophy is secondary to subpulmonary stenosis and a large ventricular septal defect.

In most cases with tetralogy, the situs is solitus and there is levocardia. The four-chamber view does not show any defect in the majority of cases. On close observation, however, one may note that the cardiac apex is deviated leftward (Figure 19.9).[23–26]

The ventricular septal defect in tetralogy involves the outlet part of the ventricular septum. It is most commonly a perimembranous defect, in which the tricuspid valve is in direct contact with the aortic valve through the posteroinferior border of the defect. The perimembranous defect is seen immediately below the overriding aortic valve in the left ventricular outflow tract view (Figure 19.10).[11,21,23,27] It is also seen in the short-axis view as a defect extending from the tricuspid annulus toward the ventricular outlet. The ventricular septal defect is less frequently a muscular outlet type, in which the posteroinferior border is the muscular rim

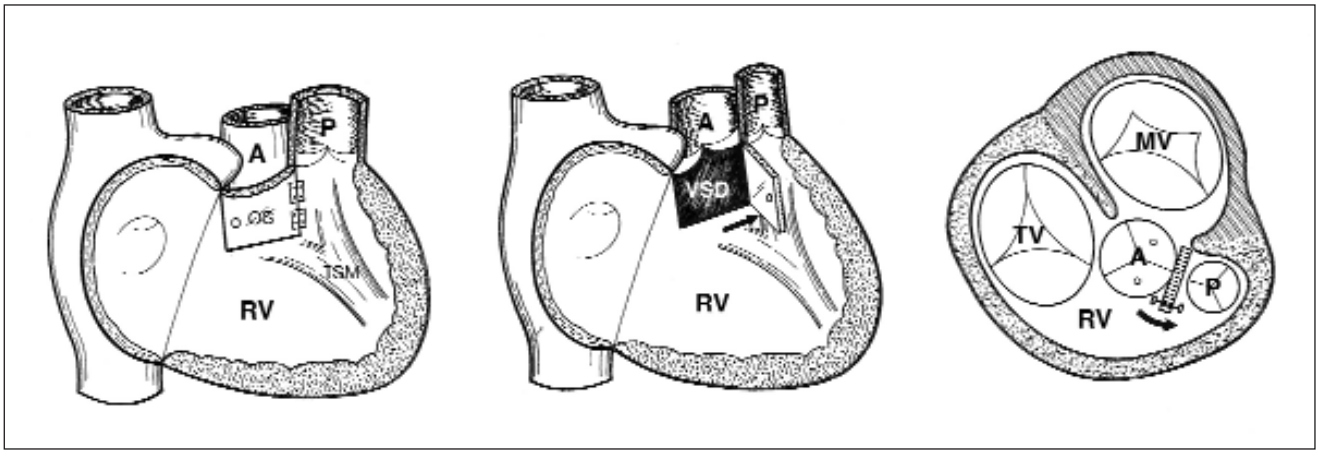


Figure 19.8
Essential pathology of tetralogy of Fallot. The outlet septum (OS) is supported by the hinges along its left anterior margin. It is pulled anteriorly into the right ventricular outflow tract. The aortic valve over-rides the ventricular septum because it is displaced anteriorly with the outlet septum. The deviated outlet septum encroaches on the subpulmonary outflow tract dimension. A, aorta; MV, mitral valve; P, pulmonary artery; RV, right ventricle; TSM, trabecula septomarginalis; TV, tricuspid valve.

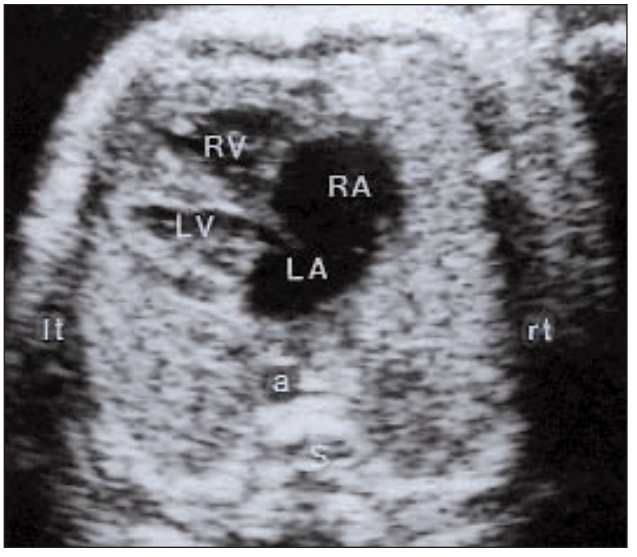
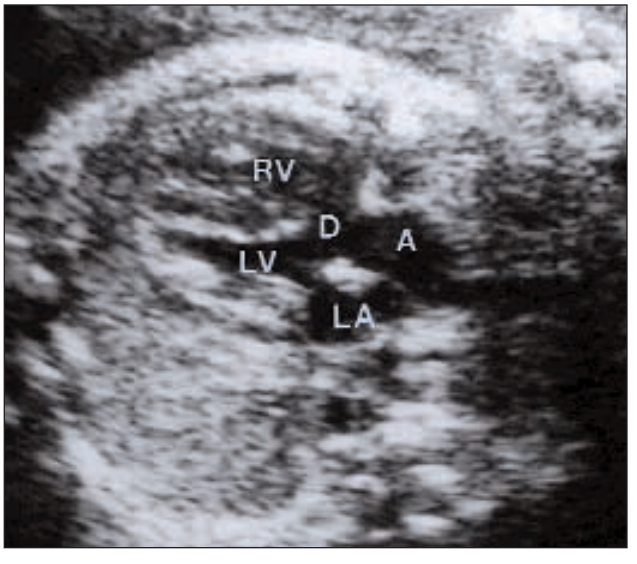


Figure 19.9
Tetralogy of Fallot. Four-chamber view shows no defect. The cardiac axis, however, is deviated to the left. a, descending aorta; LA, left atrium; lt, left; LV, left ventricle; RA, right atrium; rt, right; RV, right ventricle; S, spine. (Reprinted with permission from reference 23.)



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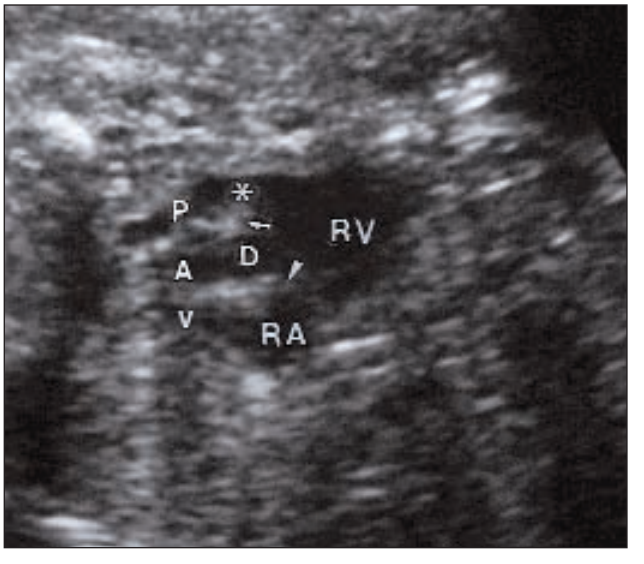

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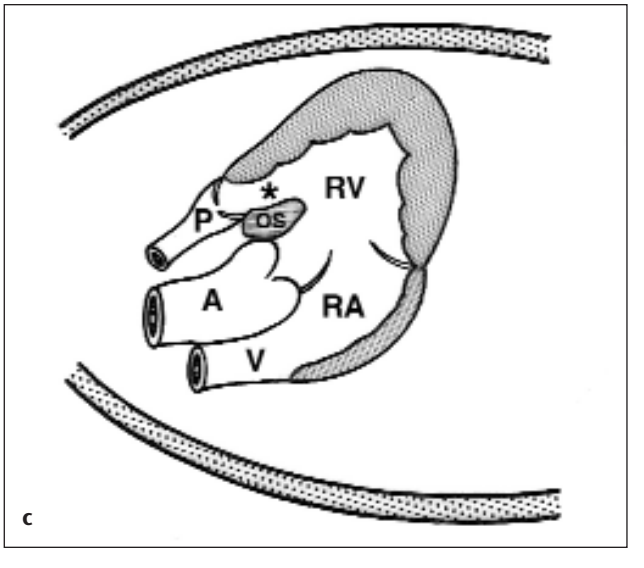

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Figure 19.10
Tetralogy of Fallot with a perimembranous ventricular septal defect. (**a**) Left ventricular outflow tract view clearly visualizes the ventricular septal defect (D). The aorta (A) over-rides the ventricular septum. (**b**, **c**) Modified short-axis view and corresponding diagram show that the subpulmonary outflow tract (asterisk) is encroached on by the deviated outlet septum (arrow in (**b**) and OS in (**c**)). The ventricular septal defect is seen behind the deviated outlet septum. The aortic and tricuspid valves are in direct contact (arrowhead). LA, left atrium; LV, left ventricle; P, pulmonary artery; RA, right atrium; RV, right ventricle; S, spine; V, superior vena cava. (Reprinted with permission from reference 23.)

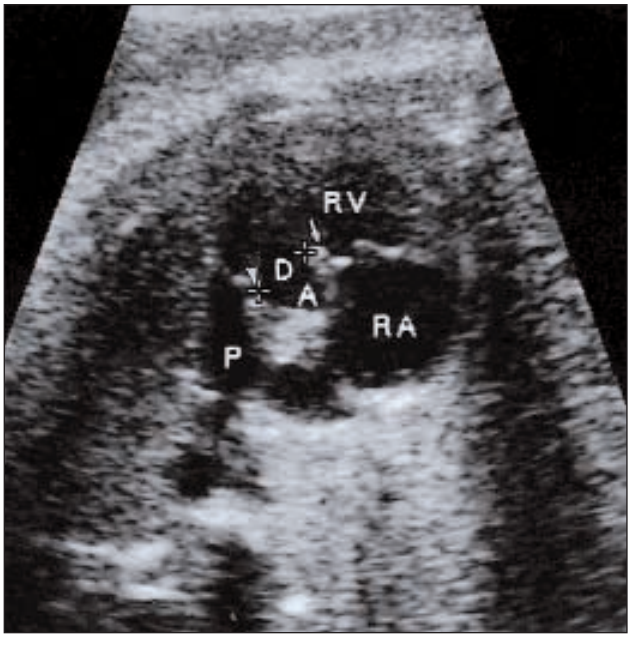


Figure 19.11
Tetralogy of Fallot with a doubly committed juxta-arterial ventricular septal defect. Short-axis view shows that the ventricular septal defect (D) involves the most cranial part of the ventricular septum. The aortic (A) and pulmonary (P) valves are in direct contact (arrowhead) through the defect. There is a muscular posteroinferior rim (arrow) between the aortic and tricuspid valves. The pulmonary valve annulus is smaller than the aortic valve annulus. RA, right atrium; RV, right ventricle. (Reprinted with permission from reference 23.)

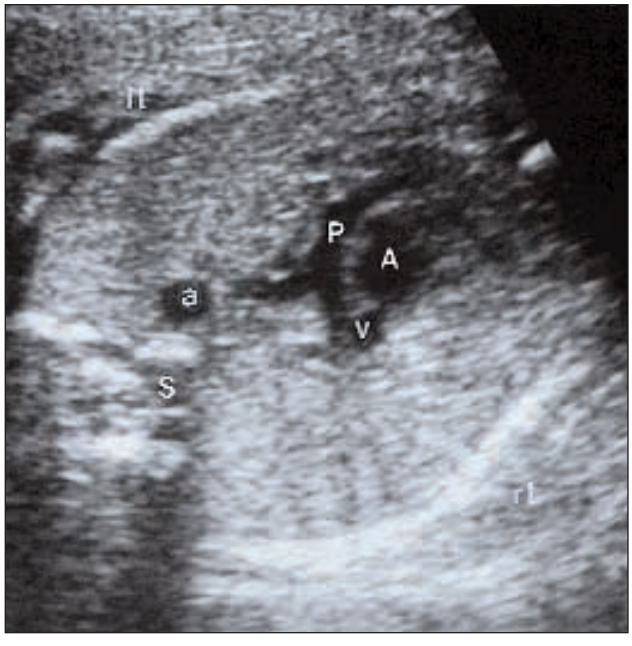


Figure 19.12
Tetralogy of Fallot. Three-vessel view shows the abnormal alignment and size of the three vessels. The ascending aorta (A) is dilated and displaced anteriorly. The main pulmonary artery (P) is small and displaced posteriorly. The branch pulmonary arteries show mild hypoplasia. a, descending aorta; lt, left; rt, right; S, spine; V, superior vena cava. (Reprinted with permission from reference 20.)

separating the tricuspid valve from the aortic valve. Least frequently, the defect is a doubly committed juxta-arterial type, which is characterized by the absence or extreme hypoplasia of the outlet septum. In this type, the aortic and pulmonary valves are in direct contact through the anterosuperior border of the defect. Both muscular outlet and doubly committed defects can not be demonstrated in the left ventricular outflow tract view but can be visualized in the short-axis view of the ventricular outflow tracts (Figure 19.11). Rarely, tetralogy may coexist with an atrioventricular septal defect. An additional muscular ventricular septal defect or defects may be present.

The essential pathology of tetralogy – the anterior, superior and leftward deviation of the outlet septum – can be demonstrated in the short-axis or right anterior oblique views (Figure 19.10b).[23] In this view, the ventricular septal defect is seen behind and below the deviated outlet septum. The deviated outlet septum encroaches on the subpulmonary outflow tract and pulmonary valve annulus. In addition, the pulmonary valve is commonly stenotic.

In the majority of cases, the ascending aorta is large and the main pulmonary artery is small.[19,20,23,28–30] The size discrepancy of the arterial trunks may not be apparent in the early stage of gestation but becomes evident with advancing gestation.[30] It has been documented that the diameters of the arterial trunks are often within normal range, suggesting that a dilated aortic root alone is not a sensitive marker of tetralogy.[27,30] Therefore, it is important to evaluate the size discrepancy by direct visual comparison or calculation of the diameter ratio.[27,29] We find direct visual comparison much easier than measurement. We use the three-vessel view for this purpose in every case (Figure 19.12).[19,20,23] Dextroposition of the aorta can also be easily recognized in the three-vessel view. As the dilated aorta is displaced anteriorly and the main pulmonary artery more or less posteriorly, the alignment of the three vessels is almost always abnormal. In our previous study, the clue to the diagnosis of tetralogy was found most commonly in the three-vessel view.[23] In contrast to the small size of the main pulmonary artery, the branch pulmonary artery size is usually normal.[30] A small branch pulmonary artery at midgestation suggests severe disease (Figure 19.13).

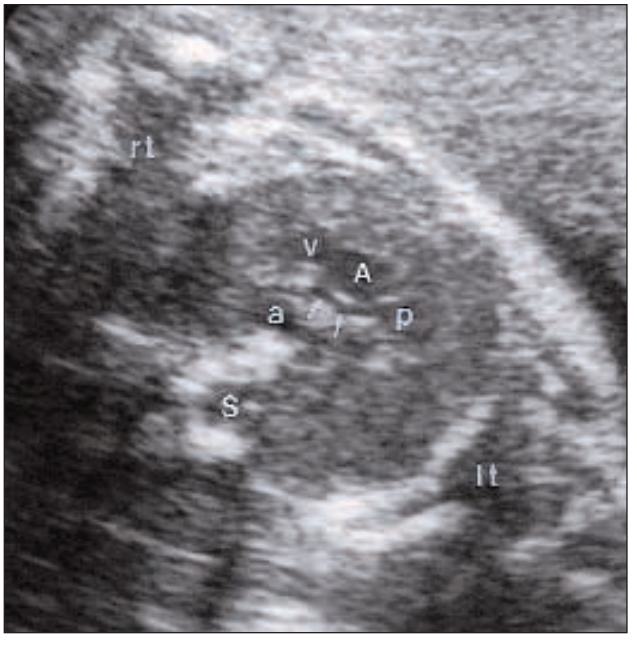


Figure 19.13
Tetralogy of Fallot with hypoplastic branch pulmonary arteries. Three-vessel view shows the abnormal alignment and size of the three vessels. The right pulmonary artery (arrows) is tiny. It progressed to pulmonary atresia in the third trimester. The descending aorta (a) is seen in the right anterior aspect of the spine (S) because there is a right aortic arch. A, ascending aorta; lt, left; rt, right; V, superior vena cava.

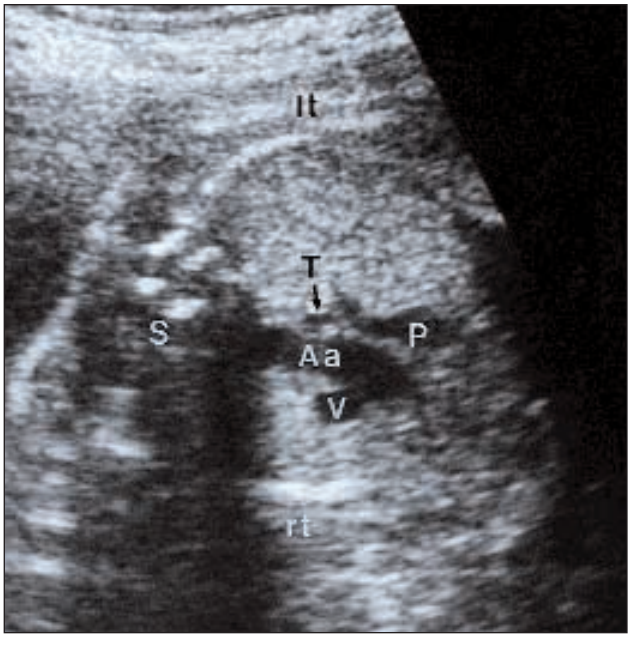


Figure 19.14
Right aortic arch in a fetus with tetralogy of Fallot. Transverse view of the upper thorax shows the aortic arch (Aa) to the right of the trachea (T). lt, left; P, main pulmonary artery; rt, right; S, spine; V, superior vena cava. (Reprinted with permission from reference 23.)

A mirror-imaged right aortic arch is common in tetralogy. When there is a right aortic arch, the descending aorta is seen on the right anterior aspect of the spine in the three-vessel view (Figure 19.13). As the trachea is filled with fluid in the fetus, the position of the aortic arch relative to the trachea can be directly visualized (Figure 19.14).[19–21] Rarely, the right or left subclavian artery may arise aberrantly from the descending aorta. The aberrant branch always courses behind the esophagus and thus is seen behind the trachea.

The ductus arteriosus is characteristically small or not identifiable in tetralogy.[23,30] The ductus often arises from the undersurface of the aortic arch (Figure 19.15). In most cases with a right aortic arch, the patent ductus is the left ductus that arises from the left innominate or subclavian artery. The blood flow through the ductus varies considerably according to the severity of subpulmonary stenosis.[23,31] The ductal flow may be right-to-left, bidirectional or left-to-right. The ductus is rarely, if ever, patent when tetralogy is complicated by absent pulmonary valve syndrome.[32–34] It is interesting, however, to note that a ductus is usually patent when absent pulmonary valve syndrome occurs with a simple ventricular septal defect.[35]

Many other lesions may be associated with tetralogy of Fallot. A secundum type of atrial septal defect may be present

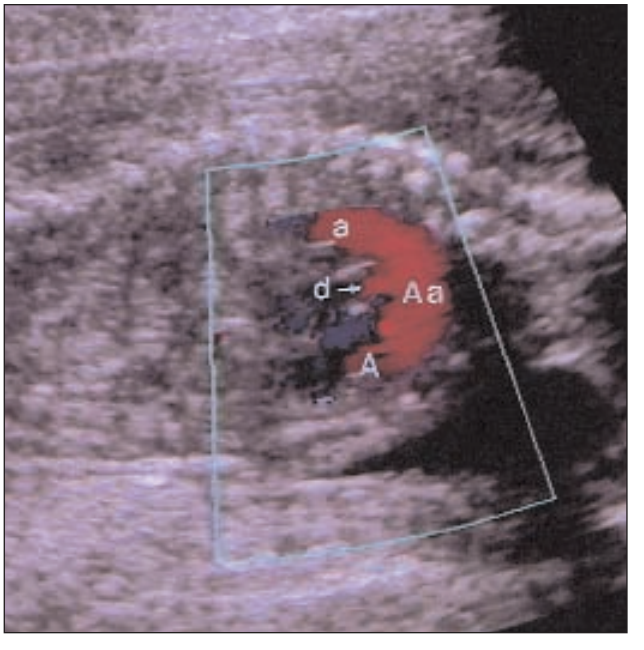
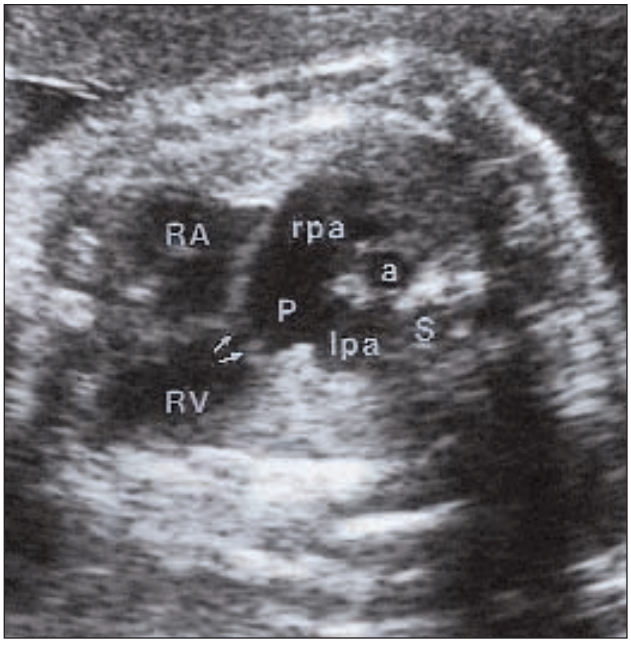


Figure 19.15
Ductus arteriosus in tetralogy of Fallot. Aortic arch view with color Doppler shows that the ductus arteriosus (d) is small and arises from the undersurface of the aortic arch (Aa). A, ascending aorta; a, descending aorta. (Reprinted with permission from reference 23.)



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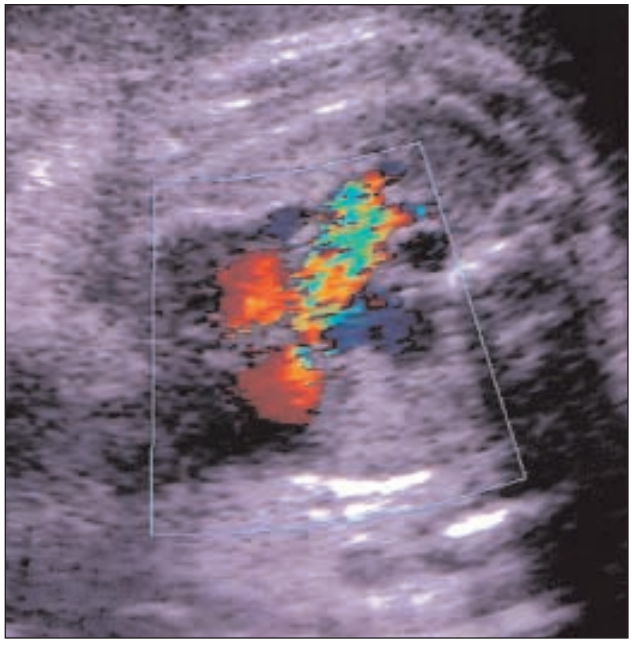

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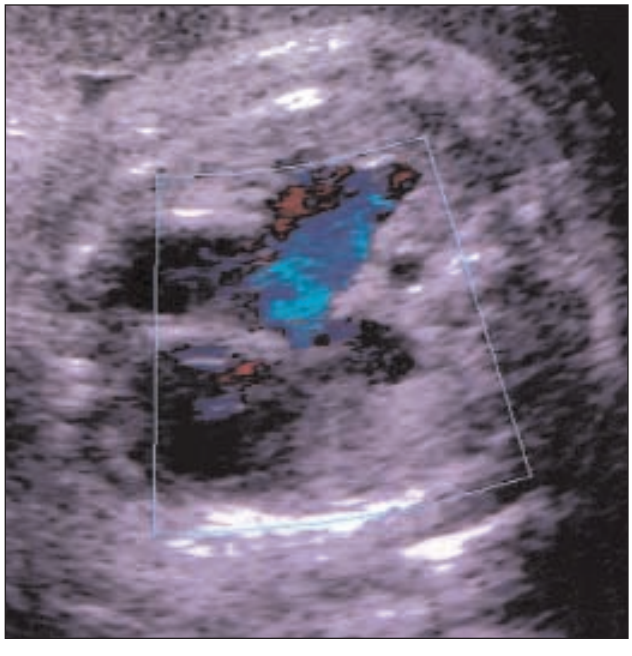

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Figure 19.16
Absent pulmonary valve syndrome in tetralogy of Fallot. (**a**) Transverse view of the right ventricular outflow tract shows marked dilatation of the main pulmonary artery (P) and branch pulmonary arteries (rpa and lpa). The pulmonary valve is guarded by the dysplastic and deficient leaflets (arrows). (**b**, **c**) Systolic and diastolic frames of color Doppler examination in the same imaging plane show turbulent forward flow in the right pulmonary artery, forward flow in the systolic phase, and reversed flow in the diastolic phase. a, descending aorta; RA, right atrium; RV, right ventricle; S, spine.

but cannot be diagnosed in the fetus. One pulmonary artery, usually the left, may be disconnected from the main pulmonary artery and connected to the ipsilateral ductus arteriosus. As mentioned, absent pulmonary valve syndrome may complicate tetralogy of Fallot.[32–35] Absent pulmonary valve syndrome is characterized by vestigial leaflets guarding the small pulmonary valve annulus, gross pulmonary regurgitation, and aneurysmal dilatation of the pulmonary arteries in the mediastinum (Figure 19.16). The dilated pulmonary arteries taper down abruptly in the lungs. The dilated pulmonary artery may be mistaken for a cystic mass in the mediastinum.[36] As the pulmonary artery compresses the tracheobronchial tree and esophagus, polyhydramnios may develop.[33] Because of severe pulmonary regurgitation, the heart is large, with right ventricular dilatation.

Pulmonary atresia with ventricular septal defect is an extreme form of tetralogy. It has been documented that tetralogy with a patent pulmonary outflow tract may progress to pulmonary atresia in fetal life.[23, 30] Pulmonary atresia may be at either infundibular or valvular level. The

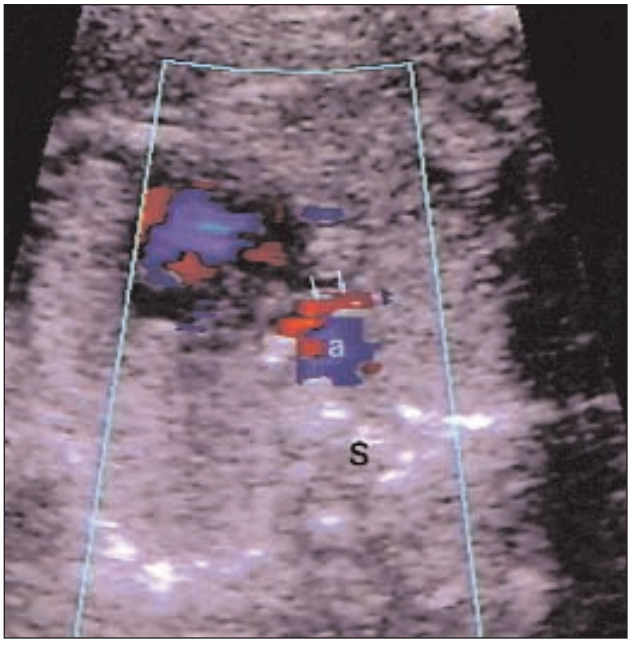


Figure 19.17
Major aortopulmonary collateral artery in pulmonary atresia with ventricular septal defect. Transverse view of thorax with color Doppler shows that the collateral artery (arrow) arises from the descending aorta (a) and supplies a part of the right lung. S, spine. (Reprinted with permission from reference 23.)

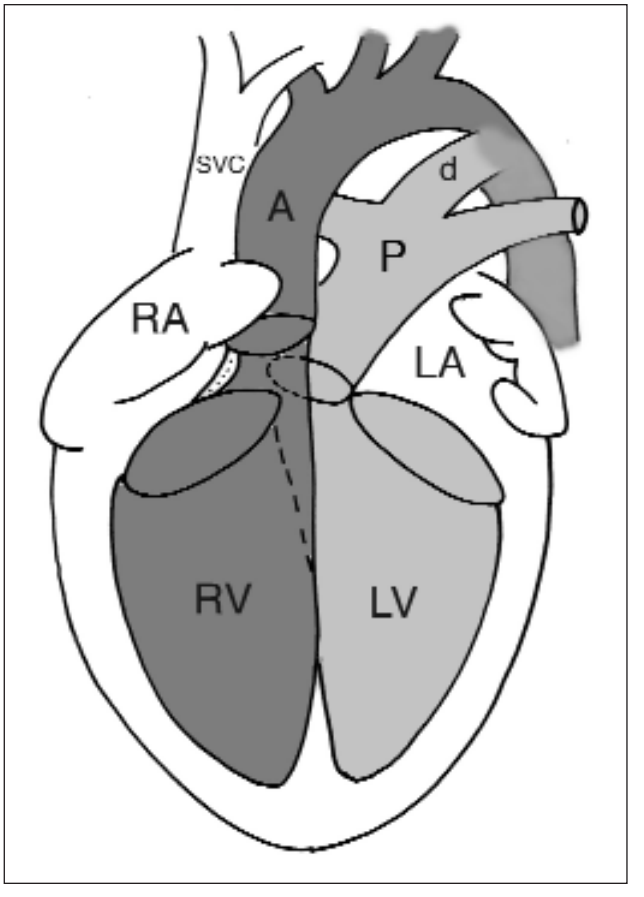


Figure 19.18
Diagram showing pathology of complete transposition of the great arteries. A, aorta; d, ductus arteriosus; LA, left atrium; LV, left ventricle; P, pulmonary artery; RA, right atrium; RV, right ventricle; SVC, superior vena cava.

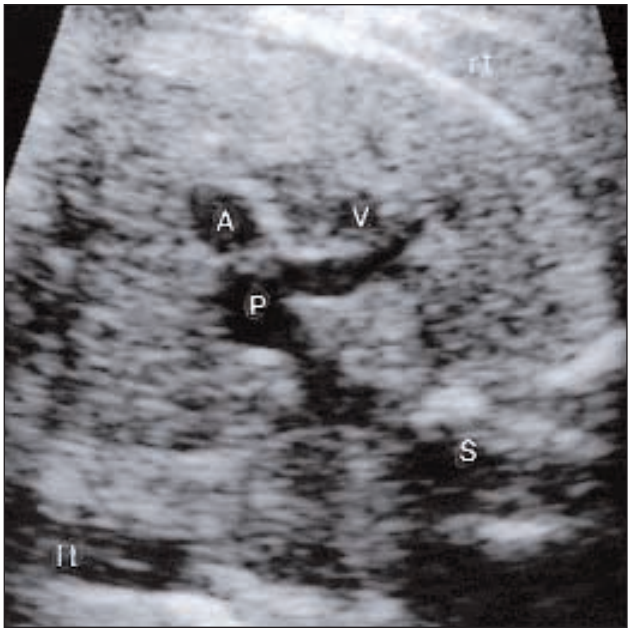
main pulmonary artery is often not formed and the branch pulmonary arteries may not be confluent. The source of blood flow to the lungs may be either the ductus arteriosus or the major aortopulmonary collateral arteries. The collateral arteries arise most commonly from the descending thoracic aorta, less often from the bracheocephalic branches, and rarely from the coronary artery. Color or power Doppler interrogation facilitates demonstration of the ductus or collateral arteries supplying the lungs (Figures 19.15 and 19.17). It is of importance that a ductus arteriosus connects with the pulmonary arteries in the mediastinum, whereas most collateral arteries connect to the pulmonary arteries at the hilum or within the lungs.

Among the various echocardiographic findings, those highly predictive of severe postnatal disease are main pulmonary artery hypoplasia at the initial examination, little or no growth of the main and branch pulmonary arteries on follow-up, and retrograde ductus arteriosus flow.[30]

Complete transposition of the great arteries

The word 'transposition' is a compound of *trans* meaning across and *ponere* meaning to place.[37] Transposition of the great arteries indicates a condition in which the great arteries are placed across the ventricular septum, and therefore the aorta arises from the right ventricle and the pulmonary artery from the left ventricle.[37,38] 'Transposition of the great arteries' is synonymous with the term 'discordant ventriculoarterial connection'. When transposition of the great arteries occurs with concordant atrioventricular connection, it is called complete transposition (Figure 19.18). When it occurs with discordant atrioventricular connection, it is called corrected transposition.

In most cases with complete transposition, the situs is solitus and there is levocardia. The right ventricle is the right-sided and anterior ventricle. The four-chamber view is usually normal when there is no associated lesion.[39,40] The great arterial relationship is almost always abnormal, which can be easily recognized at the three-vessel view (Figure 19.19a).[16,19–21] The great arterial relationship is closely related to the infundibular morphology. In classic



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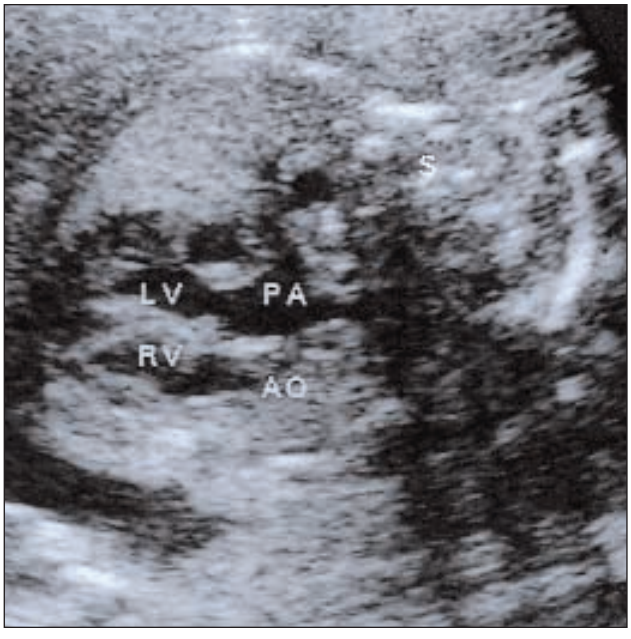

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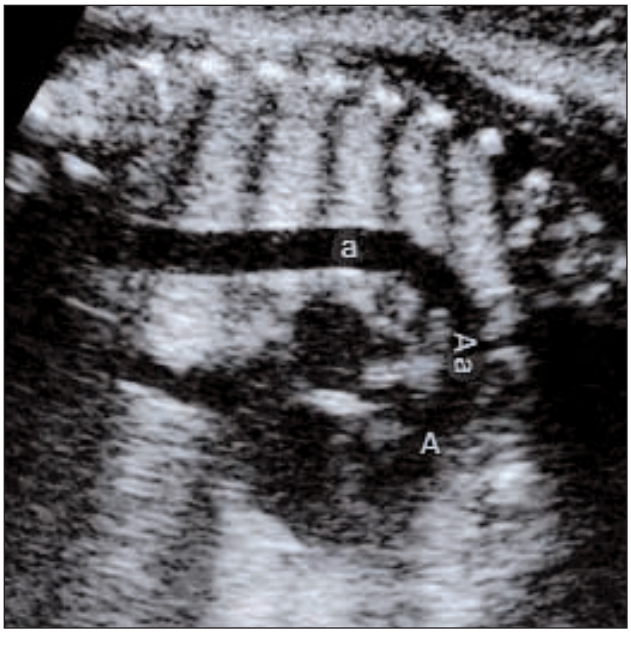

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Figure 19.19
Complete transposition of the great arteries. (**a**) Three-vessel view shows the classic great arterial relationship of complete transposition. The three vessels are arranged in a triangular fashion with the right anterior aorta (A) and left posterior pulmonary artery (P). (**b**) Ventricular outflow tract view shows discordant ventriculoarterial connection. As the right and left ventricular outflow tracts are parallel to each other, they are imaged in a single plane. (**c**) Sagittal view of the thorax shows the aortic arch (Aa). It forms a wide arc with the configuration of a hockey stick. It gives rise to the head and neck branches. a, descending aorta; AO, ascending aorta; lt, left; LV, left ventricle; PA, pulmonary artery; rt, right; RV, right ventricle; S, spine; V, superior vena cava. (**a**, reprinted with permission from reference 20.)

transposition, the aorta is supported by a completely muscular infundibulum, while the pulmonary artery is not. The pulmonary valve is in fibrous continuity with the mitral valve. In this classic setting, the aorta is anterior to and to the right of the pulmonary artery. Rarely, the aorta may be located posterior to or to the left of the pulmonary artery.[37,38] The ventricular outflow tracts are usually parallel, which allows visualization of both outflow tracts in a single imaging plane (Figure 19.19b).[16,21,39] As mentioned, normal outflow tracts cross each other and can not be visualized in a single imaging plane. The aortic arch in complete transposition is shaped like a hockey stick and can be mistaken for a ductal arch (Figure 19.19c).

In approximately 40% of the cases with complete transposition, there is an associated ventricular septal defect (Figure 19.20). It may occupy any part of the ventricular septum. Particularly common is the outlet defect with posterior or anterior malalignment of the outlet septum. When there is posterior malalignment, the subpulmonary outflow tract is narrowed and the aortic valve may over-ride the ventricular septum (Figure 19.20). When there is anterior malalignment,

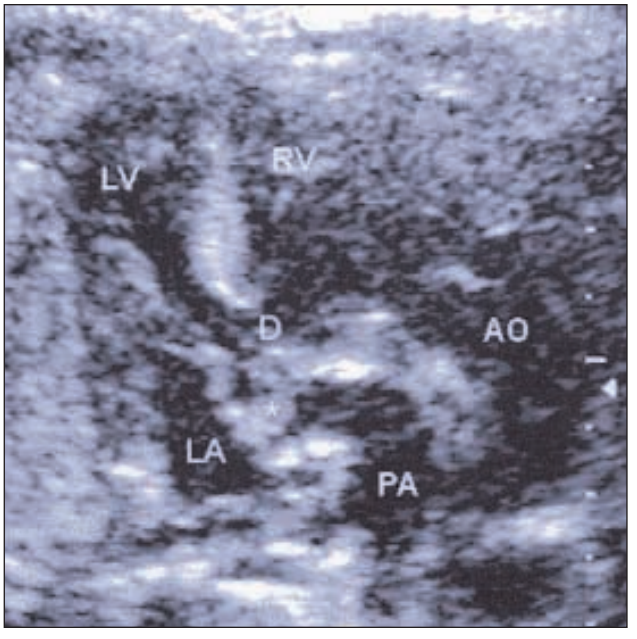


Figure 19.20
Complete transposition of the great arteries with ventricular septal defect and left ventricular outflow tract obstruction. Ventricular outflow tract view shows discordant ventriculoarterial connection. The left ventricular outflow tract is narrow because of the posteriorly deviated outlet septum. The pulmonary valve is guarded by dysplastic leaflets (asterisk). AO, aorta; d, ventricular septal defect; LA, left atrium; LV, left ventricle; PA, pulmonary artery; RV, right ventricle.

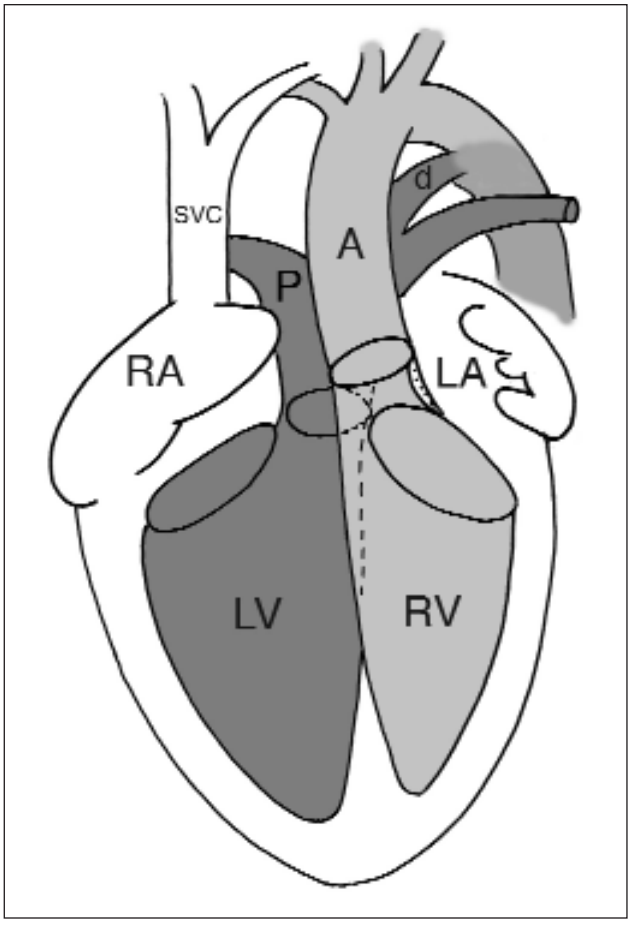


Figure 19.21
Diagram showing the pathology of corrected transposition of the great arteries. A, aorta; d, ductus arteriosus; LA, left atrium; LV, left ventricle; P, pulmonary artery; RA, right atrium; RV, right ventricle; SVC, superior vena cava.

the subaortic outflow tract is narrowed and the pulmonary valve may over-ride the ventricular septum. With a greater degree of over-riding of the aortic or pulmonary valve, transposition merges into double-outlet left or right ventricle.

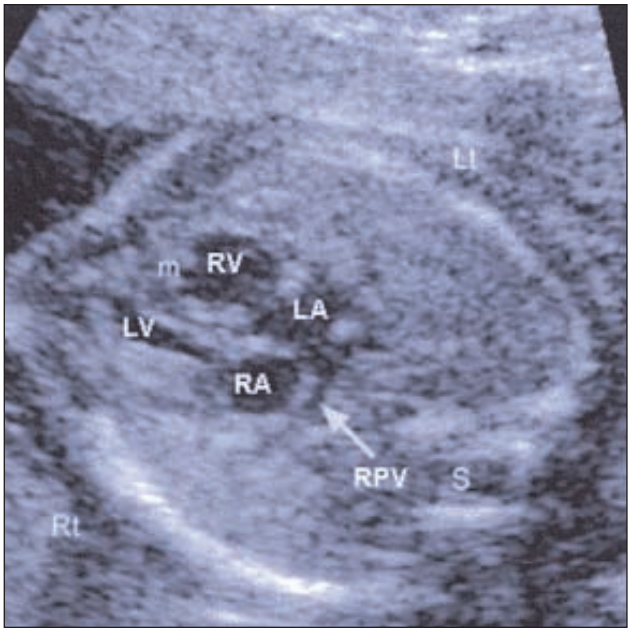
Left ventricular outflow tract obstruction is common in complete transposition. As mentioned, it may be due to posterior malalignment of the outlet septum. Obstruction due to a fibrous ridge, a fibromuscular tunnel or accessory mitral valve tissue can exist with an intact ventricular septum as well as in association with a ventricular septal defect. With a significant left ventricular outflow tract obstruction, the main pulmonary artery is smaller than the ascending aorta. The pulmonary valve may be stenotic (Figure 19.20).

Subaortic obstruction is less common. It may be due to anterior malalignment of the outlet septum. The obstruction is often dynamic. When there is subaortic stenosis, the aortic arch should be carefully searched for any obstructive lesion. Hypoplasia of the morphological right ventricle is also seen in this setting.

Corrected transposition of the great arteries

Corrected transposition is a combination of a discordant atrioventricular connection and discordant ventriculo-arterial connection (Figure 19.21).[41] Because of discordant connections at two levels, the physiological abnormality intrinsic to each discordant connection is 'corrected'.

In most cases, the situs is solitus. Although it occurs most commonly with levocardia, dextrocardia and mesocardia are not uncommon.[42] The majority of the cases with an unexpected cardiac position for the given visceral situs—i.e. dextrocardia or mesocardia in situs solitus and levocardia or mesocardia in situs inversus—will have corrected transposition (Figure 19.22). Discordant atrioventricular connection can readily be recognized at the four-chamber view (Figure 19.22a).[21,42,43] In situs solitus, the left-sided ventricle is the morphological right ventricle that can be identified by the



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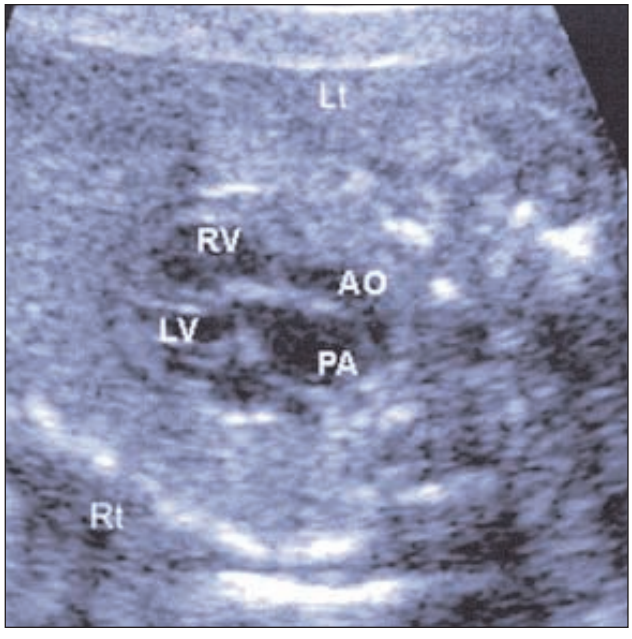

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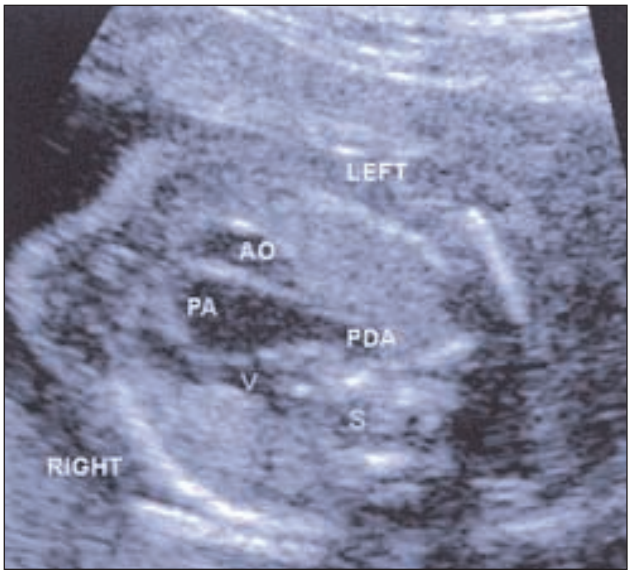

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Figure 19.22
Corrected transposition of the great arteries. (**a**) Four-chamber view shows that the cardiac apex points toward the right anterior chest. There is a discordant atrioventricular connection. The apical part of the left-sided ventricle is obliterated by the moderator band (m), suggesting that it is the morphological right ventricle (RV). (**b**) Ventricular outflow tract view shows that the right and left ventricular outflow tracts are parallel and connected to the aorta (AO) and pulmonary artery (PA), respectively. (**c**) Three-vessel view shows that the aorta is located leftward and slightly anterior to the pulmonary artery. LA, left atrium; LV, left ventricle; Lt, left; PDA, patent ductus arteriosus; RA, right atrium; RPV, right pulmonary vein; Rt, right; S, spine; V, superior vena cava.

presence of a moderator band and more apical attachment of its atrioventricular valve to the ventricular septum. Occasionally, the ventricles may be related in a superior–inferior fashion.[41] The aorta is usually supported by a completely muscular infundibulum, while the pulmonary artery is not (Figure 19.22b). The pulmonary valve is in fibrous continuity with the mitral valve. In the majority of cases, the aorta is located anterior to and to the left of the pulmonary artery, which can easily be recognized at the three-vessel view (Figure 19.22c).[19–21] The ventricular outflow tracts are usually parallel, which allows visualization of both outflow tracts in a single imaging plane (Figure 19.22b).

Approximately 60–70% of cases are associated with a ventricular septal defect. Although any type of defect can be associated, a perimembranous defect is the most common type. It tends to extend toward the ventricular inlet. Left ventricular outflow tract obstruction is also common. The nature of the obstruction may be a fibrous ridge, a fibromuscular tunnel, an aneurysm of the membranous septum, or accessory mitral valve tissue. The pulmonary valve may also be stenotic. It may infrequently be complicated by pulmonary atresia. Right ventricular outflow tract obstruction is less common.[41,43] The left-sided tricuspid valve is frequently abnormal.

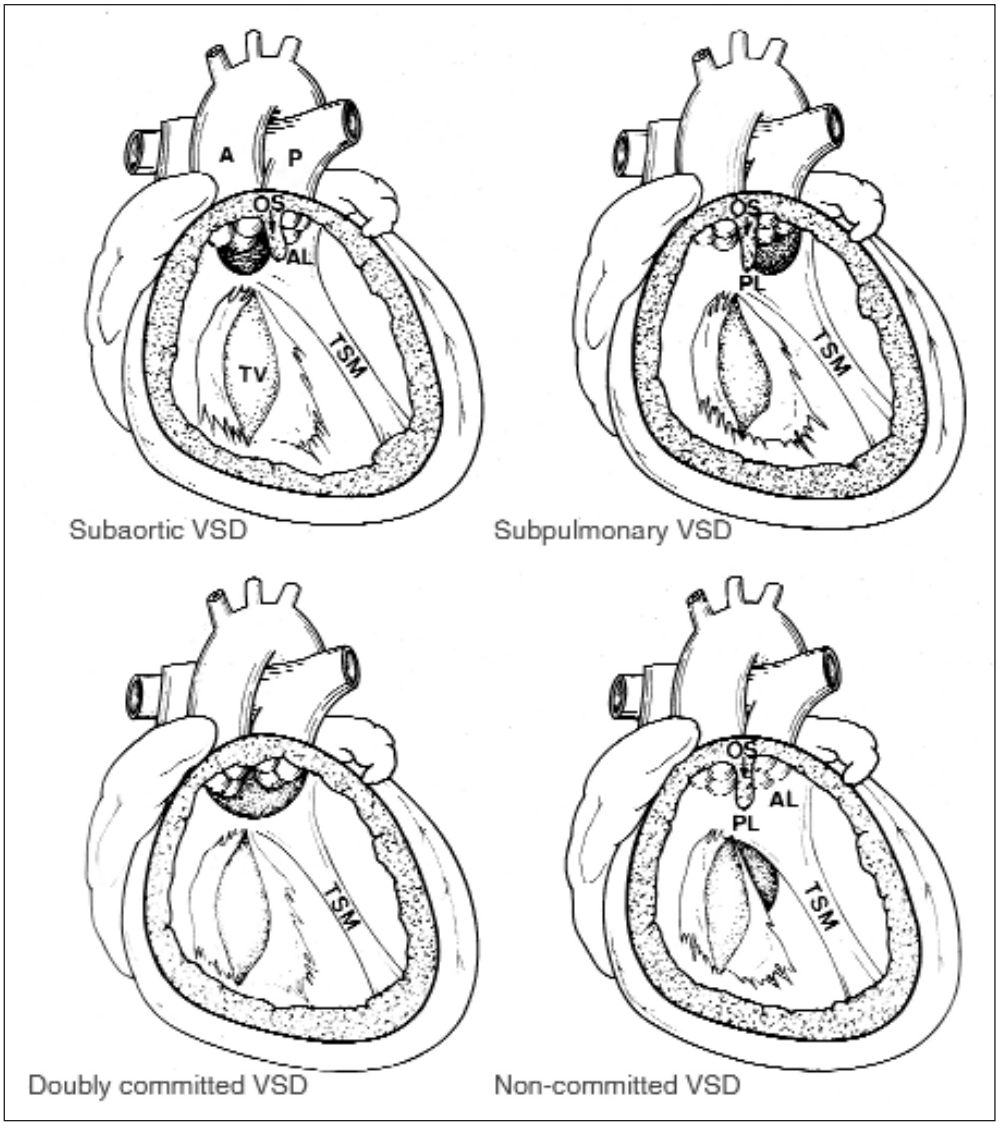


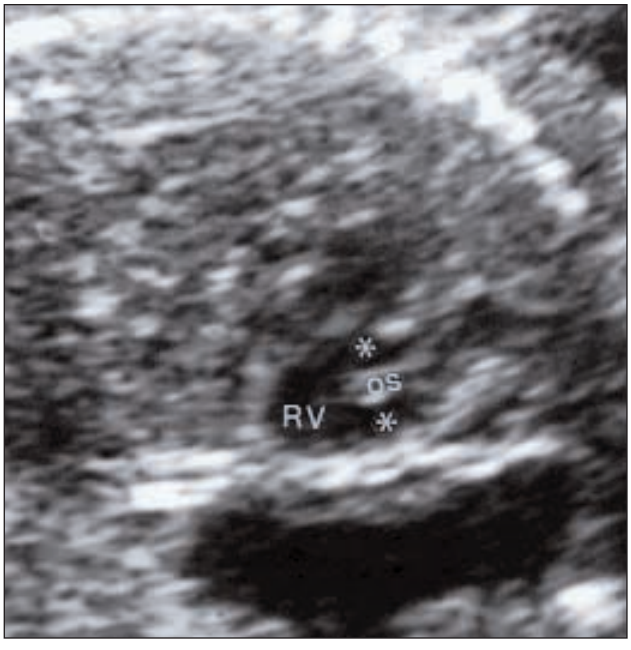
Figure 19.23
Diagrams showing pathology of double-outlet right ventricle. The ventricular septal defect is classified according to its location relative to the semilunar valves. A, aorta; AL, anterior limb of trabecula septomarginalis; OS, outlet septum; P, pulmonary artery; PL, posterior limb of trabecula septomarginalis; TSM, trabecula septomarginalis; TV, tricuspid valve.

The commonest pathology is valve dysplasia with a variable degree of regurgitation. Association of corrected transposition with Ebstein's anomaly of the tricuspid valve is well known. Delay or blocking of atrioventricular conduction is another common complication of corrected transposition. It has been documented that the incidence of arrhythmia increases with age. Its incidence in fetal life, however, has not been documented.

Double-outlet ventricle

Double-outlet right or left ventricle is a type of ventriculoarterial connection in which both great arteries arise from the morphological right or left ventricle (Figure 19.23).[44] In defining the ventriculoarterial connections, a great artery is considered to be connected to a ventricle when more than half of its valve is committed to that ventricle.

Double-outlet right ventricle can occur with any atrial situs and any atrioventricular connection. It often occurs in heterotaxy syndrome. However, it most commonly occurs with situs solitus and a concordant atrioventricular connection. In almost all cases, a large ventricular septal defect is part of the anomaly. The hemodynamic physiology of the double-outlet right ventricle after birth is determined mainly by the location of the ventricular septal defect in relation to the great arterial valves, although the presence of outflow tract obstruction is an additional important contributing factor. The defect may be subaortic, subpulmonary, doubly committed, or non-committed or remote in location (Figure 19.23). When



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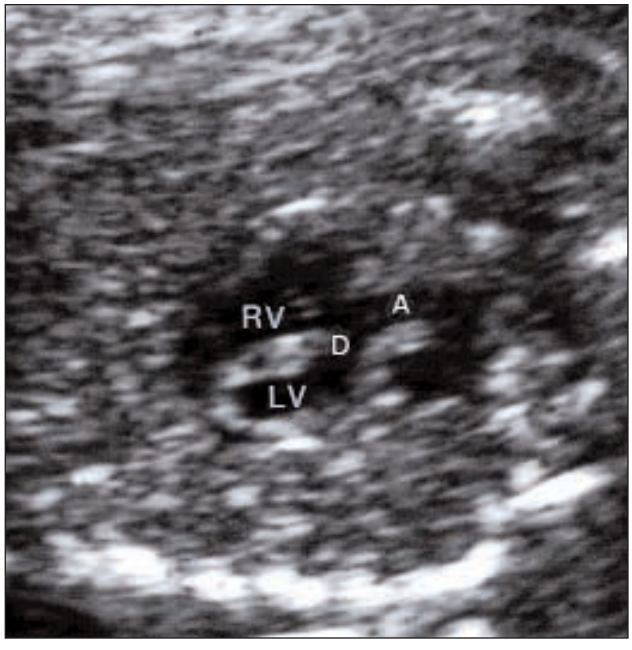

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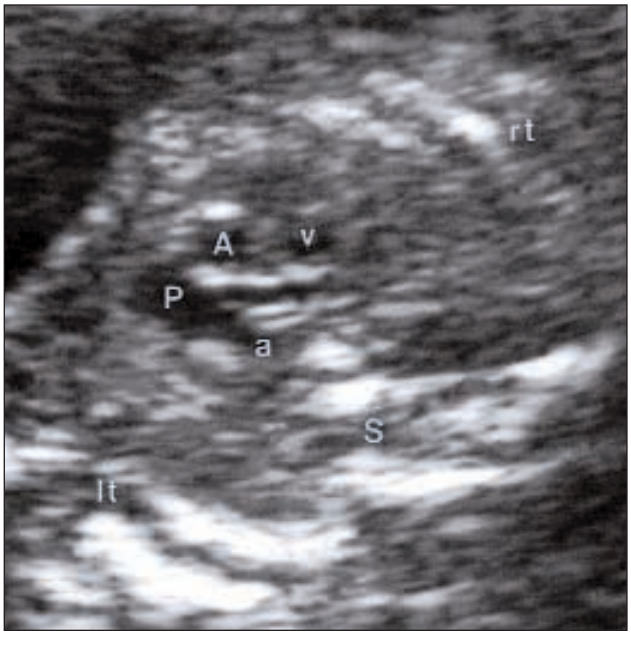

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Figure 19.24
Double-outlet right ventricle with subaortic ventricular septal defect. (**a**) Coronal view of the outlet of the right ventricle (RV) shows that it is divided into the subaortic and subpulmonary outflow tracts (asterisks) by the outlet septum (OS). (**b**) Left ventricular outflow tract view shows that the ventricular septal defect (D) is committed to the aorta (A). (**c**) Three-vessel view shows abnormal alignment of the three vessels with side-by-side aorta and pulmonary artery (P). a, descending aorta; lt, left; LV, left ventricle; rt, right; S, spine; V, superior vena cava. ((a) and (c) reprinted with permission from reference 19.)

there is a subaortic defect, the physiology is that of an isolated ventricular septal defect. When there is a subpulmonary defect, the physiology is that of complete transposition with a ventricular septal defect. A doubly committed ventricular septal defect is large and opens beneath both arterial valves. As the outlet septum is deficient or hypoplastic with this defect, a common right ventricular outflow tract leads to the great arteries. A non-committed ventricular septal defect is remote from both arterial valves. It involves either the inlet or the trabecular part of the ventricular septum. An atrioventricular septal defect is another form of non-committed defect. When the defect is doubly committed or non-committed, the physiology depends on the intracardiac streaming of blood flow. Rarely, the ventricular septal defect may be restrictive. Extremely rarely, a double outlet can be present with an intact ventricular septum.

In situs solitus, the aorta is usually located to the right of the pulmonary artery. They tend to have a side-by-side relationship (Figure 19.24). However, any relationship

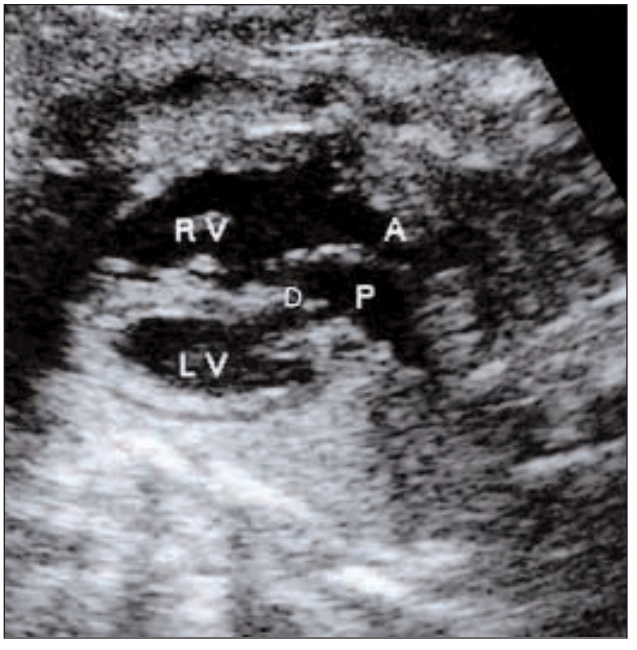


Figure 19.25
Double-outlet right ventricle with subpulmonary ventricular septal defect. Ventricular outflow tract view shows that the aorta (A) arises entirely from the right ventricle (RV). The ventricular septal defect (D) is immediately below the over-riding pulmonary artery (P). LV, left ventricle.

between the great arteries can be found. Both arterial trunks tend to have a parallel orientation.[16,39,45,46] In many cases, both great arterial valves are supported by a completely muscular infundibulum. Some consider that a bilateral infundibulum is the hallmark of double-outlet right ventricle. Most, however, have abandoned this definition. There has been some debate on the relationship between tetralogy of Fallot and double-outlet right ventricle. In this context, one should remember that the term 'tetralogy' describes the infundibular morphology, while the term 'double-outlet right ventricle' describes the type of ventriculoarterial connection.[22,44] Therefore, the type of ventriculoarterial connection in a case of tetralogy with more than 50% of aortic over-riding is unequivocally a double-outlet right ventricle.

Double-outlet right ventricle with a subaortic ventricular septal defect (Figure 19.24) is often associated with subpulmonary obstruction. Many of these combinations show an infundibular morphology that is similar or identical to that of tetralogy. In contrast, double-outlet right ventricle with a subpulmonary ventricular septal defect (Figure 19.25) is often associated with subaortic obstruction.[45] When there is subaortic obstruction, an obstructive lesion of the aortic arch is common. The obstruction in either circumstance is usually due to a deviated outlet septum, but there may be associated arterial valvular stenosis. Subpulmonary or subaortic obstruction can be suspected when one great artery is significantly smaller than the other at the three-vessel view.

Double-outlet left ventricle is extremely rare.[47] The ventricular septal defect is most commonly subaortic and less frequently subpulmonary in location. Doubly committed and non-committed defects are rare. Extremely rarely, the ventricular septum is intact. It was once considered that a bilaterally deficient infundibulum was the hallmark of double-outlet left ventricle. It was confirmed, however, that any infundibular morphology could be found in double-outlet left ventricle and that a subpulmonary or subaortic infundibulum was more common than a bilaterally deficient infundibulum.

Truncus arteriosus

Truncus arteriosus is a condition in which one arterial trunk arises from the base of the ventricles via a single

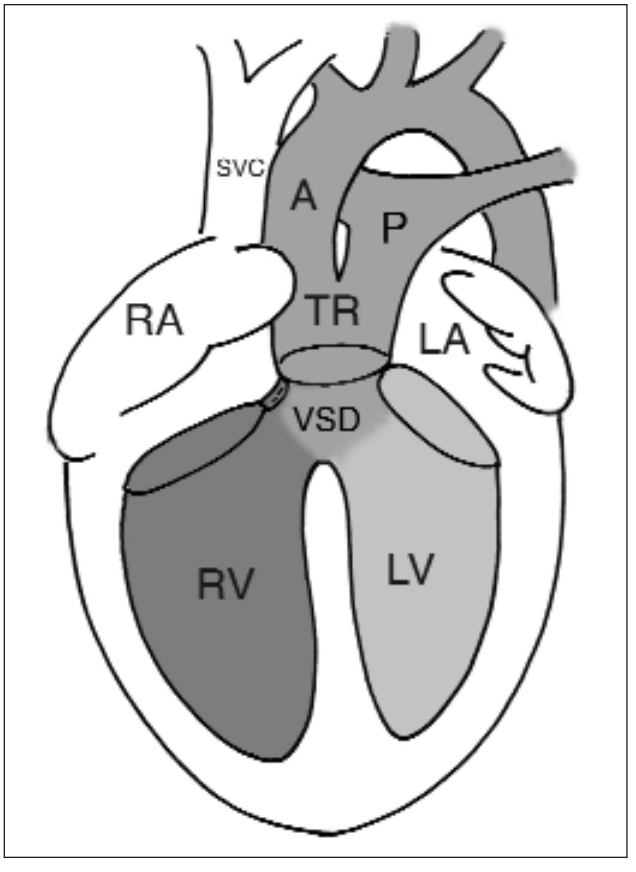
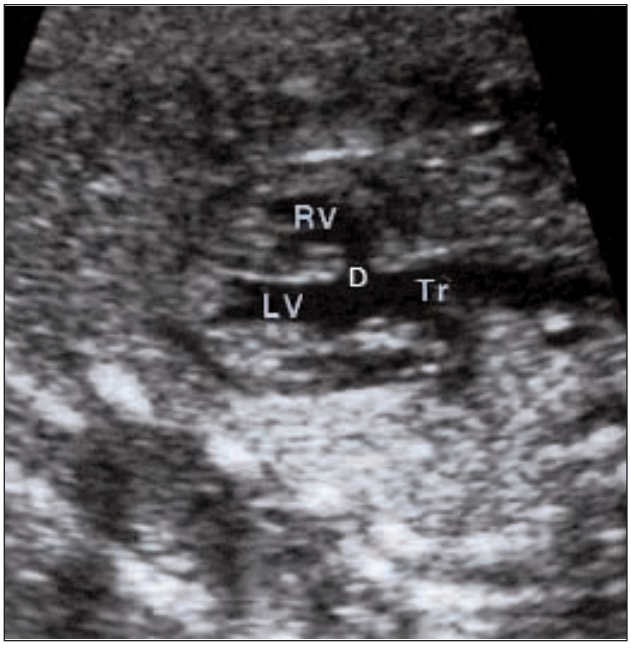


Figure 19.26
Diagram showing the pathology of the truncus arteriosus. A, aorta; LA, left atrium; LV, left ventricle; P, pulmonary artery; RA, right atrium; RV, right ventricle; SVC, superior vena cava; TR, truncus; VSD, ventricular septal defect.



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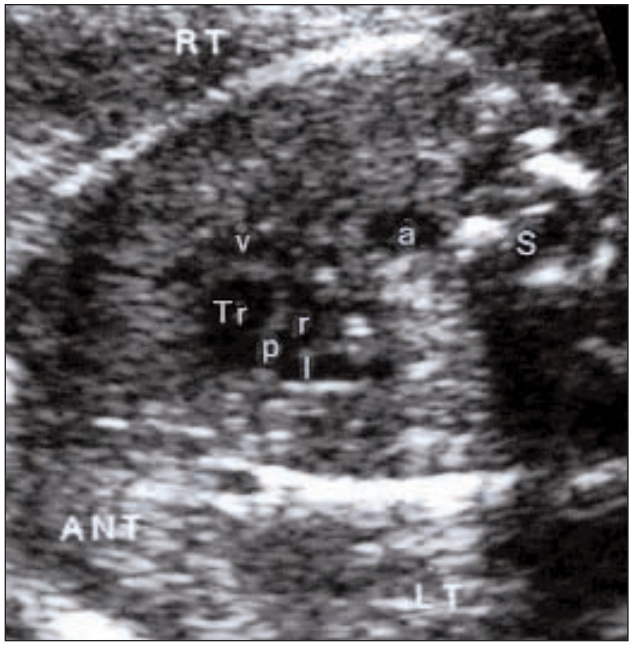

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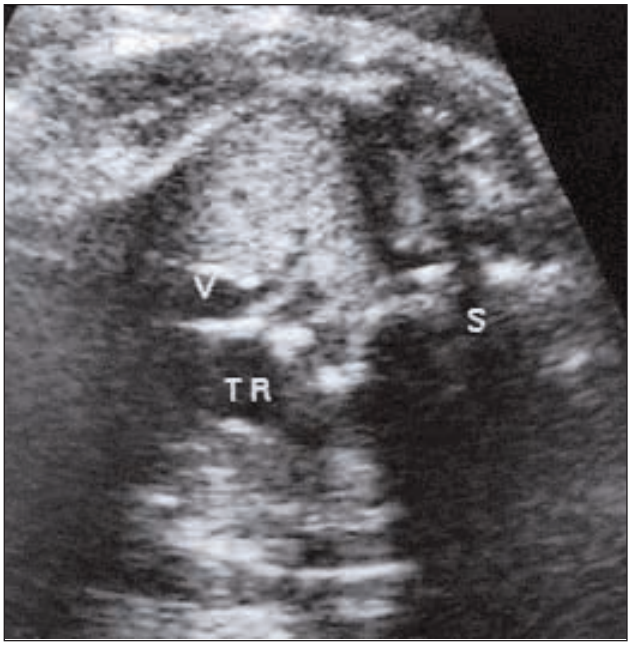
Figure 19.27
Truncus arteriosus. (**a**) Ventricular outflow tract view shows the ventricular septal defect (D) and a single arterial trunk (Tr) arising mostly from the left ventricle (LV). It more commonly arises from both ventricles. (**b**) Three-vessel view shows that the truncus gives rise to the pulmonary artery (p) that bifurcates into the right (r) and left (l) pulmonary arteries. The descending aorta (a) is seen at the right anterior aspect of the spine (S) in this case with a right aortic arch. ANT, anterior; LT, left; RT, right; RV, right ventricle; V, superior vena cava.



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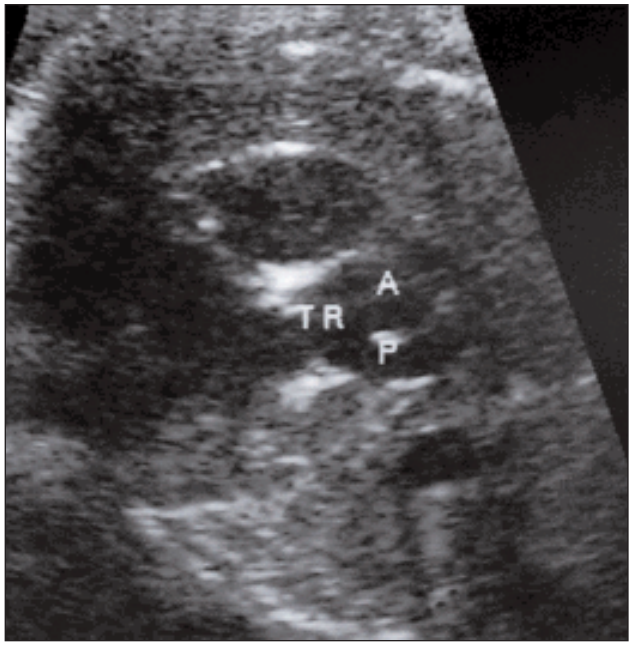

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Figure 19.28
Truncus arteriosus. (**a**) Three-vessel view shows only two vessels: the truncus (Tr) and the superior vena cava (V). (**b**) Long-axis view of the truncus shows the bifurcation of the truncus into the aorta (A) and pulmonary artery (P). S, spine. ((a), reprinted with permission from reference 20.)

arterial valve to give rise directly to the systemic, coronary, and one or both pulmonary arteries (Figure 19.26).[48]

In almost all cases, there is a large ventricular septal defect immediately underneath the common arterial valve, which usually over-rides the ventricular septum.[16,19,21,49,50] Occasionally, the common arterial trunk arises exclusively from the right or left ventricle. The ventricular septal defect can be best demonstrated in the left ventricular outflow tract view, as in tetralogy of Fallot (Figure 19.27). The truncal valve is almost always in fibrous continuity with the mitral valve. The truncal valve is often regurgitant and rarely stenotic.[50] As there is a single arterial trunk, only two vessels are seen at the three-vessel view (Figure 19.28a).[7,19–21] Two vessels at the three-vessel view can also be seen in pulmonary atresia with a ventricular septal defect and absent or hypoplastic main pulmonary arterial trunk, and in aortic atresia with a hypoplastic ascending aorta. The pulmonary arteries arise from the common arterial trunk with or without a short segment of main pulmonary artery (Figures 19.27b and 19.28b).

As in tetralogy of Fallot, leftward deviation of the cardiac axis is common in truncus arteriosus.[24–26] Otherwise, the four-chamber view usually does not show any defect. The ductus arteriosus is absent in approximately half of the cases.[51,52] A right aortic arch is also common, occurring in approximately one-third of cases (Figure 19.27b). Truncus arteriosus may be associated with interruption of the aortic arch.[48,50] Unilateral absence of one pulmonary artery is also common.

Association with chromosomal and extracardiac anomalies

A variety of karyotypic anomalies have been reported to occur with conotruncal anomalies, although they occur most commonly with trisomies 21, 18 and 13.[5,6,53] Chromosomal anomalies are detected more frequently in fetuses than in infants with a conotruncal anomaly (Table 19.2).[5,6,54] They are common in fetuses with tetralogy of Fallot, double-outlet right ventricle or truncus arteriosus, the incidence ranging from 15 to 30% in fetal series. They are rarely, if ever, found in those with complete or corrected transposition.

It has been well documented that a deletion in chromosomal region 22q11 is common in fetuses and patients with tetralogy of Fallot, truncus arteriosus, interrupted aortic arch, or aortic arch anomaly.[55–63] When it occurs with tetralogy of Fallot, the incidence is higher in those with pulmonary atresia, aortic arch anomaly, or absent pulmonary valve (Figure 19.29).[61,62] It is less frequent in those with double-outlet right ventricle and rare in those with transposition. This specific chromosomal abnormality usually occurs in a syndromic pattern, such as DiGeorge syndrome, velo-cardio-facial syndrome, conotruncal face syndrome and Cayler cardiofacial syndrome. As the patients of these groups share clinical and laboratory features, an acronym CATCH-22 (**c**ardiac defect, **a**bnormal facies, **t**hymic hypoplasia, **c**left palate, **h**ypocalcemia, and deletion in chromosome **22**) has been introduced.[57] It has been recommended that chromosomal study with fluorescent in situ hybridization (FISH) for deletion 22q11 should be performed in every fetus with a conotruncal anomaly. Although it occurs de novo in most cases, parental screening is necessary to identify familial cases.[55, 62, 63]

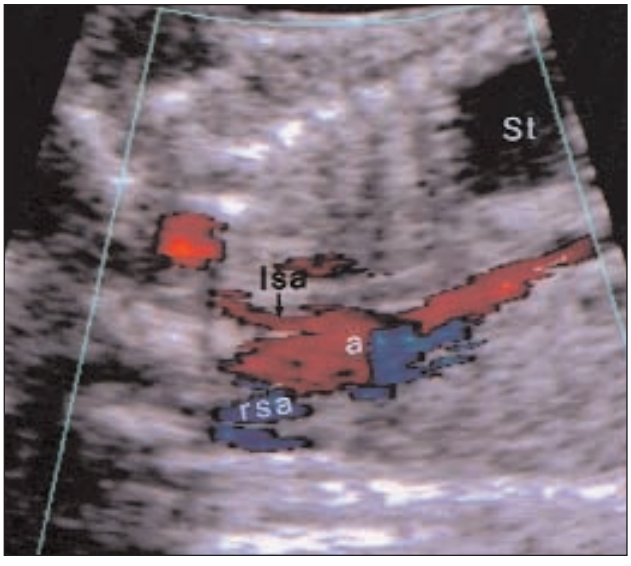
Non-chromosomal extracardiac anomalies are common in infants and fetuses with tetralogy of Fallot, double-outlet right ventricle and truncus arteriosus.[6] They are rare in those with transpositions. They are detected more frequently in fetuses than in infants with a conotruncal anomaly (Table 19.3).

Outcome and postnatal course

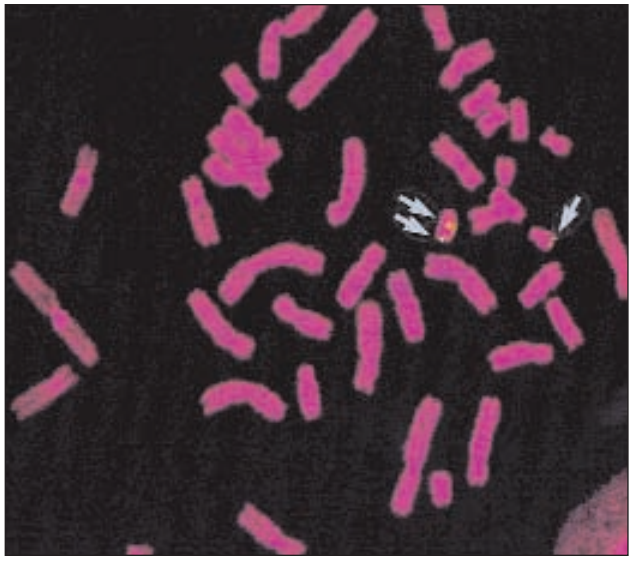
When the diagnosis of conotruncal anomaly is made, approximately 30–50% of the parents choose termination of pregnancy (Table 19.4).[5–7] The leading reason for termination is the association with chromosomal or extracardiac anomalies. A significant number of fetuses die in utero or in the neonatal period.

Table 19.2. Association of conotruncal anomalies with chromosomal anomalies

Diagnosis	*Infant series*[54]	*Fetal series A*[5]	*Fetal series B*[6]	*Fetal series A and B compiled*
Tetralogy of Fallot	33/297 (11.1%)	11/46 (24%)	5/25 (20%)	16/71 (23%)
Complete transposition	1/208 (0.5%)	0/20 (0%)	0/18 (0%)	0/38 (0%)
Corrected transposition	No data	0/3 (0%)	0/5 (0%)	0/8 (0%)
Double-outlet right ventricle	10/86 (11.6%)	4/33 (12%)	5/13 (38%)	9/46 (20%)
Truncus arteriosus	2/51 (3.9%)	2/14 (14%)	1/6 (17%)	3/20 (15%)



a



b

Figure 19.29
Deletion in chromosome 22 in a fetus with tetralogy of Fallot, right aortic arch and aberrant left subclavian artery. (**a**) Coronal color Doppler sonogram shows that the left subclavian artery (lsa) arises aberrantly from the descending aorta (a) which is seen on the opposite side of the stomach (St). The right subclavian artery (rsa) arises from the distal arotic arch. (**b**) Fluorescence in situ hybridization (FISH) to metaphase chromosomes from cultured amniocytes by using D22S75 DiGeorge chromosome region (DGCR) probe and D22S39 control probe. Two hybridization signals (arrows) are seen on the non-deleted chromosome 22. Only one hybridization signal for the control probe (arrow) is seen on the chromosome 22 with a deletion.

Table 19.3. Association (%) of conotruncal anomalies and extracardiac anomalies

Diagnosis	*Infant series*[2]	*Infant series*[4]	*Fetal series*[6]
Tetralogy of Fallot	31	28	48
Complete transposition	9	9	33
Corrected transposition	6	9	0
Double-outlet right ventricle	20	16	46
Truncus arteriosus	48	21	17

Table 19.4. Outcome of cases with fetal diagnosis of conotruncal anomalies (%)

Diagnosis	*Termination of pregnancy*	*Intrauterine death*	*Neonatal death*	*Survivors of continuing pregnancy*
Tetralogy of Fallot (n = 71)	41	7	23	36
Complete transposition (n = 38)	34	8	18	60
Corrected transposition (n = 8)	38	0	38	40
Double-outlet right ventricle (n = 46)	70	11	4	50
Truncus arteriosus (n = 20)	50	0	30	10
Total (n = 183)	48	7	19	42

Data compiled from references 5 and 6

Prenatal diagnosis of a conotruncal anomaly, however, is important because the postnatal prognosis may be improved with urgent initiation of medical or surgical intervention. Bonnet et al. emphasized the importance of fetal diagnosis of complete transposition.[40] In their study, there was no prenatal and postnatal mortality in the cases with prenatal diagnosis of complete transposition, while the prenatal and postnatal mortality rates were 15 and 20%, respectively, in their cases without prenatal diagnosis. They recommended that the fetuses with complete transposition should be delivered in a cardiac center, because a significant number of them would require a balloon atrial septostomy and prostaglandin infusion

Table 19.5. Type, timing, success rate and long-term outcome of definitive surgery in various conditions

	Preparatory procedure	*Definitive surgery*	*Timing*	*Success rate (%)*	*Long-term survival*
Tetralogy of Fallot[64–69]	Nil in most cases, BT shunt or balloon dilatation of pulmonary valve in some	Complete repair	In first 6 months	>95	>90%, 10-year
Tetralogy with pulmonary atresia[71, 72]	Nil when PDA is the only supply	Complete repair	Newborn		
	Unifocalization when there are MAPCAs	Complete repair	2–3 months	75	70%, 10-year
	BT shunt when pulmonary arterial bed is hypoplastic	Complete repair	Variable		
	Nil when there are small MAPCAs	Nil			
Complete transposition, simple[72–77]	Balloon atrial septostomy in most cases	Arterial switch	Newborn	98	>90%, 10-year
Complete transposition, with VSD and PS[72–77]	BT shunt when PS is severe	Repair with conduit or REV	1.5–2 years	93	>90%, 10-year
Corrected transposition, simple[78]	Nil	Nil in most	—	—	88%, 20-year
Corrected transposition, with VSD or with VSD and PS[78,79]	Pulmonary artery banding or BT shunt	VSD closure	In first few months		
		VSD closure with conduit	1.5–2 years	94	65%, 10-year
Double-outlet right ventricle, simple[80–84]	Pulmonary artery banding in some	VSD closure	Before 6 months	90	80%, 10-year
Double-outlet right ventricle, complex, without PS ± subaortic stenosis[80–84]	Pulmonary artery banding in some	VSD closure ± VSD enlargement, ± subaortic resection	Before 6 months		
Double-outlet right ventricle, complex, with PS or pulmonary atresia[80–84]	BT shunt in some	VSD losure ± arterial switch	Before 6 months	79	80%, 10-year
		VSD closure + conduit or REV	1.5–2 years		
Truncus arteriosus[85]	Nil	Repair with conduit	In first 2 months	>90	83%, 15-year

BT, Blalock–Taussig; MAPCA, major aortopulmonary collateral artery; PDA, patent ductus arteriosus; PS, pulmonary stenosis; REV, Réparation à l'Étage Ventriculaire procedure; RVOT, right ventricular outflow tract; VSD, ventricular septal defect

immediately after birth. The patency of the ductus should also be maintained with prostaglandin infusion when the conotruncal anomaly is associated with severe pulmonary stenosis, the pulmonary blood flow therefore being 'ductus-dependent'.[30,31] The prenatal recognition of absent ductus arteriosus in truncus arteriosus or absent pulmonary valve syndrome is also important because these lesions will not respond to prostaglandins. When the pulmonary arterial size is too small and the ductal patency can not be maintained, an urgent Blalock–Taussig shunt operation is mandatory. Table 19.5 summarizes the type, timing, success rate, and long-term outcome of the definitive surgery in various circumstances.[64–85]

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