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REVIEW ARTICLE

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Pectus excavatum: echocardiographic, pathophysiologic, and surgical insights

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Patients with pectus excavatum (PEX) may be referred for echocardiographic examination for a variety of complaints including exercise intolerance, dyspnea, palpitations, or chest pain. It is therefore important for the echocardiographer to have an appreciation of the various abnormalities associated with this disorder. Echocardiographic imaging may reveal a number of structural alterations of the right ventricle as well as a reduction in right ventricular systolic function. Interestingly, a number of these abnormalities have also been described in patients with arrhythmogenic right ventricular dysplasia, although patients with PEX do not share a predilection for malignant ventricular arrhythmias. Additional echocardiographic abnormalities associated with PEX include prolapse of the mitral and/or tricuspid valves, Marfan's aortopathy, pericardial effusion, prominence of the crista terminalis, and possibly a number of congenital cardiac anomalies. This review discusses the echocardiographic abnormalities associated with PEX and their pathophysiologic significance. The effects of corrective orthopedic surgery on cardiac function are also discussed.

KEYWORDS
arrhythmogenic right ventricular dysplasia, crista terminalis, echocardiography, Marfan's syndrome, mitral valve prolapse, pectinate muscles, pectus excavatum, right ventricular function

Pectus excavatum (PEX) accounts for >90% of all congenital chest wall abnormalities.[1] It is estimated to occur in approximately 1 of every 300 live births predominantly affecting white males.[2] Notably, PEX is rarely seen among nonwhites.[3] While the precise genetics of this disorder is not known, it has, nonetheless, been observed that about one-fourth of patients have an affected family member.[4] Finally, it is important to note that PEX is a feature of a number of connective tissue disorders, notable among them, Marfan's syndrome (MFS).[5]

Patients with PEX typically come to the attention of the cardiologist during adolescence coincident with skeletal maturity. The most common presenting complaints include exercise intolerance, dyspnea, chest pain, or palpitations.[6] Syncope secondary to inferior vena cava obstruction has also been reported but is rare.[7]

Pectus excavatum is characterized by posterior depression of the sternum and the adjacent costal cartilages. Its severity is quantified, by computed axial tomography or by chest x-ray, using the so-called *Haller index* (Fig. 1), a ratio of the width of the chest to the distance between the sternum and the spine. A ratio <2.5 is considered normal, and values >3.1 are generally regarded as severe.[8]

1 | ECHOCARDIOGRAPHIC FINDINGS

Posterior displacement of the sternum reduces the anteroposterior dimension of the chest causing compression and displacement of the heart (Fig. 1). The resulting distortion of normal cardiac geometry frequently makes transthoracic imaging quite challenging. In

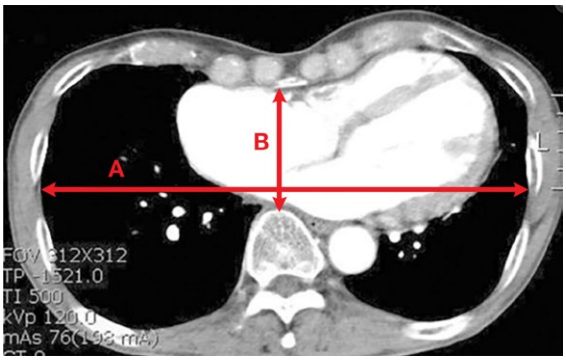

FIGURE 1 Computed axial tomographic image demonstrating how the Haller index is measured. The internal width of the thorax, labeled A, is divided by its midline anteroposterior dimension, labeled B. Values <2.5 are considered normal (Reproduced with permission from Park et al.,[30] page 64, Fig. 5)

some patients, adequate images may only be obtained from the subcostal window. These limitations are, however, largely overcome with transesophageal imaging.[9] Although seldom required for cardiac evaluation in patients with PEX, magnetic resonance imaging may prove particularly useful because, unlike echocardiographic imaging, it is unencumbered by abnormalities in body habitus. A comprehensive list of echocardiographic abnormalities found in patients with PEX is found in Table 1.

1.1 | Right ventricular abnormalities

Most of the echocardiographic abnormalities in patients with PEX involve the right ventricle (RV) likely a reflection of its location directly behind the sternal plate. Impingement by the sternum produces a characteristic reversal of the normal inward facing concavity of the RV (Fig. 2).[10] It has been suggested that confinement of the RV, between the sternum anteriorly and the spinal column posteriorly, limits its expansion during diastole.[10] In fact, the diastolic "dip and plateau" characteristically seen in RV pressure tracings of patients with constrictive pericardial disease has been reported with PEX[11]; however, a systematic study of right heart hemodynamics has yet to be undertaken. Apart from abnormalities of RV filling, patients with PEX may also demonstrate reduced systolic contractility which may be segmental or global.[12–15] Of note, the severity of RV systolic dysfunction bares no relationship to the Haller index.[16]

Whether abnormalities of RV filling and contractility are sufficient to account for the exercise intolerance observed in patients with PEX is not known. Notably, one magnetic resonance study found an average reduction of only 6 percentage points in RV ejection fraction compared to normal controls.[15] Notwithstanding the modest reduction in RV ejection fraction, it remains to be determined whether patients with PEX possess the requisite RV lusitropic and inotropic myocardial reserve necessary to sustain the marked rise in LV preload that normally accompanies exercise; failure to do so could potentially reduce exercise tolerance. Alternatively, it has been proposed that exercise intolerance in patients with PEX is the result of chronic deconditioning. This has been attributed to reluctance on the part of many patients, particularly adolescents, to participate in physically demanding activities because of poor body image and/or a perceived inability to do so.[17] Whether the improvement in exercise tolerance that is observed following corrective orthopedic surgery is related to enhanced body

TABLE 1 Echocardiographic abnormalities in patients with pectus excavatum

Right ventricle
Reversed curvature of the RV free wall
Global RV enlargement
RV outflow tract enlargement[a]
Rounding of the RV apex[a]
Sacculation of the RV free wall (microaneurysms)[a]
Regional or global RV systolic dysfunction[a]
Prominent RV trabeculations[a]
Moderator band hypertrophy
Regional RV wall thinning[a]
Heart valves
Mitral valve prolapse (functional and degenerative)
Tricuspid valve prolapse (functional and degenerative)
Congenital anomalies
Atrial septal defect (secundum-type)
Ventricular septal defect
Partial atrioventricular septal defects
Other
Aortic root enlargement
Pericardial effusion
Prominent crista terminalis
Inferior vena cava compression

[a]Echocardiographic findings shared in common with ARVD.

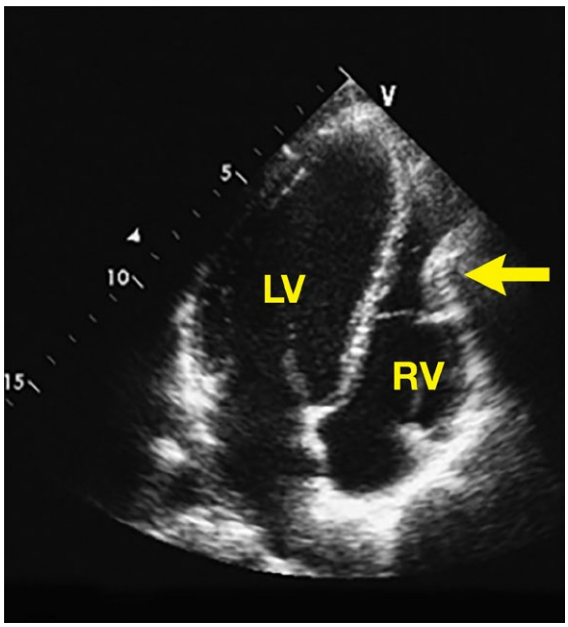

FIGURE 2 Apical 4-chamber echocardiographic image showing reversed curvature of the free wall of the right ventricle (RV) (arrow). LV = left ventricle (Reproduced with permission from Jaroszewski et al.,[10] page 191, Fig. 3)

image or to quantifiable improvements in RV function remains a matter of significant ongoing controversy.[3,6,18–20]

Other RV abnormalities that have been described in patients with PEX include global chamber dilatation or elongation.[12,13] Enlargement of the RV outflow tract, measured in the parasternal long-axis or in the aortic valve short-axis view, has also been described.[12,13] Rounding of the RV apex as well as moderator band hypertrophy (Fig. 3A) may also occur.[12,13] Additional abnormalities include small sacculations (microaneurysms) of the RV free wall (Fig. 3B), prominent trabeculae carneae (Fig. 3C), and regional wall thinning.[12,13]

Finally, it is interesting to note that a number of the RV abnormalities associated with PEX have also been described in patients with arrhythmogenic RV dysplasia (Table 1).[21–23] In fact, one or more of these abnormalities can be found in approximately one-third of patients.[13] Importantly, neither the characteristic 12-lead electrocardiographic abnormalities nor the propensity to ventricular arrhythmias seen with arrhythmogenic RV dysplasia have been described in patients with PEX.[13,21,23]

1.2 | Mitral and tricuspid valve prolapse

Prolapse of both the mitral[24] and tricuspid valves[13] has been described in patients with PEX. It has been suggested that, in some patients, leaflet prolapse is functional, representing the effects of annular distortion caused by compressive phenomena.[24] Accordingly, echocardiographic evidence of mitral and tricuspid valve prolapse is no longer evident in many patients following surgical correction.[6] It should, however, be noted that leaflet prolapse due to primary degenerative abnormalities[13,24–26] may also occur in patients with PEX, particularly those with Marfan's syndrome.[25]

1.3 | Marfan's syndrome

Marfan's syndrome is a rare, autosomal dominant disorder with skeletal and cardiovascular as well as ocular, cutaneous, and pulmonary manifestations. In fact, pectus excavatum is seen in approximately two-thirds of patients with Marfan's syndrome.[27] Interestingly, one echocardiographic study of pediatric age patients, presumed to have isolated PEX, revealed a significantly higher aortic root Z-score compared to normal controls.[5] It must, however, be emphasized that the presence of PEX and aortic root dilatation is considered insufficient to diagnose MFS when

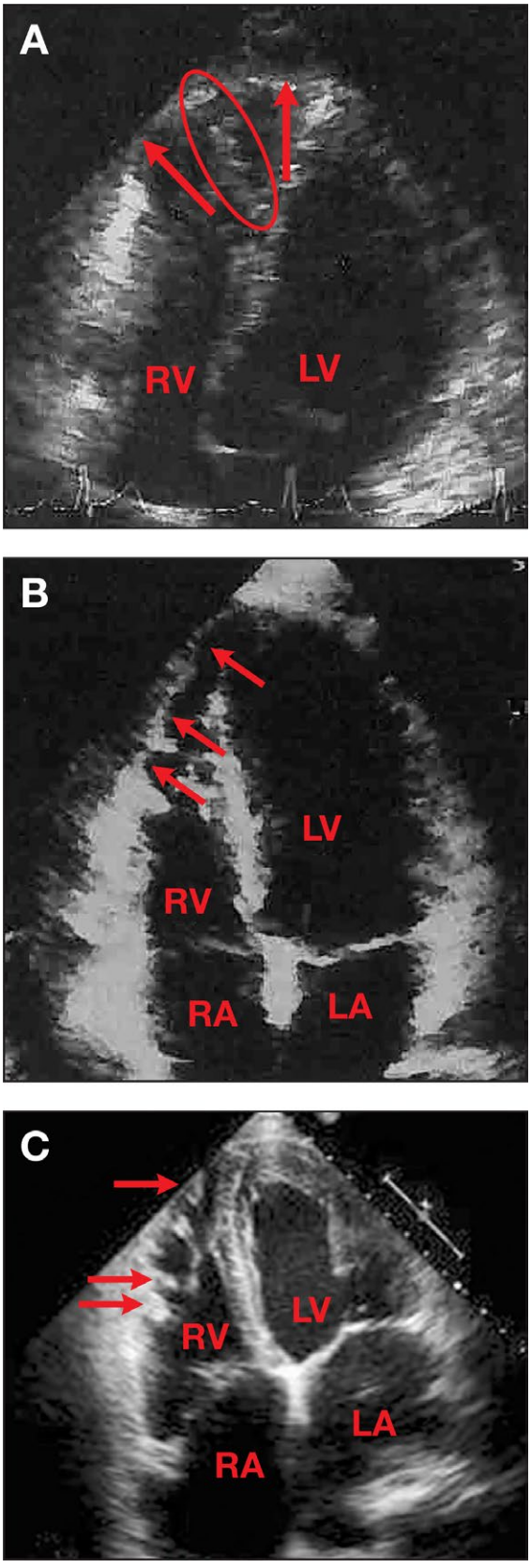

FIGURE 3 Echocardiographic abnormalities seen in patients with pectus excavatum. **A.** Note the rounding of the right ventricular (RV) apex (arrows). This region was akinetic. A hypertrophied moderator band, encircled in red, is also present. **B.** Image showing multiple sacculations (microaneurysms) of the free wall of the RV (arrows). (Panels A and B were reproduced with permission from Mocchegiani et al.,[12] pages 945–6, Fig. 1 and 2, respectively.) **C.** Prominent trabeculations are indicated by the arrows. These sometimes span the RV cavity and attach to the interventricular septum (not depicted). It is important to note that all of the foregoing abnormalities have also been described in patients with arrhythmogenic RV dysplasia. LA = left atrium; RA = right atrium; LV = left ventricle (Reproduced with permission from Park et al.,[30] page 63, Fig. 3)

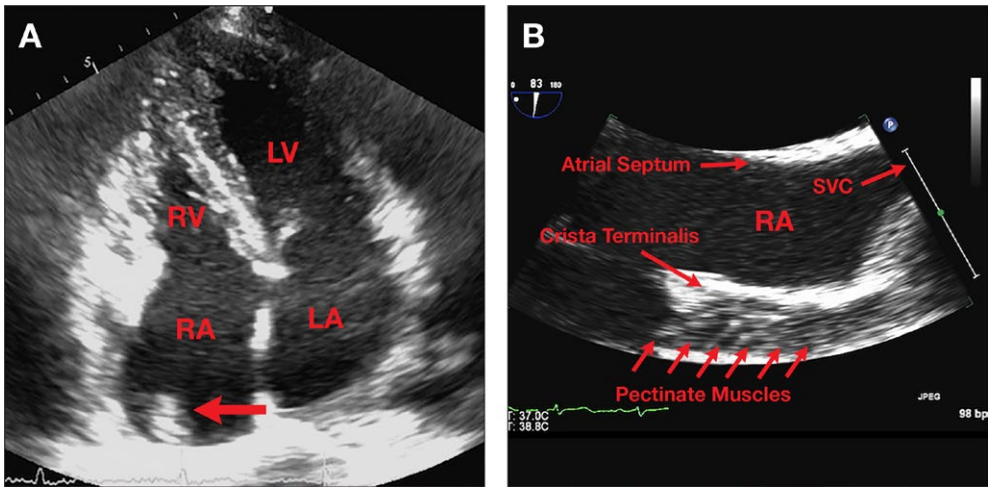

FIGURE 4 **A.** Modified apical four-chamber transthoracic echocardiographic image depicting a mass (arrow), measuring approximately 1.5 cm × 1.0 cm, attached to the roof of the right atrium (RA). LA = left atrium; LV = left ventricle; RV = right ventricle. **B.** Zoomed bicaval transesophageal image. Arrows indicate pectinate muscle attachments to the crista terminalis imparting a brush border appearance. SVC = superior vena cava

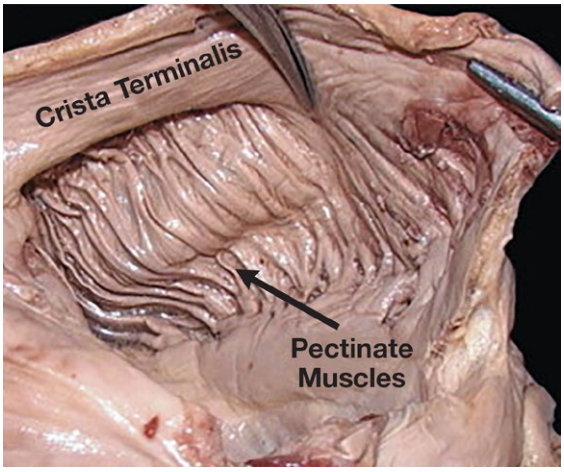

FIGURE 5 Gross specimen of a human RA showing the crista terminalis. Note the numerous connections to the pectinate muscles. LA = left atrium; LV = left ventricle; RV = right ventricle; SVC = superior vena cava. (Reproduced with permission from Loukas et al.,[29] page 83, Fig. 1)

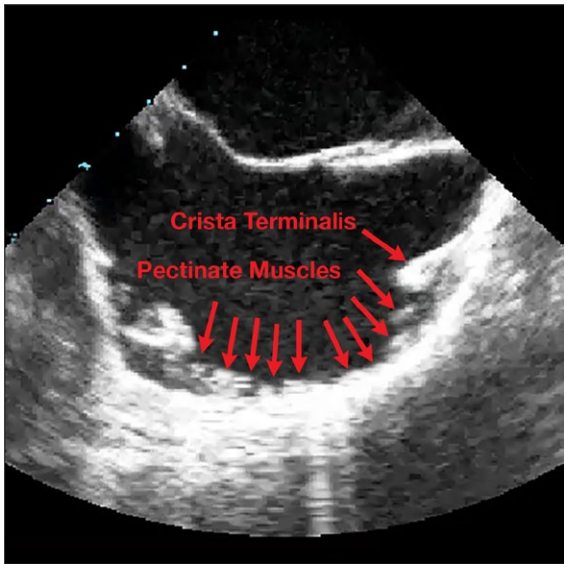

FIGURE 6 Bicaval transesophageal echocardiographic image of the RA depicting multiple pectinate muscles studding its free wall (see text for details)

the Ghent nosology criteria, currently used for the diagnosis if Marfan's syndrome, are applied.[28] This constellation of findings may, however, be regarded as a forme fruste of the disorder.[28]

1.4 | Pericardial effusion

One study found pericardial effusions in up to one-third of patients with PEX, although none were hemodynamically significant.[13] The cause of such effusions is not known. It has been suggested that they are the consequence of compressive phenomena which impede fluid resorption. Alternatively, they may represent the irritant effects of bony structures impinging upon the pericardium.[13]

1.5 | Prominent crista terminalis (CT)

Figure 4A is a transthoracic image from an otherwise-healthy 60-year-old male with PEX who was referred because of a questionable history of mitral valve prolapse. The study was notable for a prominent mass attached to the roof of the right atrium (RA) but was otherwise unremarkable. Due to the difficulties encountered during transthoracic imaging, the patient underwent a transesophageal examination. Imaging in the bicaval view (Fig. 4B) revealed numerous pectinate muscles attached to the CT imparting an unusual brush border appearance that represents the prominent RA mass seen with transthoracic imaging.

The CT is a fibromuscular ridge in the RA from which numerous pectinate muscles arise and then course anterolaterally toward the appendage (Fig. 5).[29] Pectinate muscle attachments to the CT are not ordinarily appreciated echocardiographically. Instead, the pectinate muscles are more often visualized unattached, studding the free wall of the RA as is shown in Fig. 6. We believe that the RA mass in this patient represents an instance in which otherwise "concealed" CT-pectinate attachments were brought into the echocardiographic imaging plane because of the geometric distortion of the RA that attends PEX. We, therefore, suggest that such RA "pseudomasses" be considered by the echocardiographer when imaging patients with PEX.

1.6 | Congenital anomalies of the heart

In one series, congenital abnormalities of the heart were reported in almost 20% of patients with PEX. While no one abnormality predominated, secundum-type atrial septal defect was most common. Ventricular septal defects and partial atrioventricular septal defects were seen less often.[12] Additional studies are needed to confirm these findings.

2 | CONCLUSION

Patients with PEX may be referred for echocardiographic studies for a variety of reasons. Notwithstanding the difficulties that are often encountered in imaging such patients, particular attention should be paid to RV size, function, and contour. The presence of a dilated root and mitral valve prolapse should prompt a consideration of Marfan's syndrome. Additional abnormalities which should be sought include a prominent the crista terminalis, small pericardial effusions, and possibly atrial septal defect. Finally, future research efforts will hopefully better characterize the RV hemodynamics associated with PEX and define the mechanism(s) responsible for the improvement in exercise tolerance that is observed following corrective orthopedic surgery.

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