ECHOCARDIOGRAPHY has been well established as a noninvasive tool in the diagnosis of congenital heart disease.1-3 Although endocardial cushion defects (ECD) occur with a frequency of approximately 3% of all congenital cardiac malformations,4 echocardiographic descriptions in the literature are scanty and limited to the discussion of atrial septal defect 1° (ASD 1°), the incomplete form of ECD.5,6

Due to the advances in surgical techniques, it has become increasingly important to distinguish with certainty patients with ASD 1° from those with complete A-V canal defects (CAVC). In this report we describe features common to the general classification of ECD as well as specific findings in those patients with CAVC.

Methods

Echocardiographic studies were performed in 28 patients with ECD ranging in age from two weeks to 16 years. Sixteen patients had CAVC and 12 an ASD 1°. Seven of the patients had Down’s syndrome (Tay-Sachs 21). Each patient had typical clinical and electrocardiographic findings of ECD.4 In addition, cardiac catheterization and left ventricular angiography confirmed the diagnosis in each case by catheter passage, goose-neck deformity of the mitral valve, and the scalloped right border of the left ventricular outflow tract during systole. In addition to the angiographic appearance, the presence of pressure equalization between the two ventricles was required for the diagnosis of CAVC. Further anatomic detail was obtained in nine patients at corrective surgery and in one other at post mortem examination. To define the precise spatial relationship between the A-V valves and ventricular landmarks, 30 pathological specimens of ECD were examined in detail.

Echocardiograms were performed with a Hoffrel model 101B ultrasonoscope using either an Aerotech 5.0 MHz transducer with a ¾ inch active diameter (in neonates and small children) or a Hoffrel 3.25 MHz transducer with an active diameter of ¾ inch. The echocardiographic signal was relayed to either a Cambridge Fiberoptic multichannel recorder or photographed on 35 mm film.

The transducer was placed at the mid-left sternal border until echoes from the left ventricular septum and left ventricular posterior wall were displayed. The anterior leaflet of the mitral valve was located by systematically moving the transducer superiorly and inferiorly from the second to the fifth left intercostal spaces in a progressively leftward direction. In older patients the tricuspid valve (TV) was most often recorded at the left lower sternal border. In small infants, however, it was often possible to obtain optimal TV echoes from a mid-sternal position or even at the right sternal border. In one patient with CAVC, the

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Received September 25, 1973; revision accepted for publication November 20, 1973.

SUMMARY

The echocardiographic features of endocardial cushion defects were defined in 28 patients. Two basic characteristics, common to both complete and incomplete forms of endocardial cushion defects, were found. First, the mitral valve excursion was not seen best in the usual location as defined by intraventricular landmarks. Secondly, the mitral valve echoes in cases where there are dense attachments of the mitral leaflet to the crest of the interventricular septum may be traced to the rightward and anterior border of the left ventricular outflow tract. This is the echocardiographic representation of the characteristic "goose-neck" deformity seen angiographically.

Two additional patterns were recognized in patients with complete atrioventricular (A-V) canal. In Pattern I, a large, single, A-V valve was found occupying a central location between the two ventricles. During diastole, the superior segment moved anteriorly into the right ventricle and the inferior segment posteriorly into the left ventricle. In Pattern II, a large single A-V valve moved from the left ventricle in systole into the right ventricle in diastole. This pattern was specific to patients with common A-V valve leaflets.

Additional Indexing Words:
Ultrasound Complete atrioventricular canal Atrial septal defect Ventricular septal defect

Echocardiographic Features of Endocardial Cushion Defects

By Roberta G. Williams, M.D., and Michael Rudd, B.S.
origin of the TV echo was confirmed at cardiac catheterization using a technique similar to that described by Gramiak with a power injection of 5% dextrose in 0.2% of NaCl into the right ventricle while echoes from the TV were recorded. Dense echoes produced by the cavitation effect of the injected solution filled the area between the valve echo and the anterior heart border.

Results

Characteristically, optimal mitral valve echoes in patients with ECD are not displayed in the usual location, position 2 of Feigenbaum. Although echoes from the mitral valve may be registered by scanning the area in which septal and left ventricular posterior wall echoes are found, the valve is usually seen only during systole and early diastole, moving out of the transducer beam for much of mid and late diastole (fig. 1). This feature was present to a varying degree in 23 of 28 patients, most prominently in those with CAVC and least obviously in those with ASD 1° without severe mitral regurgitation. As the transducer beam is directed in a superior and medial direction toward the left ventricular outflow tract, the anterior leaflet of the mitral valve echo is traced in a progressively more anterior direction until it forms the anterior border of the aortic root (fig. 2). This finding was not appreciated in most of the earlier studies, recorded on 35 mm film but was quite evident in six of seven patients studied with a strip chart recorder. In 14 of 28 patients, echoes of the anterior leaflet of the mitral valve could also be traced superiorly to a typically appearing mitral valve ring (fig. 3).

Figure 2

On scanning inferiorly from the base of the heart to the body of the left ventricle, anterior mitral leaflet echoes emerge from the anterior border of the left ventricular outflow tract (LVOT); this raises the floor of the LVOT to form the echocardiographic equivalent of the angiographic goose-neck deformity. See legend to figure 1 for abbreviations.

Figure 1

Typical echocardiogram of a patient with ASD 1°. The transducer is in position 2 of Feigenbaum. Mitral valve echoes are present throughout systole but disappear after the early diastolic opening movement. Note the presence of a dilated right ventricle and pattern of septal reversal which is characteristic of right ventricular diastolic overload. RVB = right ventricular body; TV = tricuspid valve; IVS = interventricular septum; ALMV = anterior leaflet of the mitral valve; LVPW = left ventricular posterior wall; LVB = left ventricular body.

Figure 3

Mitral valve echoes traced from the aortic root as seen in the patient with CAVC. Parallel echoes representing the aortic root with interposed faint echoes from a cusp of the aortic valve (AoV) are seen on the left-hand side of the illustration. As the transducer is rotated inferiorly, echoes from the posterior border of the aortic root are shown to be continuous with those of the mitral leaflet (ALMV). The slightly anterior position of the ALMV (0.5 cm) relative to the posterior border of the aortic root is most likely due to valve position.
Again, the patients in whom this feature was not recorded were studied without benefit of a strip recorder.

In patients with CAVC, two additional specific patterns of valve motion were observed. In pattern I (fig. 4), seen in ten of 16 patients, during systole the anterior leaflet of the tricuspid valve and anterior leaflet of the mitral valve were seen close together in the plane of the interventricular septum. With the onset of diastole, the leaflets move apart, the tricuspid anteriorly, with a broad excursion into the right ventricle, and the mitral valve posteriorly into the left ventricle with a somewhat damped motion and short over-all excursion (less than 1 cm). Absence of ventricular septal echoes between the two valve leaflets in diastole was noted. During systole, multiple echoes, probably representing redundant valve tissue, were seen associated with the central location of the valve leaflets.

In seven of 16 patients with CAVC, an unusual pattern of A-V valve motion could be seen (Pattern II). A single A-V valve leaflet was demonstrated in a posterior position in the left ventricle during systole and appeared to move into a markedly anterior position in the right ventricle with the onset of diastole (fig. 5). By scanning superiorly, it was possible to demonstrate that this valve leaflet was continuous with the mitral valve ring. By scanning inferiorly echoes from the valve leaflet were lost as the interventricular septum came into view. The patients in whom this pattern was seen appeared to have a common A-V valve by angiography.

Patterns I and II were present only in patients with CAVC. On one patient with CAVC, both patterns could be demonstrated.

**Discussion**

In a recent radiologic review by Baron, the anatomic features of ECD were discussed. He described the attachment of the anterior leaflet of the mitral valve to the crest of the interventricular septum. The superior and inferior segments of the cleft anterior leaflet of the mitral valve were shown to form the right border of the left ventricular outflow tract (LVOT) during systole, taking the place normally occupied by the atrioventricular septum. With diastole, both segments swing independently into the left ventricular cavity. In addition, our own review of the anatomy of ECD showed that the superior segment is stretched out like a sail across the left ventricular body with normal attachment superiorly and laterally to the mitral valve ring as well as abnormal attachment to the crest of the interventricular septum. The result of these attachments is that the superior segment of the anterior leaflet of the mitral valve moves in a plane that is roughly parallel to the ventricular septum rather than perpendicular to it as in the normal heart (fig. 6). During diastole it moves superiorly, raising the “floor” of the LVOT and forming the characteristic “goose-neck” deformity seen in the angiocardiogram of these patients. It is the result of this abnormal orientation of the anterior leaflet of the mitral valve that echoes from this structure were not usually found in the normal position. The best echoes of the anterior leaflet of the mitral valve were recorded instead from the area of the LVOT.

The two echo patterns characteristic of CAVC, not identified in any of the patients with ASD 1°, could be explained by a careful review of the anatomy. In Pattern I, two A-V valve leaflets were seen lying together in the plane of the ventricular
ENDOCARDIAL CUSHION DEFECTS BY ULTRASOUND

Figure 5

(Left panel) This is an illustration of the common atrioventricular (A-V) valve of an infant with complete atrioventricular canal (CAVC). In systole it lies just anterior to the left ventricular posterior wall (LVPW). In diastole it moves 2 cm anteriorly to touch the anterior right heart border (RHB). (The width of the entire heart in this plane is only 3 cm). (Right panel) Scanning inferiorly from the aortic root in another patient with CAVC, a large A-V valve excursion is seen which is continuous with the posterior border of the aortic root. Note the absence of interventricular septal echoes in the more inferior position where the A-V valve appears to move from its posterior position in systole to touch the RHB in diastole. No other A-V valve, separate from this one, was seen in this patient. \( RV = \) right ventricle; \( CAVV = \) common atrioventricular valve; \( RHB = \) right heart border; \( LVPW = \) left ventricular posterior wall; \( LA = \) left atrium.

Figure 6

The above diagrammatic sketch shows the position and attachments of the ALMV (dark shaded area). One can see that the ALMV is attached abnormally to the interventricular septum (IVS) as well as normally to the mitral valve ring in patients with ECD. A cleft separates the ALMV into superior and inferior portions. White arrows show the resulting direction of mitral valve movement. Tethered between its attachments to the IVS medially, the MVR superiorly, and the antero-lateral papillary muscle laterally, the superior segment of the ALMV lifts upward into the left ventricular outflow tract during diastole, forming the typical goose-neck deformity seen by angiography. Contrasted to this is the motion of the normal ALMV relatively perpendicular to the IVS.

Circulation, Volume XLIX, March 1974

Septum during systole. With the onset of diastole, these leaflets moved apart into their respective anterior and posterior ventricles. Careful examination, with a probe simulating the ultrasonic beam, of the single autopsy specimen of a patient in whom this feature was recorded suggested that the portions of the A-V valves most likely to create this pattern were the anterior leaflet of the tricuspid valve and the inferior segment of the cleft anterior leaflet of the mitral valve.

In some patients with CAVC, the superior segment of the anterior leaflet of the mitral valve is continuous with the anterior leaflet of the tricuspid valve through the ventricular septal defect, forming a common anterior leaflet. In patients with a Pattern II echocardiogram, a single A-V valve could be seen moving from a posterior position in the left ventricle during systole to an anterior position in the right ventricle in diastole. Upon review of the anatomy, it would seem impossible for any portion of the common anterior leaflet to move from the left ventricle into the right ventricle since the plane of motion is parallel to the ventricular septum, not
perpendicular to it, and because there are attachments of the leaflet in both ventricles. The mechanism postulated for this finding is shown in figure 7. The ultrasonic beam intersects the common anterior leaflet in the left ventricle during systole when it is in its closed posterior position. As the valve opens in diastole, the leaflet moves superiorly. As this occurs, the ultrasonic beam crosses the leaflet in progressively anterior positions until, in its fully open position, the beam intercepts the common leaflet well anteriorly in the right ventricle. In a sense, the transducer beam is held in one position while the common anterior leaflet is "scanning itself." An objection to this theory is the theoretical consideration that echoes only return from surfaces nearly perpendicular to the ultrasonic beam. We postulate that while the major axis of the leaflet is not perpendicular to the beam from the transducer, the nodular, redundant leaflet provides sufficient surfaces that are perpendicular to the beam.

Conclusions
Because of the abnormal orientation and chordal attachments of the anterior leaflet of the mitral valve to the ventricular septal crest in ECD, its resultant motion during the cardiac cycle is in a plane which is roughly parallel to that of the intact ventricular septum rather than perpendicular to it as is normally the case. As a result, echoes from the anterior leaflet of the mitral valves are not usually seen best in the usual transduced position which displays ventricular septal and left ventricular posterior wall echoes, but can be traced to where they form the rightward anterior border of the left ventricular outflow tract.

In patients with a large ventricular septal defect component (CAVC), two characteristic patterns of valve motion were seen. In one, the anterior leaflet of the tricuspid valve and the anterior leaflet of the mitral valve could be seen moving apart in diastole and returning to a central position in the plane of the interventricular septum during systole. The interventricular septum was not identified between them. In the other pattern, there appeared to be a common anterior leaflet which "scanned itself" during the cardiac cycle, giving a single A-V valve echo with an apparent large excursion.

Acknowledgment
We are indebted to Richard Meyer, M.D., for his helpful review of the manuscript.

References
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Circulation. 1974;49:418-422
doi: 10.1161/01.CIR.49.3.418

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Print ISSN: 0009-7322. Online ISSN: 1524-4539

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