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The spectrum of fetal cardiac malformations

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Abstract Increasingly, paediatric cardiologists are called upon to diagnose cardiac malformations prenatally. In the main, the types of malformation seen during fetal life will be similar to those documented postnatally, but the frequency with which they are encountered, as well as the views that can be used for diagnosis, will be different. This review aims to describe the anatomic spectrum of malformations seen in 917 fetal hearts examined consecutively following prenatal diagnosis. The distribution of anomalies is illustrated in terms of a simple sweep through the fetal thorax passing from the four-chamber plane to the outflow tracts, and then to more cranial views of the mediastinum. Two-thirds of the anomalies described would have been detectable in the four-chamber plane. Some, such as tricuspid valvar abnormalities, will alter the normal appearances of the four-chambers dramatically. In terms of the overall spectrum, however, such obvious abnormalities only form a minor part of the total number. Others, such as atrioventricular septal defect, will often require closer inspection of intracardiac anatomy, but will make up a large proportion of the entire cohort. Up to one third of the anomalies in the series would have required views more cranial to the four-chamber plane of section. In these, it would have been necessary to examine the nature of the left ventricular outflow tract, the crossing of the two outflows, or else the arterial arches in order to secure detection. In the fetus, these and other planes must be considered by the echocardiographer in order completely to detect and document the entire spectrum of cardiac abnormalities likely to be encountered.

Keywords: Congenital cardiac malformations; fetal cardiology; cardiac morphology

OVER THE LAST DECADE, DIAGNOSIS OF congenital cardiac malformations during fetal life has become an important part of many specialties. Experts in fetal medicine are required to examine the fetal heart, as they are with any other organ or system in the fetus. Referral to specialist fetal cardiology or medicine centres is often made by sonographers or radiologists whom, in an expanding number of countries, are required to screen the fetal heart for likely malformations as part of the their assessment of fetal anomalies in low risk populations. Consequently, paediatric cardiologists are increasingly called upon to diagnose, refine, or prognosticate on cardiac anomalies in the fetus.

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The purpose of this review is not to describe the spectrum that can be encountered during fetal life, since this has been described in detail previously[1]. Instead, it is my aim to review the distribution and morphology of the various malformations in terms of the prenatal diagnostic views that might be most useful for the pediatric cardiologist aiming to recognise the malformations. In essence, I aim to describe the morphology of the heart in the eyes of the fetal echocardiographer as well as the anatomist.

For the framework of my review, I have used the anatomy of fetal hearts examined at Guy's Hospital, London during the ten-year period from 1989 to 1999. All of the 917 cases were obtained consecutively for correlation with prenatal ultrasonic findings as part of clinical audit. Since this is an anatomic study, only structural abnormalities are included, but this is not to imply that anomalies of rhythm, such as supra-ventricular tachycardia,

or complete heart block, are not of considerable importance in fetal echocardiography.[2,3] When ranked in terms of a single primary lesion, this collection of cases reveals a spectrum of abnormalities very similar in order of frequency to that detected clinically during fetal life.[4] Thus, lesions such as the hypoplastic left heart syndrome are those most frequently encountered, while others such as complete transposition are less common (Fig. 1). At the far end of the spectrum, miscellaneous conditions such as aorto-left ventricular tunnel, or coronary arterial-ventricular communications, are to be found but are encountered extremely rarely.[5]

While useful for comparison with other series, such a method of description tells us little about how the various malformations are likely to have been suspected or recognised echocardiographically, either at a screening level or in a tertiary setting. For the remainder of this article, then, I have re-categorised these various anomalies, and interpreted them in terms of four anatomic planes of the fetus. These planes of section, within the transverse planes of the fetal body, are analogous to a theoretical 'sweep' through the fetal thorax using ultrasound, moving cranially from the diaphragm towards the fetal head. In consequence, they are slightly different from the views used by the paediatric cardiologist following birth. The most caudal section encountered using this 'sweep' of the fetal thorax will be the four-chamber plane of the heart. This view is well-known to paediatric echocardiographers and fetal sonographers alike, but it is worth taking a moment to understand why this view, and the others more superior to it, are so readily obtainable in the fetus. This information is also helpful before I explain how abnormalities alter the appearance of the normal views.

The four chamber fetal section

In the normal fetus beyond 11 weeks gestation, the apex of the heart has turned leftwards so that, when viewed from above, it lies obliquely across the chest, adopting an angle of 45 degrees to both the sagittal and coronal planes of the body (Fig. 2). Viewed from the front of the fetus, the inferior surface of the heart is virtually parallel to transverse plane of the fetus, since the apex of the heart is lifted up towards the fetal head by the large fetal liver. The result of this arrangement is that the four-chamber plane of the heart is very similar to the transverse section of the fetus (Figs 3 and 4). As in postnatal life, the right atrium and ventricle occupy the most anterior position within the chest, while the left atrium and ventricle are more posterior (Fig. 5). Contained within this section are numerous clues to cardiac normality, some of which will be described in more detail throughout this article. More importantly, in the fetus, it is possible to move in planes parallel to the four chamber plane. As the aortic valve and left ventricular

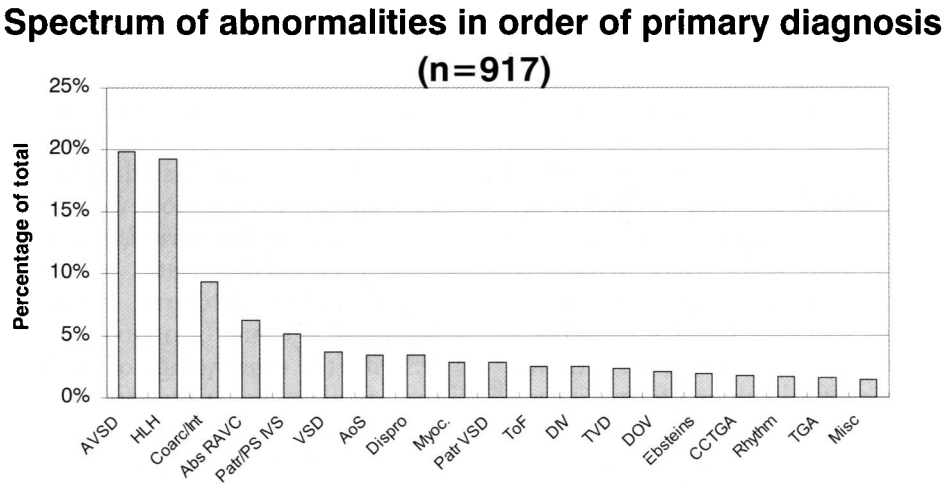

Figure 1.
The spectrum of malformations seen in 917 fetal specimens and ordered by primary diagnosis. Abbreviations as follows: AVSD=atrioventricular septal defect; HLH=Hypoplastic left heart syndrome: Coarc/Int=Coarctation or interruption of the aortic arch; Abs RAVC= Absent right atrioventricular connection; Patr/PS IVS= Pulmonary atresia or pulmonary stenosis with intact ventricular septum; VSD = ventricular septal defect; AoS=Aortic stenosis; Dispro=Disproportion; Myoc.=Myocardial anomalies; Patr VSD=Pulmonary atresia with ventricular septal defect; ToF= Tetralogy of Fallot; DIV= Double inlet ventricle; CCTGA= Congenitally corrected transposition: Rhythm=Rhythm anomalies such as supraventricular tachycardia or complete heart block: TGA= complete transposition: Misc=Miscellaneous anomalies.

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outflow tract are centrally positioned within the heart, these are the first structures that will be encountered as one moves superiorly (Figs 3, 4). Moving still further superiorly will demonstrate the right ventricular outflow tract, since this is lifted superiorly away from the tricuspid valve by the complete sub-pulmonary infundibulum (Figs 3 and 4).

Size of the heart

Perhaps the most obvious sign of a cardiac anomaly in the fetus will be a gross change in the form of the heart that affects its overall size relative to the thorax. It is well known that, in the normal fetus, the four chambers of the heart occupy approximately one third of the area of the fetal thorax.[6] When cardiomegaly is associated with structural heart disease, the increase in cardiac size is often evident from the four-chamber plane, the heart stretching from one side of the thorax to the other in severe cases. Consequently, to varying degrees, the lungs are squeezed to the back of the thorax in the four-chamber plane (Fig. 6). In the present series of fetal hearts, this form of overall enlargement was most commonly associated with enlargement of the right-side of the heart as a result of tricuspid valvar abnormalities (Fig. 7). As a result, the right atrium and ventricle filled the anterior thorax, and the axis of the heart was rotated posteriorly (Fig. 6). Tricuspid valvar abnormalities associated with such changes range from dysplasia of the leaflets to Ebstein's malformation, and rarely complete agenesis of tricuspid valvar tissue (Fig. 7). In all forms, incompetence of the tricuspid valve is accentuated by dilation of the

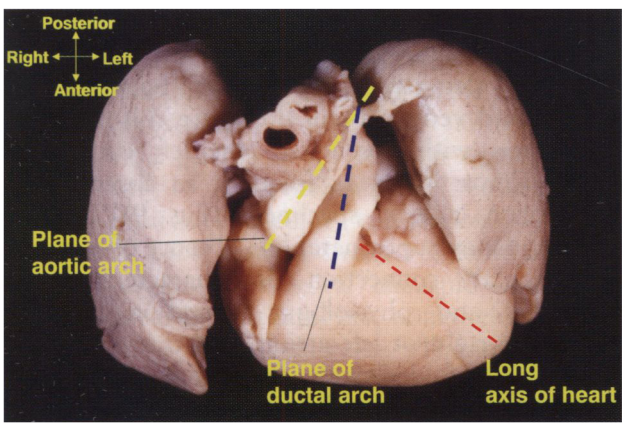

Figure 2.
This normal fetal heart at 22 weeks' gestation is viewed from above to show the oblique planes of the ventricular septum, ductal and aortic arches to the axis of the body.

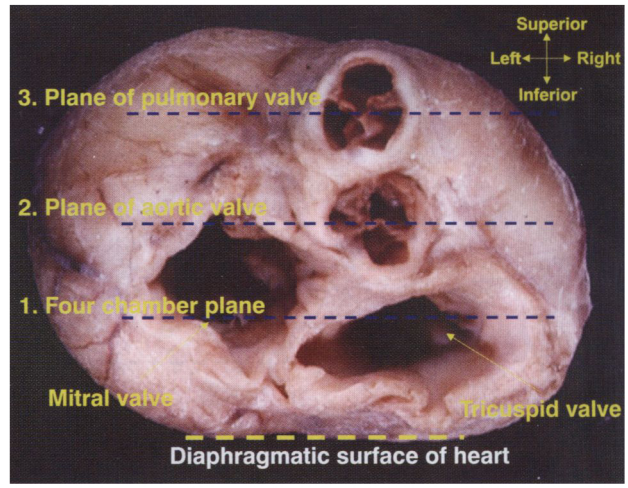

Figure 3.
The relative position of the cardiac valves to one another can be appreciated when looking at the mid-trimester fetal heart from its base. Planes of section have been superimposed to illustrate how each valve can be imaged in parallel transverse sections.

right atrioventricular junction, which has the effect of further preventing coaption of the already dysplastic leaflets.[7] All these entities can be associated with stenosis or atresia of the pulmonary valve, but are rarely associated with infundibular atresia.[8] A much smaller proportion of hearts with cardiomegaly will result from enlargement of the left-side of the heart due to mitral valvar regurgitation and dilation of the left atrium (Fig. 7). In this small group, left atrial and ventricular enlargement are associated with aortic valvar stenosis, and with premature closure or severe restriction of the atrial septum. All these conditions, in their severe forms, are obvious abnormalities in the four-chamber view. Their poor outcome following birth is well recognised, and is primarily due in those with abnormalities of the tricuspid valve to respiratory insufficiency, although the lungs show normal maturation.[9] In those with aortic stenosis or atresia and premature closure of the ventricular septum, there is commonly associated lymphangectasia of the lungs. This feature, along with accompanying arterialisation of the pulmonary veins, has been shown to affect the outcome of palliative surgery.[10] Consequently, one might expect these abnormalities to be much more frequently detected during fetal life than in neonates, and indeed they appear to be more prevalent.[1] Their influence on the spectrum of disease seen prenatally needs to be put into perspective, however, since these conditions are still relatively rare in fetal life, accounting for only 5% of the total series (Fig. 8)

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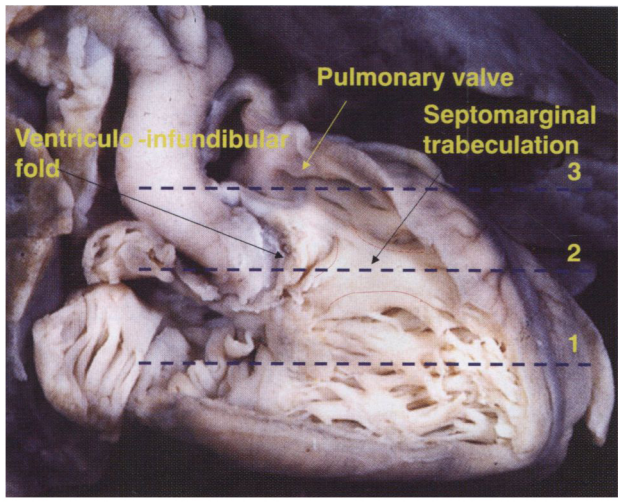

Figure 4.
When viewed from the front, the near horizontal nature of the normal fetal heart can be appreciated. In this specimen, the free wall of the right ventricle has been removed so as to demonstrate the relationship of the outflow tracts and the position of the septomarginal trabeculation which cradles the ventriculo-infundibular fold. The planes of section are numbered as in figure 3.

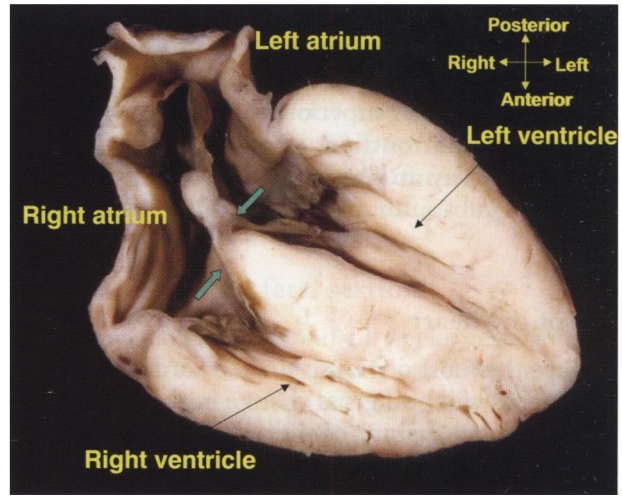

Figure 5.
This view of an 18 week fetal heart shows the normal four-chamber view. The cardiac axis is at a 45 degree angle to the planes of the body and the two sides are of similar size. The connection between the atria and ventricles can be seen as can the differential insertion of the two atrioventricular valves (arrows)

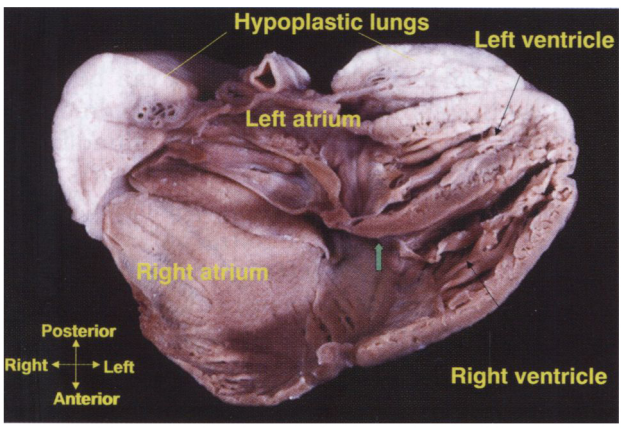

Figure 6.
Four-chamber plane of section in a fetus with tricuspid valvar dysplasia. There is marked dilation of the right heart which fills the anterior part of the chest, rotates the axis of the heart posteriorly and compresses the lungs. The hingepoint of the tricuspid valve at the ventricular septum is normal (arrow) in this instance but can be displaced in similar fetuses with Ebstein's malformation.

Atrioventricular connections

In order to account for the detection of more anomalies from this series, it is necessary to focus closely on the morphologies demonstrated from the four-chamber view. Since the two atriums and ventricles are visible in transverse sections of the fetal thorax, so are the junctions between them, as well as the tricuspid and mitral valves that guard these junctions. As in the neonate, in the normal fetal heart the atrioventricular valves are positioned in the long axis of the fetal body, at right angles to axis of the heart, and therefore at an angle of 45 degrees to both sagittal and coronal planes (Fig. 5). A section of the fetal heart across its base demonstrates the diaphragmatic position of the tricuspid valve, this being more anterior than the mitral valve (Fig. 3). Consequently, the next most obvious forms of abnormality will be those showing univentricular atrioventricular connections. In total, these abnormalities will account for much larger proportion of the overall spectrum of abnormalities within the pathological series (Fig. 8). Complete absence of the either the anterior (right-sided) or posterior (left-sided) connection between the atrium and ventricle will dominate this group of anomalies, with absence of the left connection being almost twice as frequent as absent right connection in fetal specimens (Fig. 9). In fetal hearts, just as in postnatal life, absence of one atrioventricular connection is characterized by anatomic and physiologic separation between either the right or left atrium and the ventricular mass by an infolded sandwich of tissue composed of atrial myocardium, fibrous tissue that later becomes infiltrated with fat, and ventricular myocardium. Consequently, atrial myocardium joins the base of the atrial septum, thereby walling off one atrium from the rest of the heart. Since the ventricle beneath the absent connection is usually

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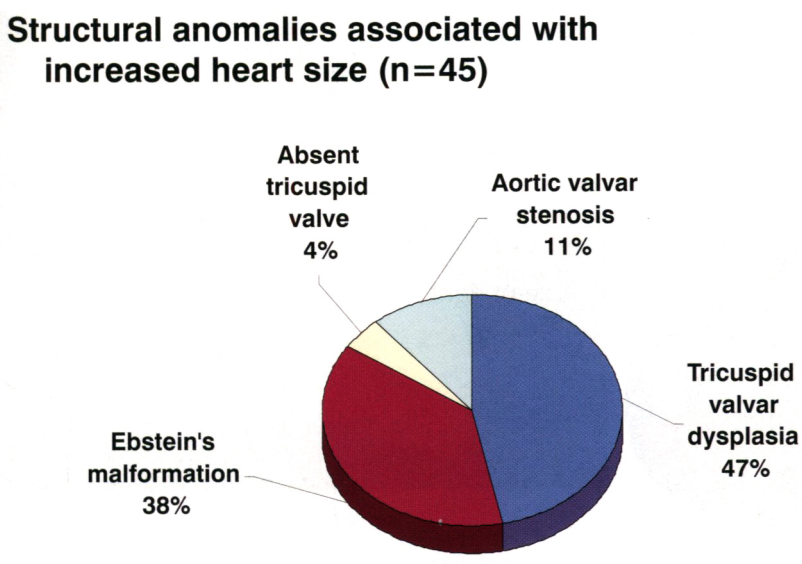

Figure 7.
The frequency of anomalies associated with increased cardiac size in the pathological series.

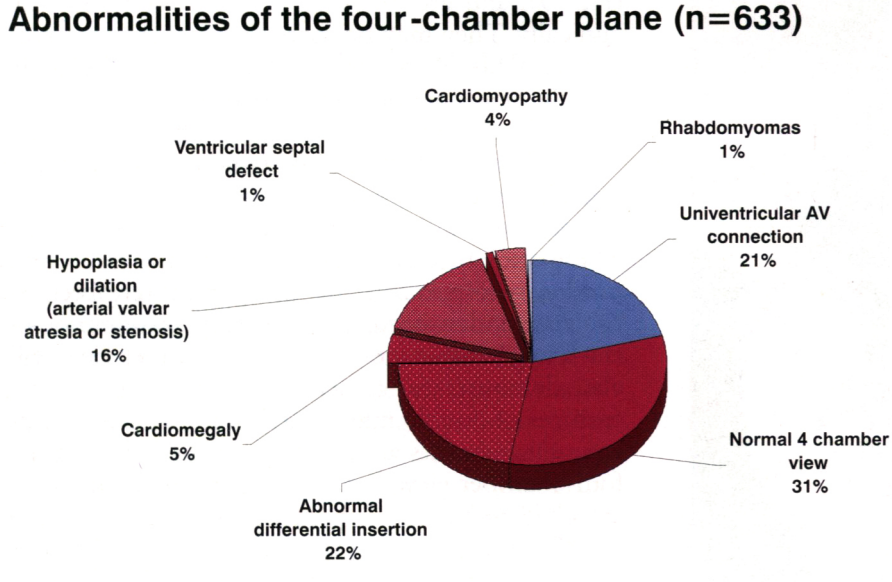

Figure 8.
The distribution of anomalies detectable in four chamber sections in the pathological series.

hypoplastic, there will also be alteration in ventricular proportions. This is readily seen in the four chamber plane, as is a shifting of the axis of the heart either towards the midline (absent right connection, Fig. 10) or to the left (absent left connection). In the majority of fetuses, the remaining atrioventricular connection will be concordant. Most fetuses, therefore, could also be classified as having classical tricuspid or mitral atresia. Just as in postnatal life, nonetheless, other more complex variants do exist.[11] The importance of knowing the nature of the persisting atrioventricular connection is that the nature of the ventriculo-arterial connection, as seen in views outside the four-chamber plane, will depend upon it. In the majority of fetuses with absence of the right atrioventricular connection due to tricuspid atresia, the two great arteries are concordantly connected to their respective ventricles, while in those with more complex forms of absent right connection, the ventriculo-arterial connections are likely to be abnormal. Likewise, in fetuses with absence of the left atrioventricular connection due to mitral atresia, the commonest ventriculo-arterial connection is aortic atresia, with the pulmonary trunk arising from the right ventricle, or else double outlet right ventricle. In more complex forms of absent left connection, however, the ventriculo-arterial connections will most likely be discordant. This discussion of arterial complexity is a digression from the abnormalities visible in the four-chamber plane, but it does serve to illustrate why it is vital, at least in the tertiary setting, accurately to define the morphology of the chambers and connections under examination even during fetal life.

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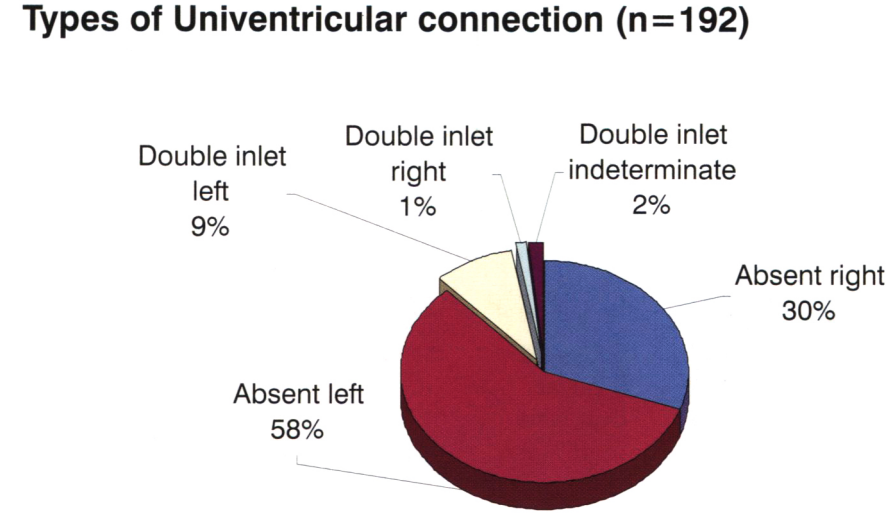

Figure 9.
The types of univentricular atrioventricular connections found in 192 fetal specimens from the series.

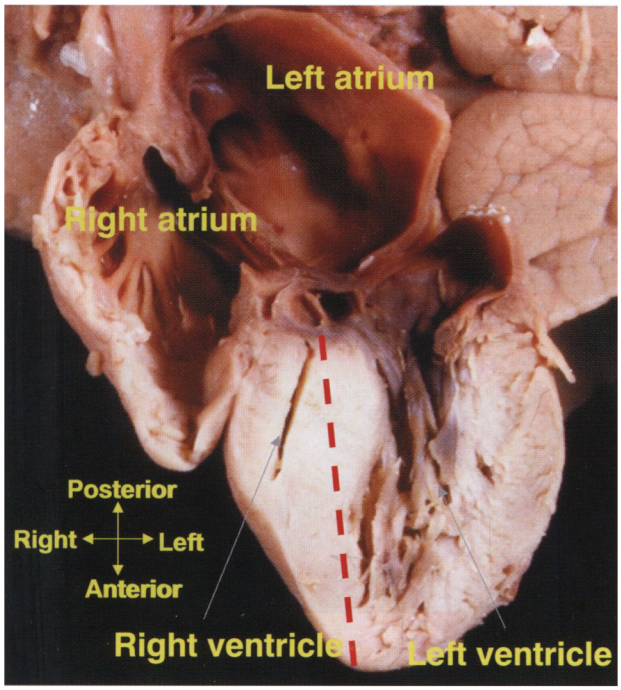

Figure 10.
A four chamber section of a fetal heart with absence of the right atrioventricular connection due to tricuspid atresia. There is separation of the right atrium from the a hypoplastic right ventricle by a fibromuscular sandwich (arrow). The axis of the heart is deviated towards the midline (dashed line)

This then, is the simple end of the spectrum of hearts with univentricular atrioventricular connections, all of which will generally require palliation to create a functionally univentricular circulation following birth rather than complete repair. Equally important to detect in terms of prognosis will be those hearts at the more complex end of the spectrum, namely those with double inlet ventricle. These hearts form a smaller proportion of hearts with univentricular atrioventricular connections (Fig. 9), and are likely to be seen less frequently clinically. They may be more problematic to detect during fetal life since, in this setting, there are usually not one but two atrioventricular valves visible in the four chamber plane (Figs 11 & 12). Closer inspection will also demonstrate papillary muscles groups running in between and supporting the anterior and posterior atrioventricular valves. These are particularly prominent in the fetus, and produce the potential for being misinterpreted as the ventricular septum on a cursory examination of the four-chamber view. The four-chamber view will also demonstrate, nonetheless, the characteristic features of double inlet ventricle familiar to paediatric cardiologists after birth[12]. In the centre of the four-chamber view, both atrioventricular valves are attached to the base of the atrial septum, and are attached at the same level (Figs 11 & 12). The ventricular septum, if present, is deviated either antero-superiorly as in double inlet left ventricle, (Fig. 11), or postero-inferiorly as seen in double inlet right ventricle, (Fig. 12). The essence of double inlet is that both atrioventricular valves are malaligned relative to the rudimentary ventricular septum. The majority of each valve leads into either a dominant left ventricle, which is located posteriorly, or a dominant right ventricle, seen anteriorly. Only extremely rarely will both atrioventricular valves lead into a solitary ventricle that is of indeterminate morphology (Fig. 9).[6]

Overall, hearts with cardiomegaly or univentricular atrioventricular connections accounted for the detection of approximately one quarter of the total number of hearts seen at Guy's Hospital. This still leaves three-quarters unaccounted for. For these, it will be necessary to take note of other features present in the four-chamber view.

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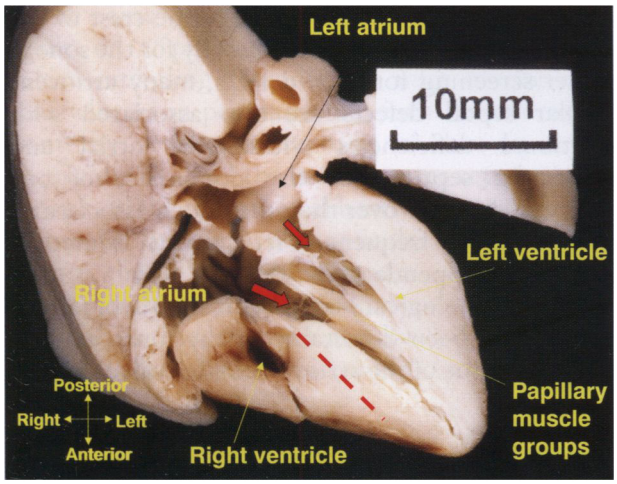

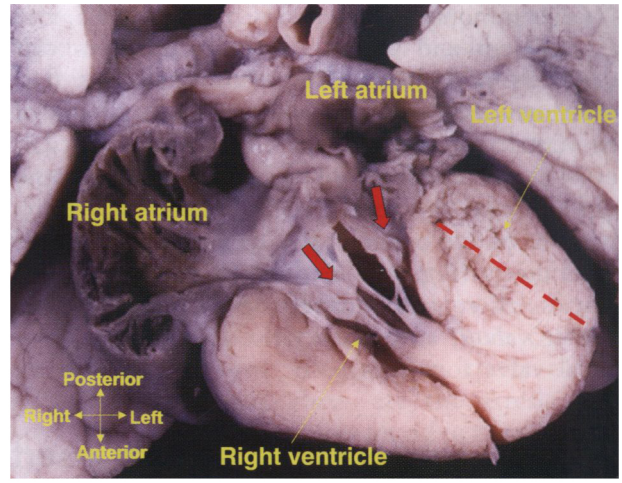

Figures 11 and 12.
These sections, taken in the 'four-chamber' plane are from mid-trimester fetuses with double inlet left (Fig 11) and double inlet right (Fig 12) ventricle. Two atrioventricular valves are evident in this plane (arrows) and are malaligned with respect to the ventricular septum (dashed line)

Differential insertion of the hinges of the atrioventricular valves

Documentation of some of these features will require a closer inspection on the part of the echocardiographer. When abnormal, nonetheless, these features permit detection of the larger proportion of cases seen in the pathological series. Of particular note is the 'crux' of the heart at the centre of the four-chamber plane of section. Such a synonym, in reality, hides the vital aspect of anatomy in this region of the heart, since 'crosses' are not usually thought of as being asymmetric. For the prenatal sonographer, as for the paediatric cardiologist, it is the very asymmetry of the 'crux' of the heart that is crucial to the diagnosis of normality. The offsetting between the atrioventricular valves is apparent from early in gestation, in fact as soon as the atrioventricular junction has been divided into right and left halves. It increases in magnitude with age from approximately 1mm at 18 weeks (Fig. 5) to 3.5mm at 40 weeks gestation. On the anterior, and morphologically right-side of the normal ventricular septum, the septal leaflet of the tricuspid valve inserts into the ventricular septum towards the apex of the heart, whereas on the left side, the arterial, or anterior leaflet of the mitral valve is lifted away from the ventricular septum and hinges more towards its base (Fig. 5). In my overall series, abnormalities of differential insertion accounted for a further quarter of the total (Fig. 8). Accentuation of the degree of offsetting will already have been a feature in fetuses with increased cardiac size and Ebstein's malformation. This accounts for a further 3% of the total of 917 fetuses. Reversal of the normal pattern is seen in congenitally corrected transposition due to discordant atrioventricular connections. This is another potential reason for recognition of fetal cardiac abnormalities, but in reality accounts for only a small fraction of cases (Figs 13,14).

Far more frequently encountered will be those fetuses in which there is loss of the normal offsetting of the atrioventricular valves (Fig. 13), either as a result of atrioventricular, or perimembranous ventricular, septal defects. As in post-natal life, it is the offsetting between the two atrioventricular valves that gives rise to the small region between the two sides of the heart in the fetus that neither separates right atrium from left atrium nor right ventricle from left ventricle. Instead, this area, separates right atrium from the left ventricle, and is therefore 'atrioventricular'. For many years, this portion of the heart was defined as a septal structure. Recent work has proven that it is composed of a fibro-muscular wedge that pushes into the diaphragmatic surface of the heart, separating the normal atrioventricular junctions into right and left halves[13]. Atrioventricular 'septal' defects, in contrast, occur when the junction is not divided into right and left halves, but rather remains a common structure. In fact, it is the common junction which is their pathognomonic feature. Be that as it may, atrioventricular septal defects, with common atrioventricular junction, in all their various guises, are encountered particularly frequently in fetal life.[14,15] They account for nearly one fifth of the total series of 917 hearts, and yet in their simplest form produce only relatively minor changes to the four-chamber views of the heart.

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Thus, commonly in fetuses with Trisomy 21, the arrangement and size of the four chambers will appear normal. The atrial septum, and its flap valve, may also be present, as can be the majority of the ventricular septum, but closer inspection will reveal than both the atrial and ventricular septums stop short of the centre of the heart in a four-chamber plane, producing a true atrioventricular septal defect (Fig. 15). Because of the septal deficiencies, there is no offsetting of the atrioventricular valves, and neither the tricuspid nor the mitral valve form normally. Instead, two leaflets common to both ventricles bridge the crest of the ventricular septum (Fig. 15). Luckily for the sonographer screening for such defects, many atrioventricular septal defects are also associated with additional deficiencies within the atrial and ventricular septum, or else dominance of one ventricle chamber over the other. Such associations are particularly frequent in fetuses with isomerism of the atrial appendages.[14]

In my pathological series, a further 3% of fetuses would have been detectable in the four chamber plane by virtue of the atrioventricular valves arising

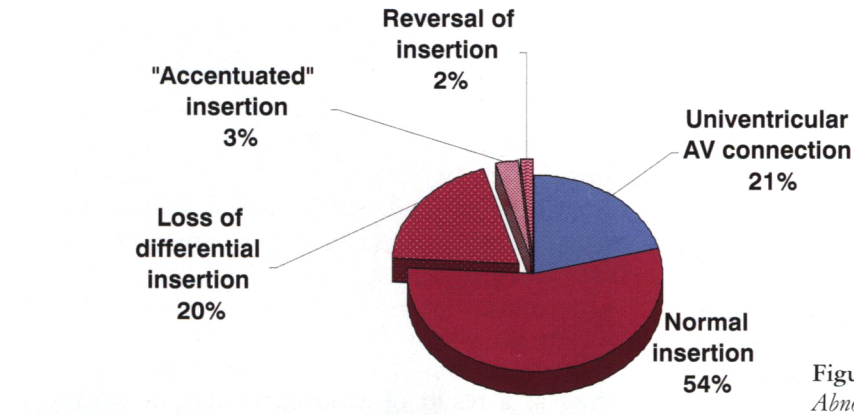

Figure 13.
Abnormalities of differential insertion found in 211 fetuses from the total series.

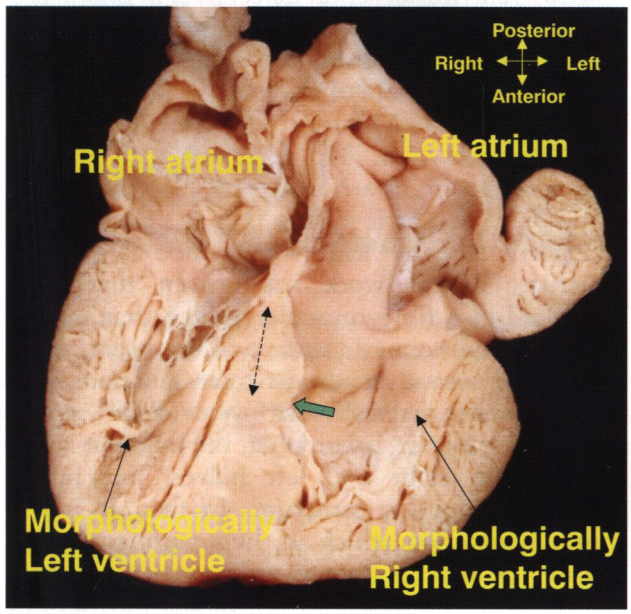

Figure 14.
This four-chamber section shows the anatomy of discordant atrioventricular connections in the fetus. There is reversal of the normal pattern of offsetting of the atrioventricular valves at the crux of the heart. This is accentuated in this particular case by Ebstein's malformation of the left-sided tricuspid valve.

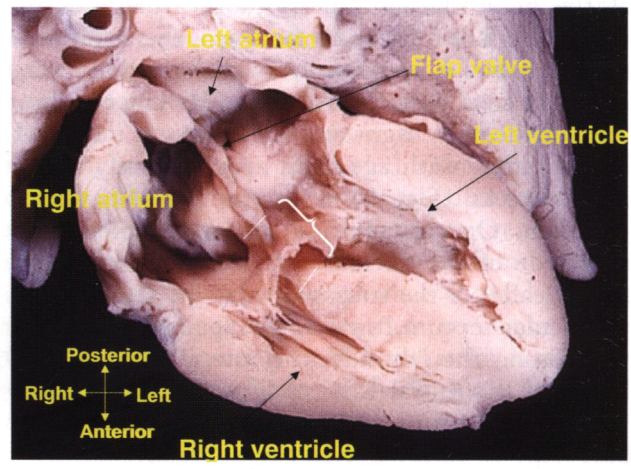

Figure 15.
The anatomy of a small atrioventricular septal defect (bracketed) as seen in a four-chamber section in a fetus with Trisomy 21. The majority of the atrial and ventricular septums are complete but the crux of the heart is deficient.

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at the same level in four chamber sections as a result of a perimembranous ventricular septal defect extending towards the inlet of the right ventricle. As in postnatal life, such defects extend from the ventricular septum to abut upon the tricuspid valve and the membranous septum, even though, in the normal fetal heart, the membranous septum is mainly atrioventricular.[16] As a consequence, the tricuspid valve cannot attach normally to the right side of the muscular ventricular septum. Instead, it arises at a similar level to the aortic leaflet of the mitral valve when seen in four-chamber sections of the heart.

Miscellaneous abnormalities of four-chamber sections

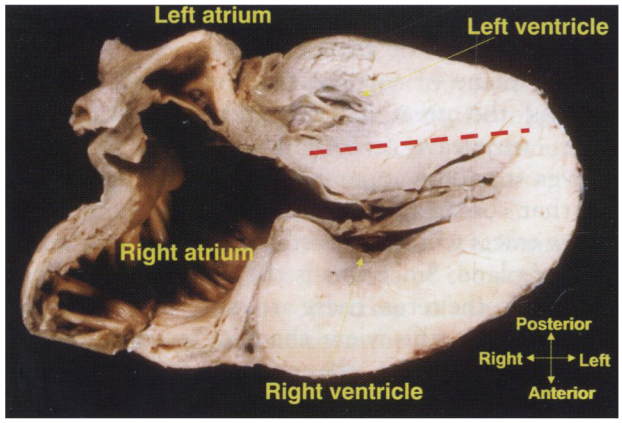
It would be misleading to leave the four-chamber plane without commenting on the variety of miscellaneous lesions that, although infrequent, together account for a small percentage of anomalies detectable in this view (Fig. 8). The four-chamber appearances of these anomalies will vary from fetus to fetus, but would include such minor lesions as small ventricular septal defects within the muscular portion of the septum. In my pathological series, such defects were infrequent, and were usually an incidental finding in fetuses with extra-cardiac abnormalities. This is, of course, in stark contrast to the prevalence seen in clinical practice.[17] Since these defects do not abut on the atrioventricular valves, the echocardiographic crux of the heart will be normal in four-chamber views.[6] The miscellaneous group of four-chamber anomalies will also include diverse anomalies such as cardiomyopathies which, in the pathological series, were seen to affect all four chambers, both ventricles, only one ventricle, or both atriums.[6] They also include certain forms of cardiac tumours.[18] In particular, rhabdomyomas are commonly seen within four-chamber sections of the heart, in keeping with their myocytic origin. These are usually variable in size and number. In contrast, teratomas were always singular, more commonly originating around the arterial roots, and therefore outside the four-chamber plane of section. Nonetheless, the series of 917 cases includes a single fetus with an intracardiac teratoma within the right ventricle that would have been visible in the four-chamber plane.

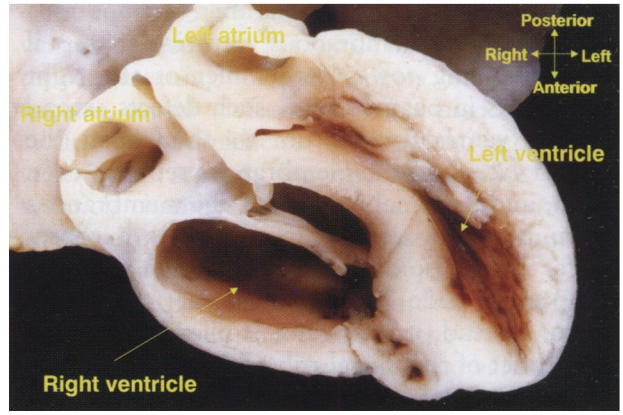
Abnormal chamber proportions reflecting lesions out of the four-chamber view

Thus far then, we have accounted for the detection of approximately half of the total 917 fetal hearts in the pathological series. In order to include still more anomalies, it is necessary to begin to think of abnormalities of the fetal heart outside the four-chamber plane of section, in other words, those involving the great arteries and outflow tracts. Before we leave the four-chamber sections, however, it will be apparent to paediatric cardiologists that some of these anomalies involving the outflow tracts will also be reflected within the four-chamber plane. Since this is the starting point for diagnosis in the fetus, these are more likely first to be detected in this view than in more complex views demonstrating the outflow tracts of the heart. These lesions, then, are the atresias and stenoses of the outflow tracts found in the setting of an intact ventricular septum. Together, these anomalies account for a further one-sixth of the pathological series, with valvar atresia or stenosis on the left side dominating that on the right (Fig. 8). Although these obstructive lesions are downstream of the four chambers of the heart, they nonetheless have marked effects on the four chamber view of the fetus, as they also do in the neonate.[19] As already mentioned, aortic valvar stenosis, when associated with mitral valvar regurgitation and premature closure of the atrial septum, will readily be detectable by an overall increase in cardiac size due to enlargement of the left atrium. But there is a wider variety of changes that can occur in association with atresia or stenosis of the outflow tracts and valves in fetal life. These range from dilation of the ventricle to hypoplasia associated with thickening of its walls (Figs 16 & 17). Such appearances are often particularly noticeable since the ventricular walls will often also be echogenic due to endocardial fibroelastosis and calcification of the affected chamber. All are readily visible in the four-chamber view of the fetal heart.

Views of the outflow tracts

In order to begin to account for the final third of fetal cardiac anomalies, we must concentrate on lesions that are *only* detectable in views outside the four-chamber view of the heart. But first, before continuing the sweep through the fetal thorax, it is useful to be reminded of the position of the outflow tracts to one another in the normal heart. In the normal fetal heart, the two atrioventricular valves are within the diaphragmatic plane of the heart. Moving superiorly towards the fetal head in a parallel section, the first arterial valve to be encountered should be the aortic valve, since this is positioned above the tricuspid and mitral valves in the centre of the heart (Fig. 3). Consequently, sections of the fetal thorax just above and parallel to the four chamber view, or else those obtained by tilting





Figures 16 and 17.
These four-chamber sections show the variable nature of ventricular size in fetuses with arterial valvar stenosis and atresia. In figure 16, the left ventricle is hypoplastic and the axis of the heart is deviated posteriorly (dashed line). The aortic valve was atretic. In figure 17, the pulmonary valve was imperforate and the anterior (morphologically right) ventricle is dilated.

from the four chamber view towards the fetal head, will demonstrate the left ventricular outflow tract leading into the aorta, the latter trunk being directed towards the right shoulder of the fetus (Figs 3, 4 & 18).

In the normal fetal heart, the anterior wall of the aorta will be continuous with the line of the ventricular septum. It is abnormal separation of these two components of the heart than can account for a diverse range of anomalies involving the outflow tracts, and hence set the scene for their likely detection. The conditions detectable in such a view will range from tetralogy of Fallot with and without pulmonary atresia, to common arterial trunk and double outlet right ventricle, as well as rare anomalies such as aortic atresia with sub-pulmonary ventricular septal defect, or so-called 'absent' pulmonary valve syndrome. All these abnormalities commonly share the same singular anomaly, namely a ventricular septal defect cradled within the arms of the septomarginal trabeculation (Fig. 19). Consequently, regardless of whether the posterior rims of the defect are muscular or perimembranous, or indeed whether the defect extends to become juxta-arterial and doubly committed, all will be seen as anomalies of the first outflow tract in this theoretical ultrasound 'sweep' of the fetal thorax (Fig. 20). Of course, in addition, it would be difficult to ignore the hypoplasia of the pulmonary trunk seen in cross section in this view in fetuses with tetralogy of Fallot, just as it would be difficult to ignore the fact of a solitary outlet in pulmonary atresia or common arterial trunk (Fig. 20). Or to ignore dilation of the pulmonary arterial branches in 'absent' pulmonary valve syndrome.[20]

Such abnormalities are likely also to be confirmed in the next plane of the fetal thorax. In this view, which once again is superior and parallel to the left ventricular outflow tract, it is possible to section the pulmonary trunk. Since, in the fetus, this trunk originates close to the anterior chest wall, and passes directly back to join the descending aorta via the arterial duct, it can easily be imaged in the transverse plane. As in the neonate, it also branches transversely, so slight tilting of the plane of section towards the right or left will demonstrate its right and/or left pulmonary branches, and/or the arterial duct (Fig. 21). It is, however, the direction of the arteries at the level of the outflow tracts that is of equal importance for the detection of abnormalities

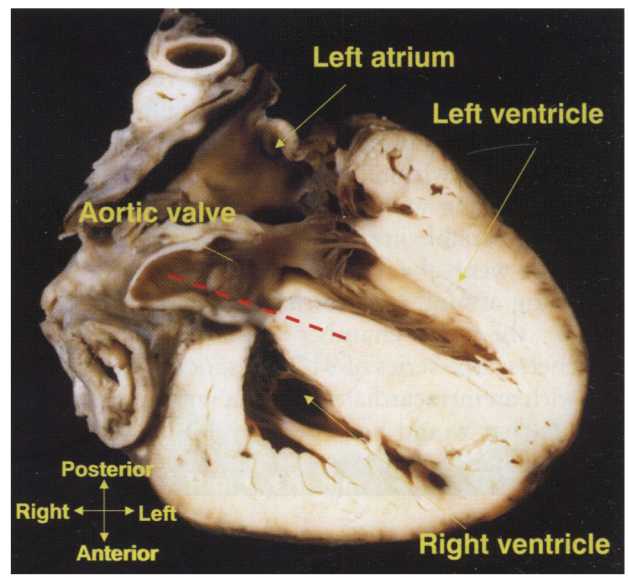


Figure 18.
The normal left ventricular outflow tract in a mid-trimester fetus showing its oblique course towards the right.

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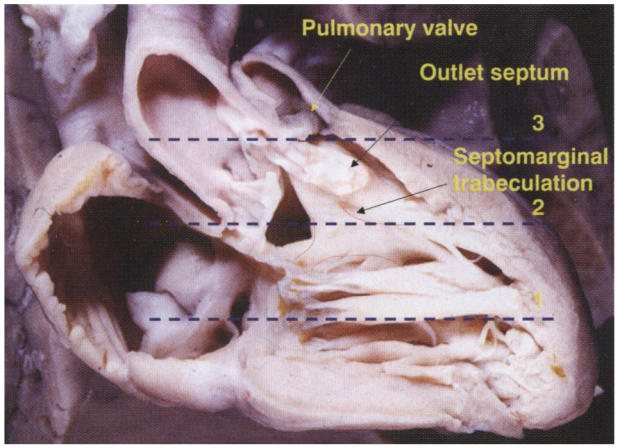

Figure 19.
The location of the ventricular septal defect in a fetus with tetralogy of Fallot, within the arms of the septomarginal trabeculation. The planes of section are numbered as in figure 3.

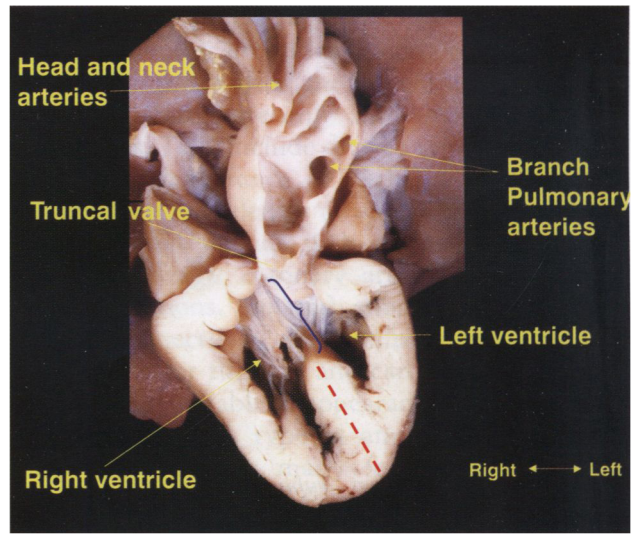

Figure 20.
View of the left ventricular outflow tract in a fetus with a common arterial trunk demonstrating lack of continuity between the anterior wall of the trunk and ventricular septum (bracketed). The plane of section in analogous to section 2 in figures 3, 4 and 19.

associated with abnormal ventriculo-arterial connections. Given that, in transverse sections, the first outflow tract in the fetus is normally directed towards its right shoulder (Fig. 18), and the second is directed towards the left of the spine (Fig. 21), the two should cross one another at the level of the arterial valves, passing from one outflow tract section to another. Again, such relationships are set very early in gestation at the time of division of the arterial trunks[21]. Failure to visualise this feature indicates abnormal ventriculo-arterial connections, the nature of which can be variable. In my pathological series, it most often indicated discordant connections. Overall, anomalies of the outflow tracts, either involving a ventricular septal defect or parallel outflow tracts, accounted for a further fifth of the series, leaving only one-tenth unaccounted for (Fig. 22).

High mediastinal views – the aortic and ductal arches

In order to detect the final group of anomalies, it is necessary to consider views taken much higher in the fetal thorax. These can be obtained by continuing the theoretical sweep cranially, to first the so-called 'three vessel view', and then to sections which visualise the ductal and aortic arches. The 'three-vessel' view may be a useful confirmatory or screening view, since it shows the relationship and size of the pulmonary trunk, ascending aorta, and superior caval vein.[22] In most instances, it will be superficial to views already described. For instance, the superior caval vein, aorta and a hypoplastic pulmonary trunk will be normally arranged in tetralogy of Fallot,[22] but this will have already been encountered in more caudal views. Moreover, the three vessel view will tell the fetal echocardiographer little or nothing about the connection of the aorta to the left or right ventricles, a feature which is more readily assessed in views of the left ventricular outflow tract.

In contrast, transverse views of the two arterial arches have considerable utility in the diagnosis of

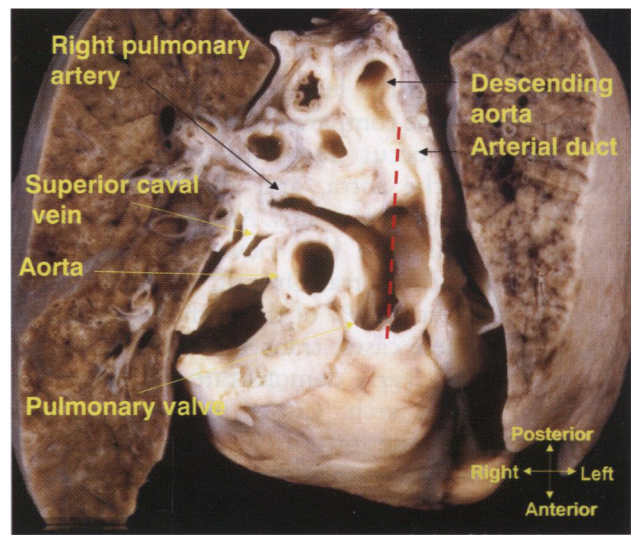

Figure 21.
The normal right ventricular outflow tract in mid-gestation which is the sagittal plane of the body.

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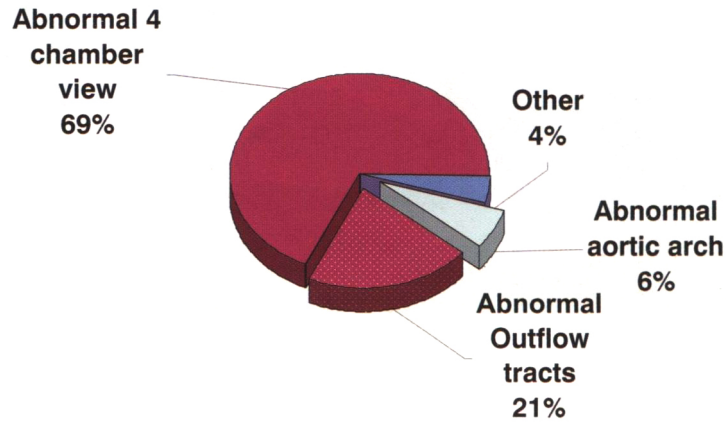

Figure 22.
Proportion of fetal specimens within the series accounted for by views of the outflow tracts and aortic arch.

anomalies of the aortic arch. When looking at the ductal and aortic arches from the above (Fig. 2), or from the left side (Fig. 23), it is evident that they are in differing planes of section. The aorta, arising from the center of the heart, initially passes towards the right shoulder, and then traverses the fetal thorax obliquely in order to reach the descending aorta to the left of the spine. The arterial duct is the direct continuation of the pulmonary trunk. It originates at the sternocostal margin of the heart, and sweeps directly back to meet the descending aorta in a sagittal plane. In addition, the aortic arch, when present, is always more cranial than the ductal arch. Consequently, only an oblique transverse section of both arches will allow direct comparison of their sizes (Figs 23, 24). In this view, the ductal arch is towards the left, and the aortic arch is more medial (Fig. 24). The two converge as an inverted 'Y' to the left of the spine, and their sizes can readily be compared. Differences in size of the two arches occur frequently in fetuses with coarctation of the aorta, and also in those rare fetuses with ductal aneurysms. Absence of the more medial aortic arch in this view should indicate interruption of the aorta. Overall, such anomalies account for a further 6% of the pathological series (Fig. 22). In these cases, transverse sections of the two arches can clearly demonstrate hypoplasia of the more medial arch. Closer inspection would demonstrate that the aortic arch enters into the side of the ductal arch, rather than merging with it, owing to the infolding of the aortic wall proximal to the origin of the arterial duct. This feature may be beyond the resolution of current ultrasound machines used for fetal echocardiography. It would be misleading not to state, however, that in many fetuses with coarctation of the aorta there may also be associated asymmetry between the right and left sides of the heart which is visible in the four-chamber plane. Although this feature was initially considered to be a sign of fetal coarctation, and can occasionally be quite marked, evidence obtained during fetal life shows that it is more variable than the presence of hypoplasia of the arch.[23] From an anatomic standpoint, this is no more than would be expected, since it is the malformation of the arch that is being diagnosed. Consequently, for the

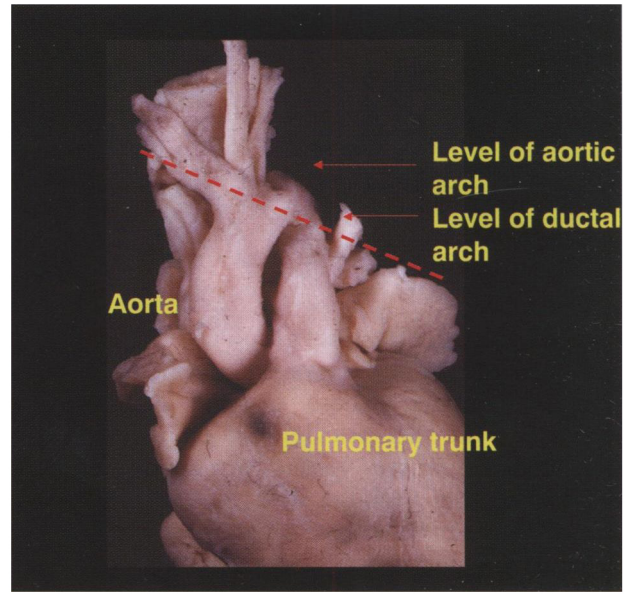

Figure 23.
The aortic and ductal arches as seen from the front of a normal 22 week gestation fetus, showing the plane of section required to image them simultaneously.

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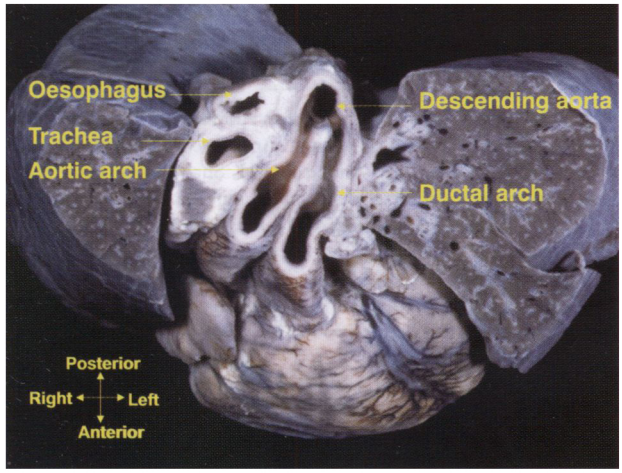

Figure 24.
Section obtained when cutting across both the aortic and ductal arch in the plane illustrated in Figure 23. This plane is useful for comparison of the size of the two arches that are normally similar but differ in coarctation of the aorta.

purposes of illustration, this sign has been conveniently ignored in my analysis.

Conclusions

This analysis has shown the basic manner in which cardiac defects can be detected from a sequential sweep through the fetal thorax. It demonstrates how the majority of cardiac defects obtained for correlation with fetal echocardiography in a tertiary setting will show abnormalities within the four-chamber plane. Nonetheless, up to one-third of the spectrum will require the fetal echocardiographer to examine more cranial views of the fetal thorax. The remaining group of cases, which are not accounted for within the four transverse views described, constitute only one twenty-fifth of the total series. They include diverse anomalies that are variable in appearance. It is, therefore, difficult to assign them confidently to any of the views so far covered. They include such anomalies as vascular rings and slings, coronary arterial to ventricular communications, isolated anomalous pulmonary venous connections, aorto-pulmonary window or left ventricular tunnel,[5] and cardiac teratomas, all of which are variable in appearance.

Of course, this type of analysis is only illustrative. Defects in many fetuses will show as abnormalities in both four-chamber and outflow tract sections. Once the main abnormalities have been identified, it is possible to refine the diagnosis further by introducing additional views of the fetus. For instance, it will usually be necessary to determine the position of the heart and stomach relative to the left and right of the fetus, as well assessing as the vascular arrangement within the thorax as a clue to atrial arrangement. Such associations are not infrequent in the fetus (Fig. 25), being abnormal in 12% of the current series of cases.[24] Their assessment cannot be ignored if the echocardiographer is to provide accurate prognostic information on the spectrum of cardiac anomalies found in the fetus.

Acknowledgement

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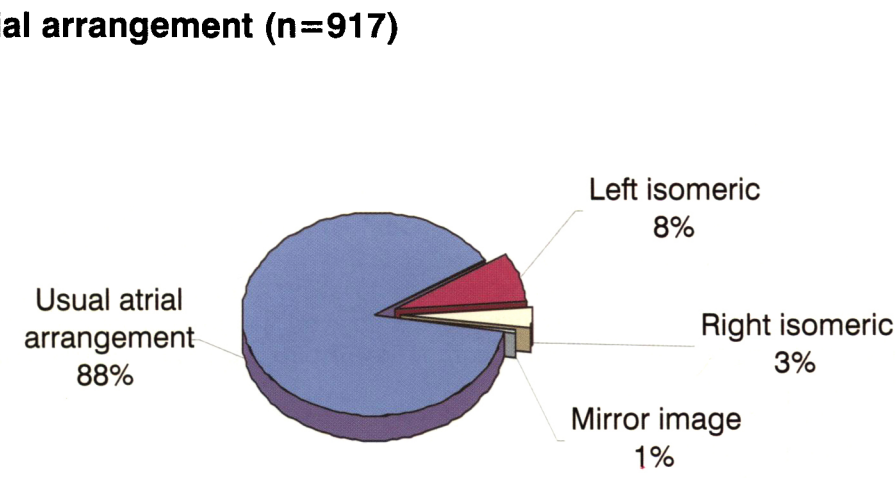

Figure 25.
Distribution of atrial arrangement amongst the total cohort of 917 fetal specimens.

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