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Echo-morphological correlates in atrioventricular valvar atresia

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As we described in the previous review,[1] double inlet ventricle is usually found with the atrial chambers connected to a dominant left ventricle, less frequently to a dominant right ventricle, and rarely to a solitary and indeterminate ventricle. As we have also discussed in this supplement,[2] double inlet to the left ventricle was, for many years, considered the exemplar of so-called "single ventricle", despite the fact that such patients unequivocally possess one big and one small ventricle. Echocardiographic interrogation has served to resolve this controversy, showing that such patients make up a significant proportion of those having functionally univentricular hearts. Such echocardiographic investigation has also served to resolve similar controversies regarding patients having tricuspid atresia. For some time, it was argued that patients with tricuspid atresia also had "univentricular hearts",[3] but the logic used to underscore this approach was just as flawed as that used to justify the use of "single ventricle" in patients with double inlet atrioventricular connection.[4,5] The increasing use of the Fontan procedure has served to demonstrate that these patients, along with many having mitral atresia in the setting of hypoplastic left heart syndrome, also have functionally univentricular arrangements. As we will show in this review, however, the anatomical substrates found in patients with atrioventricular valvar atresia are much more complex than those seen in the setting of double inlet ventricle. This is because atrioventricular valvar atresia can be produced either by absence of one atrioventricular connection, or by presence of an imperforate valvar membranes closing completely one or other of the two normal atrioventricular junctions. This important difference, combined with multiple segmental combinations, produces a bewildering array of potential anatomical substrates, with the complications magnified by the fact that, when one atrioventricular connection is absent, the other atrioventricular junction can be shared between the two ventricles, the so-called uniatrial and biventricular arrangement.[6] In our review, we will first describe the anatomical options, before concentrating our attention on the more frequent patterns seen in clinical practice.

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Anatomical substrates for atrioventricular valvar atresia

As indicated in our introduction, the key distinction to be made when analysing the substrates for atrioventricular valvar atresia is the difference between absence of one atrioventricular connection and an imperforate atrioventricular valve. For many years, it was believed that the substrate for "classical" tricuspid atresia was an imperforate valve interposed between the floor of the right atrium and the small right ventricle. This purported imperforate valve was believed to correspond with the so-called "dimple" seen in the atrial floor (Fig. 1a). In reality, the "dimple" overlies the atrioventricular component of the fibrous atrioventricular septum (Fig. 1b). If perforated using a pin, it can be shown that this structure separates the floor of the right atrium not from the small right ventricle, but rather from the subaortic outflow tract of the left ventricle. The muscular floor of the right atrium can

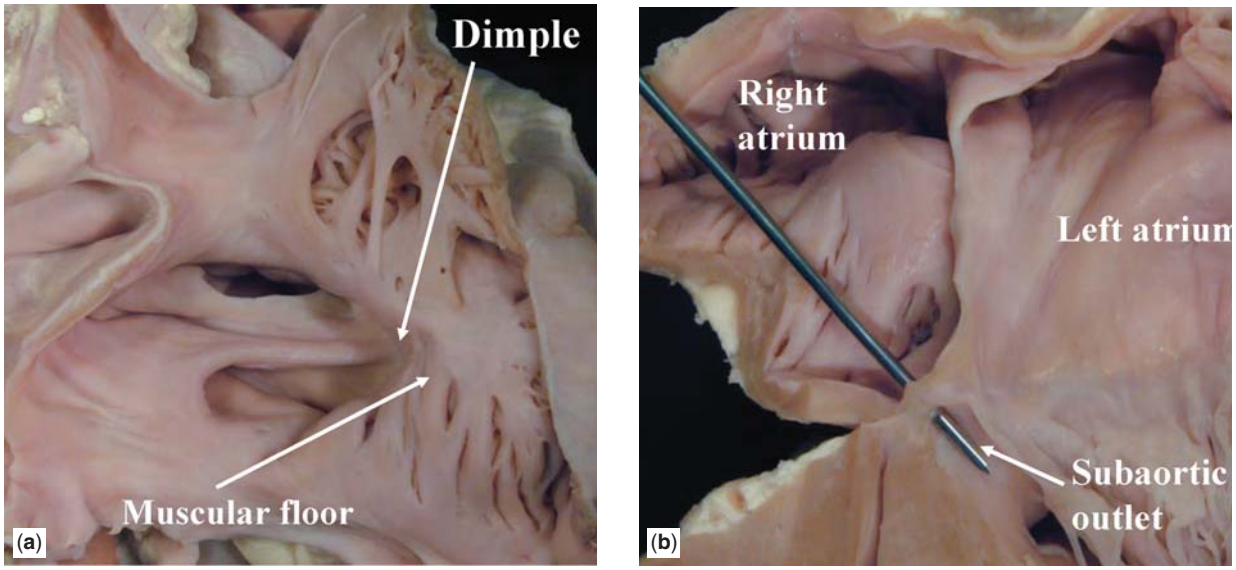

Figure 1.
*The photograph (**a**) shows the typical muscular floor of the right atrium seen when tricuspid atresia is produced because of absence of the right atrioventricular connection. Note the location of the so-called dimple. In the past, it was often thought that the dimple represented an imperforate valve, and separated the right atrium from the underlying right ventricle. As the section in (**b**) shows, however, the dimple overlies the atrioventricular component of the fibrous ventricular septum. There is no communication, either real or potential, between the right atrium and the incomplete right ventricle because of the absence of the right atrioventricular connection.*

be liberated in its entirety from the ventricular base, since the essence of the "classical" variant of tricuspid atresia is complete absence of the atrioventricular junction, including the inlet component of the morphologically right ventricle.[7] Thus, as we will show in our section devoted to echocardiographic findings, the majority of patients with tricuspid atresia have absence of the right atrioventricular connection (Fig. 2a). In this setting, the right ventricle is incomplete, being made up only of the apical trabecular and outlet components, albeit that rarely both arterial trunks can be supported by the right ventricle, or even more rarely both arterial trunks can arise from the dominant left ventricle. In this latter situation, the right ventricle will be no more than a trabeculated pouch. More usually, the right ventricle gives rise to the pulmonary trunk, in other words the ventriculo-arterial connections are concordant, although discordant ventriculo-arterial connections, or transposition, are by no means rare in patients with tricuspid atresia. Determination of the ventriculo-arterial connections, and establishment of the size and location of the ventricular septal defects, are amongst the most important tasks for the echocardiographer assessing the anatomy of the patient with tricuspid atresia, although the state of the atrial septum, and assessment of any other associated malformations, must not be ignored.

In a small proportion of cases, the echocardiographer will find unusual morphology at the site of the right atrioventricular junction. This is when the atresia is produced by an imperforate valve rather than by absence of the right atrioventricular connection. The imperforate valve can be a hypoplastic structure, and this is typically seen in the setting of hypoplasia of the right heart with intact septum, when not only the pulmonary valve but also the tricuspid valve is imperforate (Fig. 2b). Less frequently, the hypoplastic imperforate right valve can separate the floor of the right atrium from the dominant left ventricle, producing double inlet left ventricle with imperforate right atrioventricular valve. It is also possible to find a much larger imperforate valve interposed between an atrialised part of the right ventricle and the apical trabecular component of the right ventricle. This is seen when the imperforate valve is also deformed by Ebstein's malformation (Fig. 2c). It is also important for the echocardiographer to be aware of, and recognise, the unusual situation in which the right atrioventricular connection is absent, as in classical tricuspid atresia, but in which the left atrioventricular valve straddles across the ventricular septum, with overriding of the left atrioventricular junction (Fig. 2d). This variant, the uniatrial but biventricular atrioventricular connection, can be seen not only with right hand ventricular topology, when the incomplete right ventricle will be positioned anteriorly and superiorly relative to the morphologically left ventricle (Fig. 3a), but also with left hand topology, when the right-sided ventricle will possess the apical trabeculations of the morphologically left ventricle, and will be positioned postero-inferiorly and to the right of the

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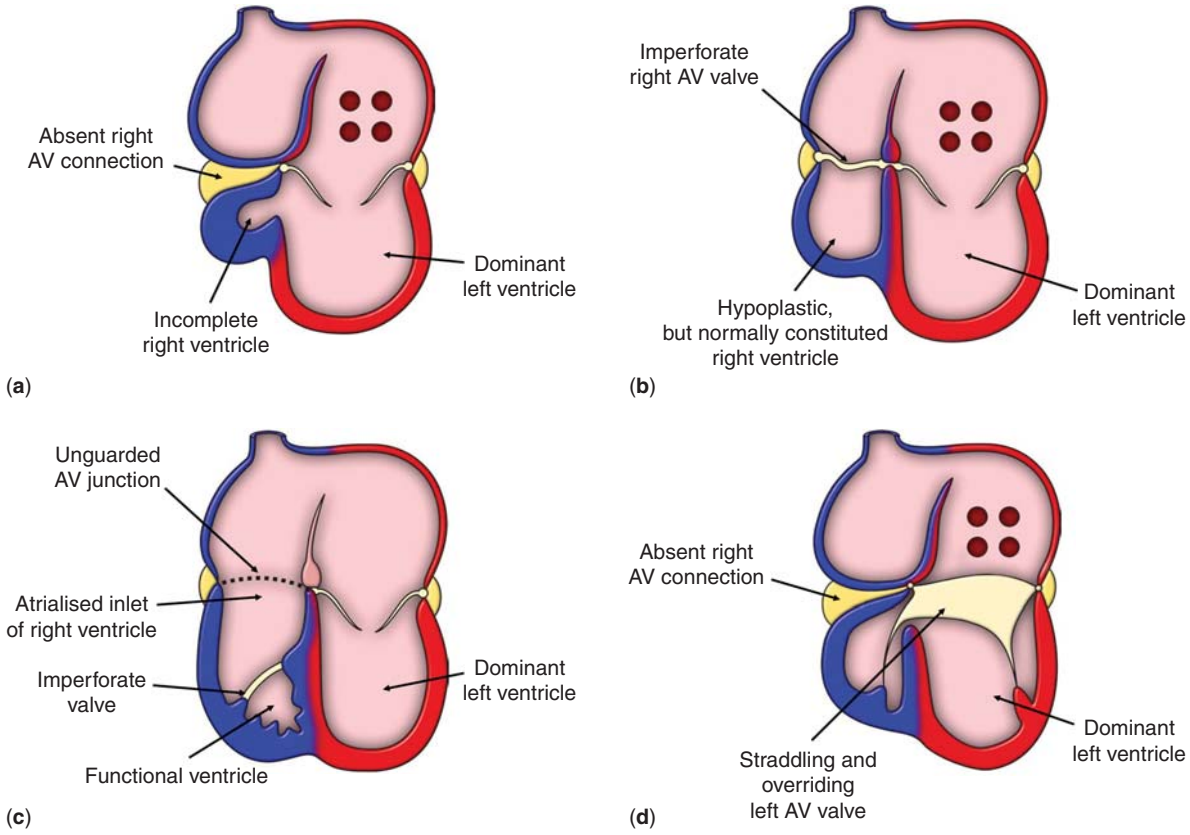

Figure 2.
*The figures show the different phenotypic features of tricuspid atresia as produced by (**a**) absence of the right atrioventricular connection, (**b**) an imperforate atrioventricular valve with biventricular atrioventricular connections, (**c**) an imperforate atrioventricular valve with biventricular atrioventricular connections in the setting of Ebstein's malformation, and (**d**) when the solitary atrioventricular valve straddles and overrides in combination with absence of one atrioventricular connection, producing a uniatrial but biventricular atrioventricular connection. Morphologically right structures are coloured blue, whilst morphologically left structures are coloured red.*

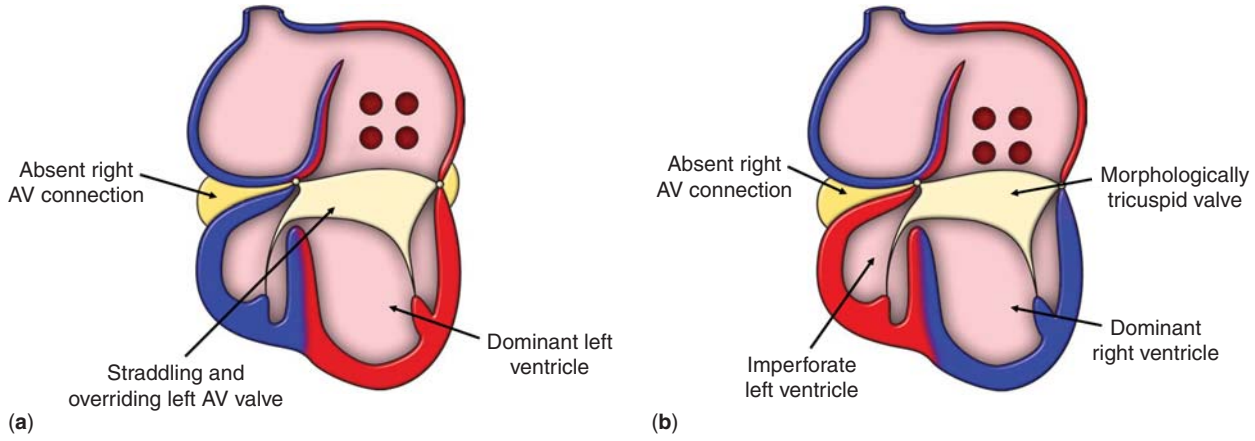

Figure 3.
*The figures show how absence of the right atrioventricular connection, when found with straddling and overriding of the left atrioventricular valve, can exist with either (**a**) right handed ventricular topology or (**b**) with left handed topology. With right handed topology, the left valve will likely be of mitral morphology, whereas it will be of tricuspid morphology with left hand topology. The structure of the right atrium, however, will not change. The colour coding is as for Figure 2.*

morphologically right ventricle (Fig. 3b). In this setting of left hand topology, the straddling left-sided atrioventricular valve will be of tricuspid morphology, and a strong case can be made for describing the heart as showing mitral atresia (Fig. 3b). The morphology of the blind-ending right atrium, nonetheless, is indistinguishable from that seen in classical tricuspid atresia. As we will see, this problem of

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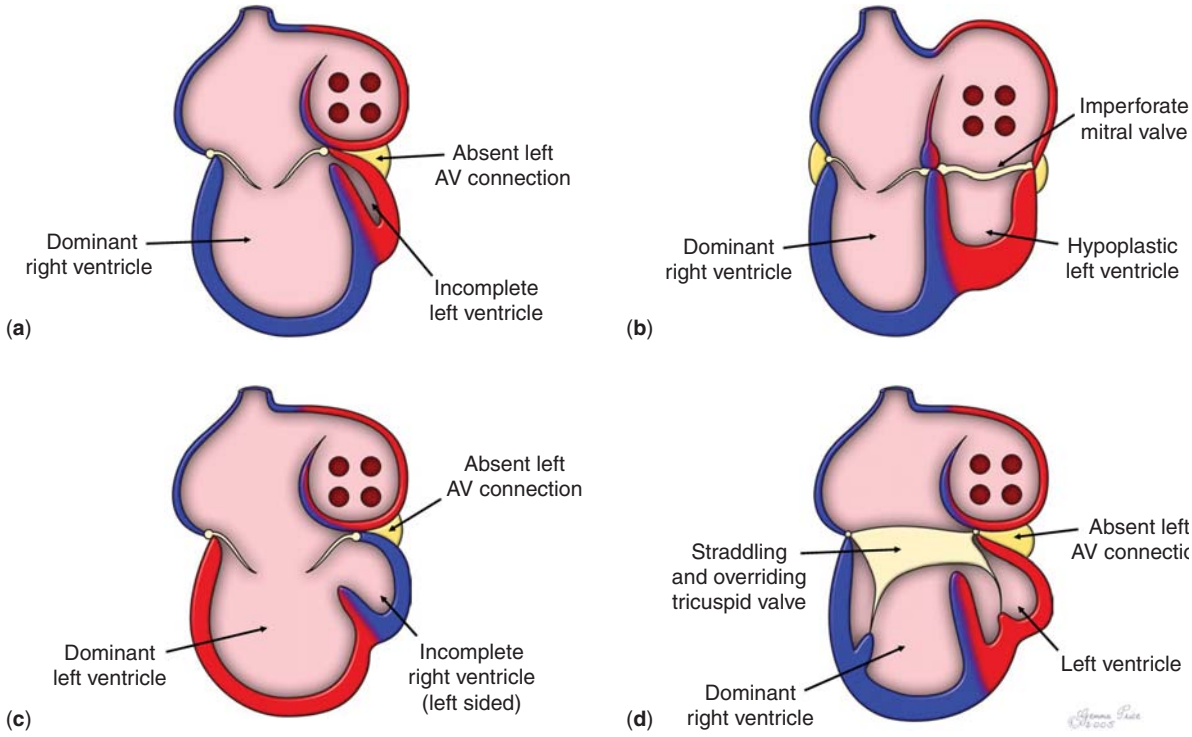

Figure 4.
*The figures show the varying phenotypic arrangements to be found producing so-called "mitral atresia". In (**a**), the typical form of hypoplasia of the left heart is shown with absence of the left atrioventricular connection, whereas (**b**) shows the variant with concordant atrioventricular connections and an imperforate mitral valve. Figure (**c**) shows the situation where the left atrioventricular connections is absent, but the right atrium is connected to a dominant left ventricle. In this setting, the incomplete right ventricle will be positioned antero-superiorly, and the right atrioventricular valve will be of mitral morphology. Figure (**d**) shows the variant with overriding and straddling of the right atrioventricular valve. This can be found with right hand topology, as illustrated, but also with left hand topology. The colour coding is as for Figures 2 and 3.*

naming the atretic valve can also arise when the left-sided atrioventricular connections is absent. In such rare cases, therefore, there is much to be said for describing the side of the absent connection, along with the position of the ventricles, rather than seeking to nominate the case as showing either tricuspid or mitral atresia.

The distinction between an absent connection and an imperforate valvar membrane must also be made when it is the left, rather than the right, atrium which is blind ending.[8–10] In the patient with usual atrial arrangement, this situation produces mitral atresia. Either absence of the connection, or an imperforate valve, can be seen when the heart itself falls into the category of hypoplastic left heart syndrome (Fig. 4a, b). When there is typical hypoplasia of the left heart, then it is the morphologically right ventricle that is dominant, and the heart functionally univentricular, irrespective of whether the left ventricle is itself complete or incomplete. Absence of the left atrioventricular connection can also be found with some frequency when the right atrium is connected to a dominant left ventricle (Fig. 4c). In this setting, the right ventricle is typically left sided, and positioned antero-superiorly relative to the dominant morphologically left ventricle. Usually the right ventricle gives rise to the aorta. Had a left-sided atrioventricular connection developed in this setting, then almost certainly it would have been guarded by a morphologically tricuspid valve. Thus, a case can again be made for describing this particular combination as tricuspid atresia, despite the fact that it is the pulmonary venous atrium that is blind ending. We find this potentially confusing, and would recommend describing absence of the left atrioventricular connection, along with connection of the right atrium to the left ventricle, and adding the information with regard to the ventriculo-arterial connections.[11] Similar sequential segmental descriptions also facilitate understanding of those rare cases in which the right atrioventricular valve straddles and overrides the ventricular septum when the left atrioventricular connection is absent (Fig. 4d). These rare lesions, like their right-sided counterparts, show the uniatrial

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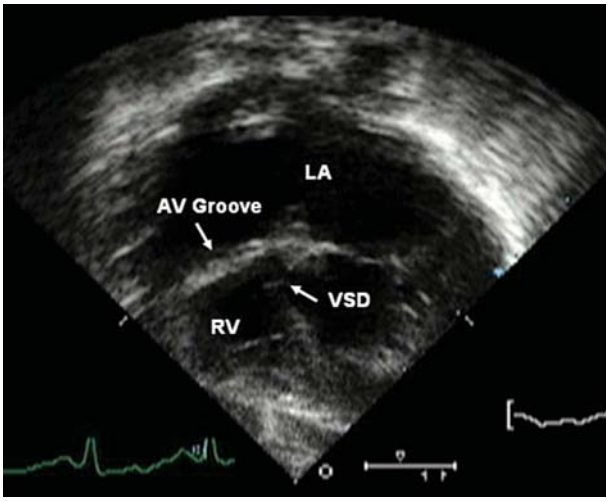

Figure 5.
This subcostal view is from a patient with the commonest variant of tricuspid atresia, that with absence of the right atrioventricular connection. Note the fibro-fatty tissue of the atrioventricular (AV) groove, which interposes between the left atrium (LA) and the incomplete right ventricle (RV), and the location of the ventricular septal defect (VSD).

and biventricular connection. They can be described as "double outlet atrium", but it must then be remembered that this label is non-specific, since there can be right or left hand topology (Fig. 3). Imperforate valves must also be anticipated to exist in any possible type of atrioventricular connection, such as double inlet left or right ventricle. Either an absent left atrioventricular connection, or an imperforate left atrioventricular valve, is a possibility when there is a solitary and indeterminate ventricle, as indeed is an imperforate right atrioventricular valve or an absent right atrioventricular connection. The anatomical heterogeneity in the setting of atrioventricular valvar atresia, therefore, is considerable and potentially bewildering, albeit that all the features described above are readily recognised using cross-sectional echocardiographic interrogation.

Echocardiographic interrogation

It is now expected that the echocardiographic evaluation of patients with suspected atrioventricular valvar atresia should follow the segmental approach, where we examine first the venous segment, then the veno-atrial connections, before turning to interrogation of the atrial chambers themselves, the atrioventricular connections, the make-up of the ventricular mass, the ventriculo-arterial connections, and finally the arrangement of the intrapericardial arterial trunks and their branches.

When considering the specific details of tricuspid and mitral atresia, the key features demanding

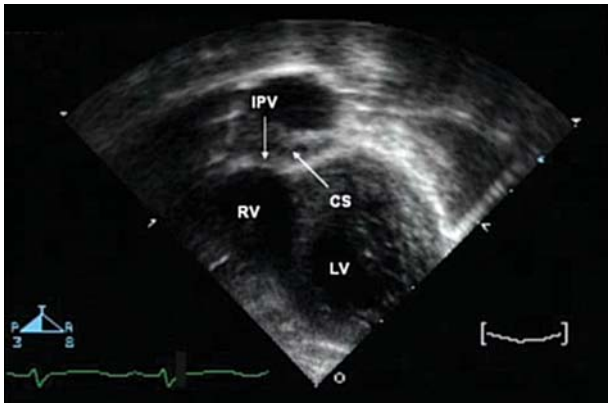

Figure 6.
This four-chamber image shows tricuspid atresia produced by an imperforate tricuspid valve (IPV) in the setting of concordant atrioventricular connections. Note the relationship to the coronary sinus (CS). RV, LV: right and left ventricles.

recognition are the morphological substrate producing the valvar atresia, the characteristics of the dominant ventricle, and the position and size of a second incomplete ventricle if present. Attention should then be directed to visualisation of the nature and number of ventricular septal defects, characterisation of the ventriculo-arterial connections, paying particular attention to the presence or absence of obstruction, and the nature of any and all associated lesions. The latter feature is particularly significant, since up to one-sixth of patients with tricuspid atresia may have a persistent left superior caval vein draining to the coronary sinus, while aortic coarctation is a frequent accompaniment of discordant ventriculo-arterial connections.[12]

In the greater majority of patients with tricuspid atresia, 95% in a recent study,[7] it is the fibro-fatty tissue of the atrioventricular groove interposed between the muscular floor of the right atrium and the parietal wall of the ventricular mass that is the substrate for the valvar atresia. This is because of absence of the right atrioventricular connection. The subcostal and apical views show well the salient anatomy, illustrating the location of the atrioventricular groove between the right atrium and incomplete right ventricle (Fig. 5). These views also show well the size of the ventricular septal defect, and the features of the incomplete right ventricle, which usually possesses only apical trabecular and outlet components. In the remaining one-twentieth of cases, the valvar atresia will be produced by an imperforate tricuspid valve. As discussed above, this morphology is typically encountered either with coexisting pulmonary atresia and an intact ventricular septum, or in the setting of imperforate Ebstein's malformation. The arrangement is shown to advantage using the four-chamber approach (Fig. 6). The

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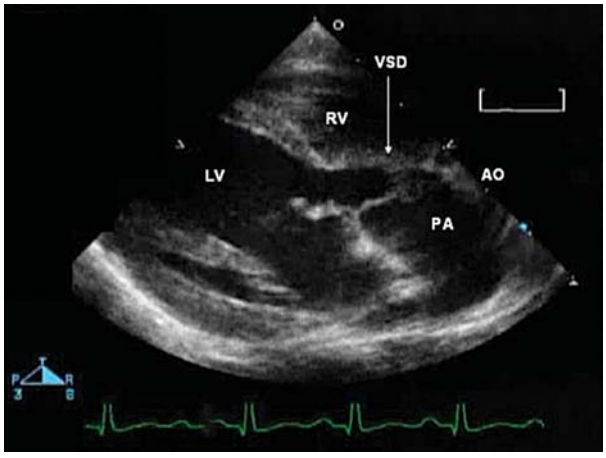

Figure 7.
This parasternal long axis view is form a patient with tricuspid atresia due to absence of the right atrioventricular connection, with dominant left (LV) and incomplete right (RV) ventricles. The image shows well the morphology of the ventricular septal defect (VSD), in this case with discordant ventriculo-arterial connections (transposition). AO: aorta; PA: pulmonary trunk.

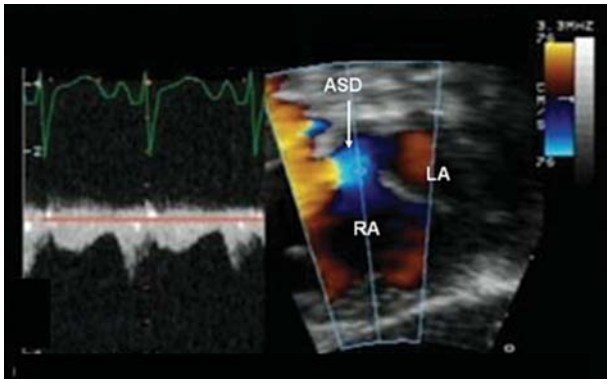

Figure 8.
In this subcostal image of a patient with tricuspid atresia, the cursor has been placed to permit colour flow and pulse wave Doppler interrogation of the flow across the interatrial communication (ASD), showing no restriction of flow. RA, LA: right and left atriums.

parasternal long axis views are of greatest value in assessing the relationships of the intrapericardial arterial trunks, and in confirming the location and number of ventricular septal defects (Fig. 7).

Doppler is useful in the assessment of the state of the atrial septum. The right-to-left shunt is obligatory in the setting of tricuspid atresia (Fig. 8), but occasionally the interatrial communication, which may be no more that a patent oval foramen, can be restrictive. It is particularly important to be aware that absence of the right atrioventricular connection can co-exist with straddling and overriding of the left atrioventricular valve. This produces the uniatrial but biventricular connection. The echocardiographic images will show the left atrium in connection with

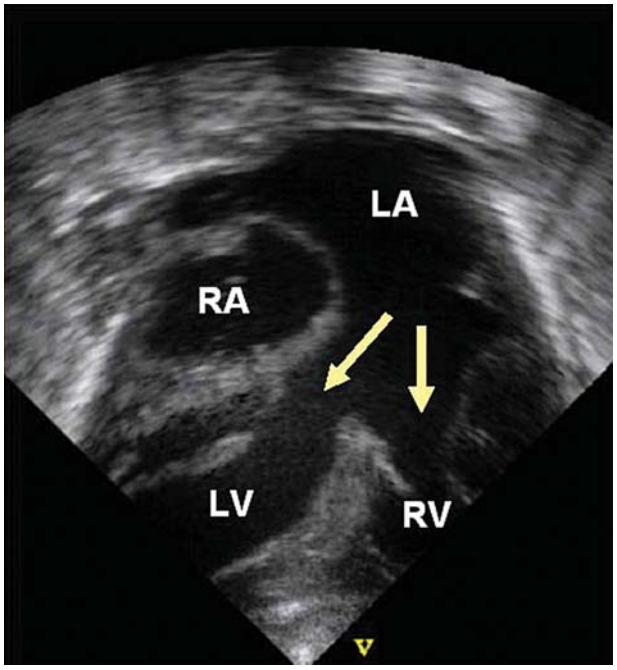

Figure 9.
This four-chamber section, reproduced by kind permission of Dr Jan Marek, from Great Ormond Street Hospital for Children, London, shows absence of the right atrioventricular connection with straddling and overriding of the left atrioventricular valve, which connects the left atrium (LA) to both ventricles (arrows). The fibro-fatty tissue of the right atrioventricular groove separates the right atrium (RA) from the ventricular mass. Note that both ventricles (RV, LV) are incomplete in this lesion, which produces a uniatrial but biventricular connection.

both the right and left ventricles, both ventricles them being incomplete to greater or lesser degree (Fig. 9). When found, it is also important to confirm the ventricular topology, since remarkably similar atrial anatomy can be found when the incomplete left ventricle is the smaller chamber, being positioned posteroinferiorly and rightward within the ventricular mass.

Atresia of the left atrioventricular valve is assessed echocardiographically in a similar manner to that described above. Using subcostal and apical views, it should always be possible to distinguish absence of the left atrioventricular connection, the most common pattern, from an imperforate left atrioventricular valve (Fig. 10). Note should be taken, of course, of the morphology of the dominant ventricle, since when there is atresia of the left-sided atrioventricular valve, the morphologically right atrium may drain into the dominant morphologically right ventricle, or to a dominant morphologically left ventricle (Fig. 11). As with tricuspid atresia, it is essential to establish the relationships of the intrapericardial arterial trunks, and the state of the atrial septum (Fig. 12). This latter feature is the more important in the setting of left-sided atrioventricular valvar atresia, since

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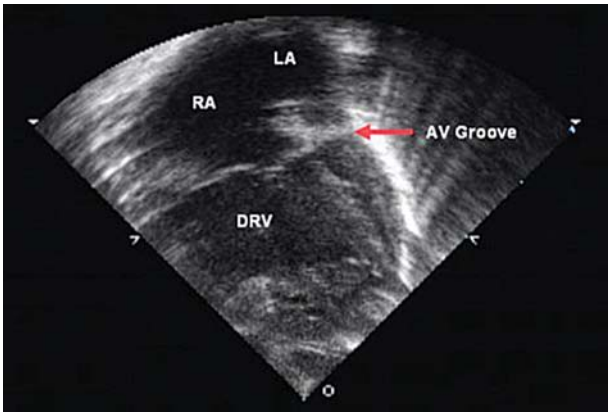

Figure 10.
This apical view shows atresia of the left-sided atrioventricular valve due to complete absence of the atrioventricular connection. Note that fibro-fatty tissue of the atrioventricular (AV) groove interposes between the left atrium (LA) and the ventricular mass. The right atrium (RA) connects to a dominant morphologically right ventricle (DRV). Note also the position of the incomplete left ventricle postero-inferiorly and to the left.

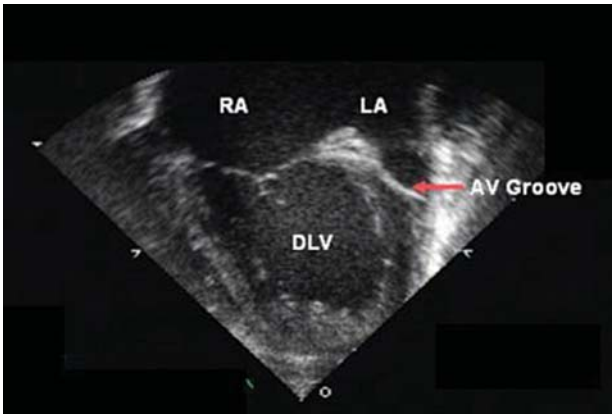

Figure 11.
This four-chamber view shows absence of the left-sided atrioventricular connection, again with the fibro-fatty tissue of the atrioventricular (AV) groove underlying the floor of the left atrium (LA), but this time with the right atrium connected to a dominant morphologically left ventricle (DLV). The incomplete right ventricle is not seen. It was positioned antero-superiorly and to the left.

a restrictive atrial septum reduces markedly the egress of pulmonary venous return.

Many patients with atresia of the left atrioventricular valve will have hypoplastic left heart syndrome. In this setting, the left ventricle will be grossly hypoplastic, typically with co-existing aortic atresia. The echocardiographic assessment, which again will include a full segmental approach, should include subcostal or apical views. These will show the size of the left ventricle, often no more than a virtual slit, and should be helpful in assessing the state of the atrial septum, which could again be restrictive. Parasternal

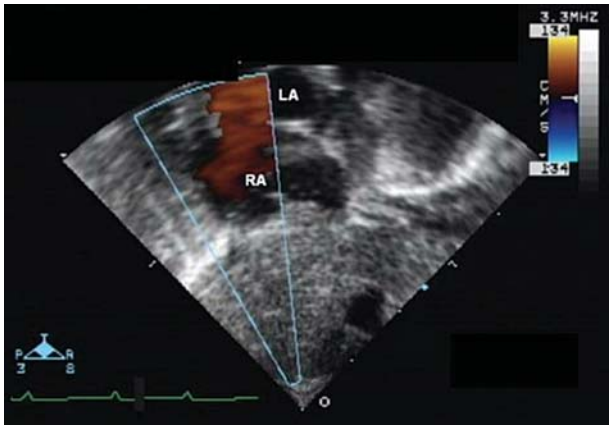

Figure 12.
This subcostal view shows the obligatory left-to-right shunt found at atrial level in the setting of mitral atresia, the red colour showing the flow from left (LA) to right atrium (RA).

long axis views will confirm the size of the small or virtually absent left ventricle, showing also the small ascending aorta and the dominant right ventricle. Care must be taken to identify a ventricular septal defect if present.

Conclusion

When the investigator is aware of the anatomical potential for variability, then echocardiographic interrogation is fully capable of revealing all the necessary details of anatomy and flow required to determine the appropriate therapeutic approach to patients with atrioventricular valvar atresia. Assessment nowadays begins with the prenatal scan. Even at this stage, most of the anatomical details should be discernible. Continuing assessment will then be needed as the patients are followed through childhood, during adolescence, and into adulthood. Visualisation of the various malformations will become even better, and more precise, with the more widespread availability and use of three-dimensional techniques.

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