

CLINICAL REVIEW

Diagnosis and Management of Pectus Excavatum in Primary Care

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Pectus is the most common congenital chest wall deformity, with pectus excavatum being the most prevalent type characterized by a sunken sternum. It has been proven that in severe cases this anomaly can result in cardiopulmonary compromise affecting patients' daily activity and ability to exercise. Despite the latest research, the deformity may still be viewed by some as a cosmetic issue that does not necessitate treatment. Since primary care physicians are often the first to evaluate these patients, a thorough understanding and comprehensive treatment approach for pectus is needed. Patients with pectus excavatum can present with a wide range of symptoms, although some may be entirely asymptomatic. Depending on the cardiopulmonary impact of the deformity, the severity of symptoms can range from mild exertional dyspnea to syncopal episodes, which may progress with aging. There have been several publications that have documented the improvement in quality of life and cardiopulmonary function that can occur in patients who have undergone repair of their pectus excavatum deformity. This article reviews updated information and recommendations and can serve as a guide to primary care physicians in the diagnosis, evaluation, and referral of pectus excavatum patients.

Keywords: Access to Care, Cardiology, Care Coordination, Chest Wall, Minimally Invasive Surgical Procedures, Pectus Excavatum, Primary Care Physicians, Primary Health Care, Quality of Life, Referral and Consultation

Background

Pectus excavatum (PE) is an anterior chest wall deformity that is likely to be encountered in the primary care setting. There has been a long-standing controversy over its clinical implications, with one side reporting a significant cardiopulmonary effect and the other believing it to be merely cosmetic.¹⁻³ Over the past decade, several publications have better defined the physiological significance of PE in addition to documenting progression of symptoms as the PE patient ages.⁴⁻⁷ The belief that PE is cosmetic may have led to patients with cardiopulmonary symptoms being overlooked. In a survey of 432 adult PE patients, 87% of women and 66% of men with PE felt that their symptoms had been dismissed by physicians when evaluated.^{8,9} Today, with extensive internet information available and a rise in patient awareness, many PE patients are seeking evaluation and treatment, even at an older age.^{8,10} Primary care physicians are often the first to evaluate PE patients, therefore an updated review is presented for evaluation and indications for surgical referral.

Epidemiology

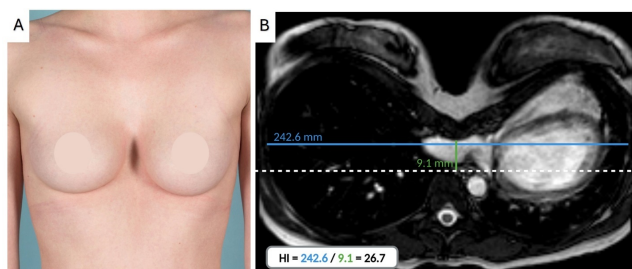
Pectus deformities are the most common congenital chest wall anomalies, with PE being the most common variant. In

early congenital publications, it was reported as more common in Caucasian males with a prevalence of 1 in 300-1000 live births.¹¹ Turkey, Argentina, Brazil, China, Korea, and other countries have also reported on large series of PE.¹²⁻¹⁶ While early reports indicated a 4-5x higher prevalence in males, recent literature has called this into question.⁸ A longitudinal study involving 2,687 adult patients who underwent radiographic evaluation, revealed a prevalence of pectus at 0.4% (1 in 250) and noted slightly more women than men (0.5% to 0.3%).¹⁷ It is likely that PE occurs equally in both sexes however, it may be harder to visually detect in females due to the presence of breast tissue (*Figure 1*).⁸

Presentation and Pathophysiology

It is important to prioritize the patient's symptoms over the visual appearance of the deformity, as even mild appearing deformities can cause underlying cardiac compression.⁴ The inward deviation can present symmetrically or asymmetrically (*Figure 2*), exhibiting varying degrees of costochondral and sternal concavity. Wider flat defects can appear visually less severe; however may have significant underlying cardiac compression (*Figure 3 A-F*).¹⁸ The Haller Index (HI) is a widely recognized radiologic measure for assessing the severity of PE and a value of 3.25 and greater

Figure 1. A 24-year-old female dancer presented with worsening exertional dyspnea, anxiety, and chest pain, noting a progressive decline in her ability to exercise. On physical exam, a severe pectus excavatum deformity is obscured by breast tissue (A). Although echocardiography appeared “within normal limits,” an Electrocardiogram revealed an incomplete right bundle branch block with secondary ST-segment changes. Due to persistent symptoms, a cardiac MRI was obtained, demonstrating a severe deformity (Haller Index (HI) 26.7) with compression of the right ventricle, leftward displacement of the heart, and altered right ventricular morphology (B).



is considered a severe deformity.¹⁹ The HI is a ratio which can be subject to variability due to differences in the shape of the chest and may not accurately reflect PE severity in patients with significant discrepancies of anterior-posterior to medial-lateral dimensions of the chest wall (Figure 4).¹⁸ The correction index (CI) can be a more accurate assessment of the severity of PE and is not variable among chest morphologies. It represents the percentage of chest depth to be corrected by bar placement (Figure 4).¹⁸

The cause of PE is unknown, but there are several hypothesis including unbalanced costochondral overgrowth and tethered diaphragm which cause the inward pull of the lower chest wall during development.¹¹ While PE is often an isolated anomaly, it is found to have familial inheritance in 30-40%.²⁰ The genetics of pectus have remained elusive and are likely a complex multi-factorial polygenic cause.²¹ Approximately 13-28% of patients with PE have associated skeletal disorders such as scoliosis.²² Several connective tissue diseases, including Marfan Familial Syndrome, Ehlers-Danlos, and Noonan syndromes can also be asso-

ciated with PE. Clinical findings such as hyper flexibility, elongated extremities, and other classic findings can be seen.²⁰ Genetic testing can be considered if suspicion is high.

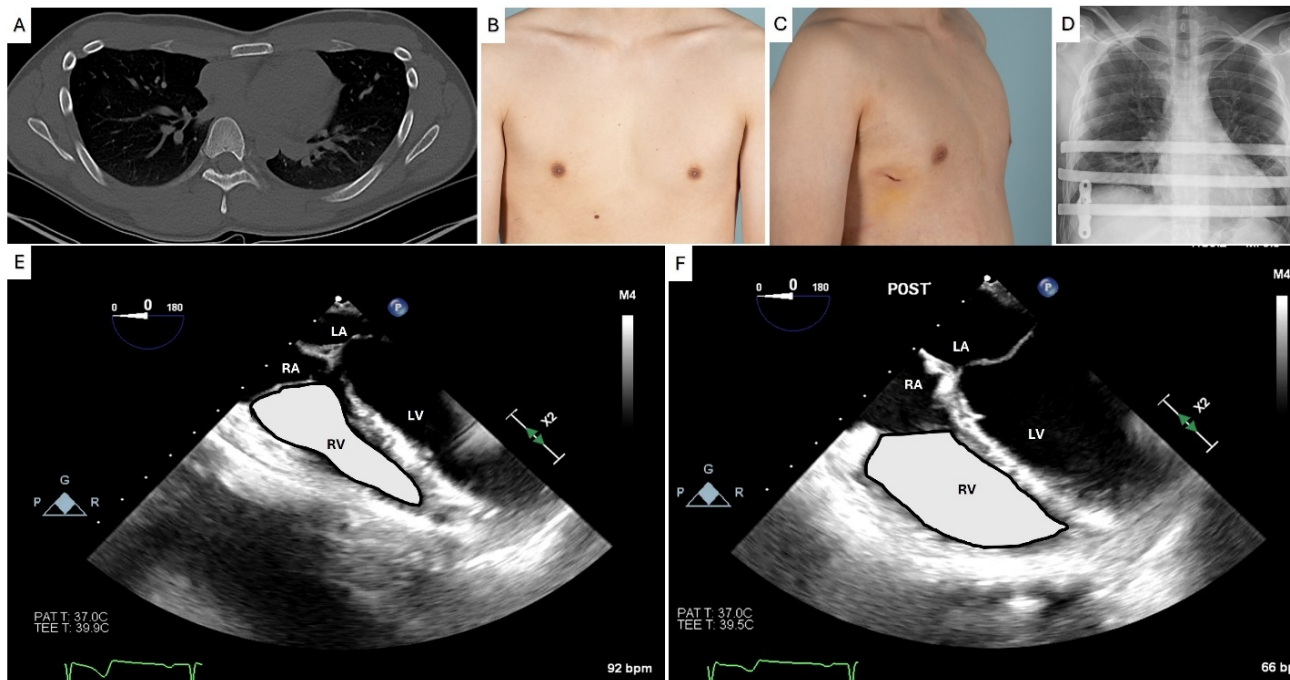
Adolescents may be highly vulnerable to psychosocial stress due to peer interactions and the effects of deformity can be detrimental.²³ Studies have indicated that older patients with PE also experience diminished social presence, reduced quality of life, increased depressive symptoms, and heightened social anxiety.²⁴ In a study of 432 adults (mean age of 30), 68% avoided activities in which their chest might be visible, and 65% reported feeling sad or depressed about their chest appearance at least some of the time.⁸

While the psychological ramifications of PE are well-documented, the physiological impacts of this deformity are less understood. The inward sternal concavity can compress the right heart to varying degrees depending on the location and depth of the deformity.⁷ When the right chambers are compressed or distorted, reduced cardiac output, stroke volume, ventricular strain, and overall reduction in systolic and diastolic function can be seen.^{1,7} To a lesser degree, sternal depression can reduce intra thoracic volume, affecting tidal volume and in severe cases, restrictive pulmonary function may be seen.^{1,3,25} While symptoms of PE can present at any age, many first report symptoms during adolescence, coinciding with the pubertal growth spurt.¹⁵ However, onset of symptom in the adult years can occur. One study noted 36% of adult patients reported new symptom onset after age 20 and over 53% progression after the age of 18; underscoring the variability in clinical presentation.^{8,10} Variable degrees of symptoms are described, with the most frequently reported being exercise intolerance, chest discomfort, and exertional dyspnea.^{11,26,27} There are several validated questionnaires available to assess symptoms including the original Nuss questionnaire (NQ), modified Nuss questionnaire (NQ-mA), and the Phoenix Comprehensive Assessment of Pectus Excavatum Symptoms (PCAPES) survey which takes into account neurological, cardiovascular, pulmonary, gastrointestinal, and psychosocial symptoms.^{8,28} An overview of the symptoms is provided in Table 1.

Figure 2. (A) MRI scan of a 20-year-old male with asymmetric pectus excavatum deformity with a Haller index >8, sternal torsion 38.1 degrees. (B) Corresponding pre-operative clinical photo showing anterior chest wall asymmetry and severe pectus excavatum deformity (C) post-operative clinical photo after pectus repair. (D) Post-operative chest roentgenogram after minimally invasive surgical repair with 3 bars and link bridges used for stabilization.

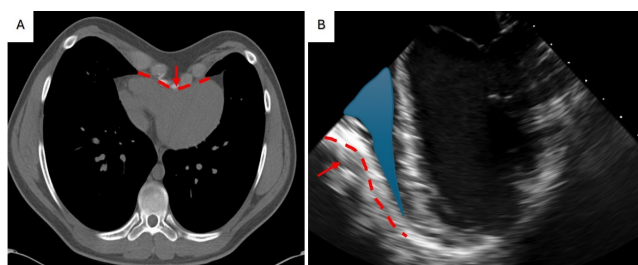


Figure 3. In wide, flat defects, pectus can appear less severe on physical exam despite underlying cardiac compression. (A) CT scan of a 27-year-old male patient with pectus excavatum Haller index of 3.6 and correction index of 12%. (B) Pre-operative image showing a relatively mild appearing deformity (C) post-operative image of the same patient. (D) Post-operative roentgenogram displaying three Nuss bars in place after minimally invasive surgical repair with a medial stabilizer on the lower bar. (E) Pre-operative transesophageal echocardiogram (TEE) of the same patient showing small right ventricular chamber volume due to the extrinsic compression by the pectus deformity. (F) Postoperative TEE showing resolution of right ventricle compression.



Abbreviations: RV: right ventricle, LV: left ventricle, RA: right atrium, LA: left atrium.

Figure 4. (A) Axial computer tomography (CT) demonstrating a Haller Index (HI) of 2.7 and a Corrective Index (COI) of 35.7% in a patient with pectus excavatum. Although the HI is below the commonly cited threshold of 3.25, this deformity was associated with clinically significant symptoms secondary to underlying cardiac compression (red dashed lines and arrow). (B) Corresponding echocardiographic views demonstrate the sternal deformity compressing the right ventricular and show a reduced chamber size (blue area).



Evaluation and Diagnosis

Even mild deformities can cause underlying cardiac compression (Figure 3).³¹ A thorough history, physical exam, and rule out of other medical causes for symptoms can de-

termine the need for referral to a pectus center or surgeon who specializes in pectus deformities. Many pectus centers offer specialty testing with protocols tailored to the pectus patient.²

Table 2 summarizes current imaging methods for assessing the PE deformity.

Some of the common testing includes:

Chest X-ray

While CT scan is more accurate and recommended for surgical assessment of pectus cases, plain film radiographs can be used for an initial evaluation and estimation of both the HI and CI as well as assessment of cardiac displacement (Figure 5).³²

Computed Tomography Scan

Non-contrast, low dose radiation CT scans are the gold standard for assessing severity of PE and an expiratory view is recommended.³³ Significant differences can be found when comparing end-expiration to end-inspiration, with a large influence on calculation of pectus indices and cardiac compression.³² Information including asymmetry and sternal torsion, cardiac compression and heart displacement, calcification of cartilage, and pulmonary abnormalities can be additionally assessed (Figure 6).³²

Table 1. Symptoms of Pectus Excavatum Noted by Patients.

	Symptoms	Percentage Reported in the Literature
Psychological Symptoms	Avoidance of activities that expose the chest	68% ⁸
	Sad or depressed about the way their chest looks	31% ⁹ - 65% ⁸
Cardiopulmonary Symptoms	Chest pain or pressure	88% ⁸
	Tachycardia	76% ⁸ - 94% ²⁹
	Palpitations	25% ²⁷ - 72% ^{4,8}
	Exercise intolerance	62% ³ - 83% ⁸
	Difficulty keeping up with peers	67% - 86% ^{4,8}
	Dyspnea	64% ³ - 96% ^{4,8}
Neurological Symptoms	Headaches	74% ⁸
	Positional Dizziness	67% ⁸
	Exertional Dizziness	59% - 61% ⁸
Gastrointestinal Symptoms	Dysphagia	2% ³⁰ - 48% ⁸
	Post-prandial dyspnea	52% ⁸
	Early Satiety	84% ⁸

Table 2. Advantages and Disadvantages of the Imaging Techniques Assessing Pectus Excavatum.³²

Modality	Advantages	Disadvantages
Chest X-ray	Can be used for diagnosis and calculating pectus indices	Less accurate
	Lower cost and less exposure to radiation	Cannot assess asymmetric chest defects and sternal torsion
Computed Tomography (CT) Scan Low dose radiation imaging without contrast should be performed	Excellent for diagnosing pectus excavatum, calculating indices, assessing cardiac compression, and evaluating calcification of the cartilage	Higher cost and exposure to ionizing radiation
	Can assess asymmetric chest defects and sternal torsion	
Magnetic Resonance Imaging (MRI) (Chest/ Cardiac)	Excellent for diagnosing pectus excavatum, calculating pectus indices, and assessing cardiac compression	Higher cost and less commonly available due to insurance restrictions
	Cardiac MRI can be used to evaluate cardiac function as well as the degree of compressions	Requires sedation or other interventions to perform on claustrophobic individuals

The CI, defined as the percentage of chest that is in an abnormal position or depressed, may be a superior assessment of severity.¹⁸ This is particularly true in broad-chested patients where the maximal A/P diameter is disproportionately wide, leading to a low HI despite a severe deformity (*Figure 4*).

Magnetic Resonance Imaging (MRI)/ Cardiac MRI

Chest MRI serves as an alternative imaging option, without exposure to radiation. A cardiac MRI can be performed with the added benefit of evaluating cardiovascular dynamic function through real-time imaging.³² Conventional methods assess wall motion abnormalities and ejection fraction, however ventricular myocardial strain imaging has been shown to be more sensitive in detecting right ventricular dysfunction due to sternal depression and subtle septal mo-

tion abnormalities in PE patients during both inspiration and expiration.⁷

Electrocardiogram (ECG)

Although most patients will have a normal 12 lead ECG, several abnormalities can be seen. These include an increased P wave amplitude in lead II, dominant negative P wave morphology in lead V1, rsr' pattern in lead V1, right bundle branch block (RBBB), T wave inversion in lead V1-V3 and right axis deviation.³⁴ In a report on 310 adult PE patients, it was found that RBBB was present in 9.4% of patients, an rsr' pattern in 40.6%, and T wave inversions lead V1-V3 in 62.3% of patients. Adolescents with PE have also been noted to have higher P-wave voltage in lead II ("Eiffel tower" P waves) with a prolonged PR interval and high prevalence of RBBB.³⁵ One publication found that RBBB and T wave inversion were associated with a lower exercise capacity in PE evaluated with a cardiopulmonary

Figure 5. (A) Lateral X-ray highlighting the sternal concavity (orange arrow and dotted line) showing the minimum A/P distance between the anterior vertebral body and the posterior sternal body (green line) needed to calculate the Haller Index. (B) Anteroposterior x-ray view showing the maximum transverse thoracic diameter (blue line).

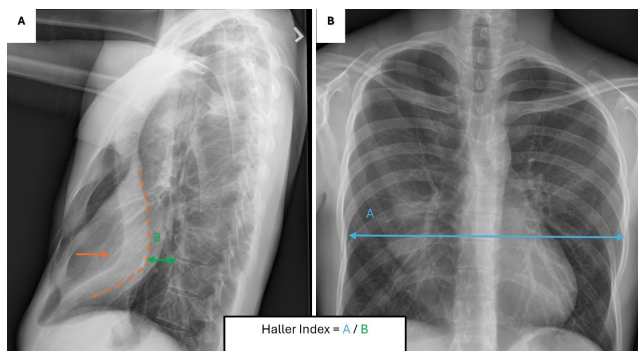
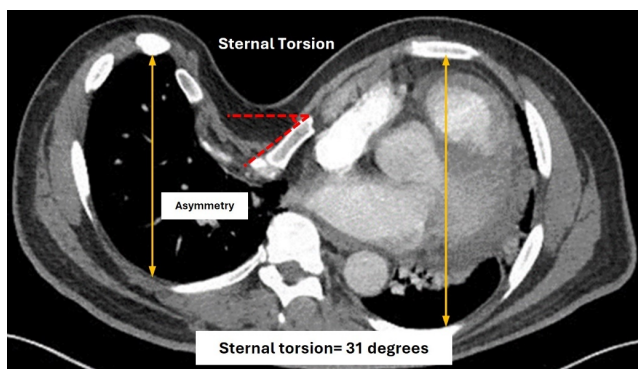
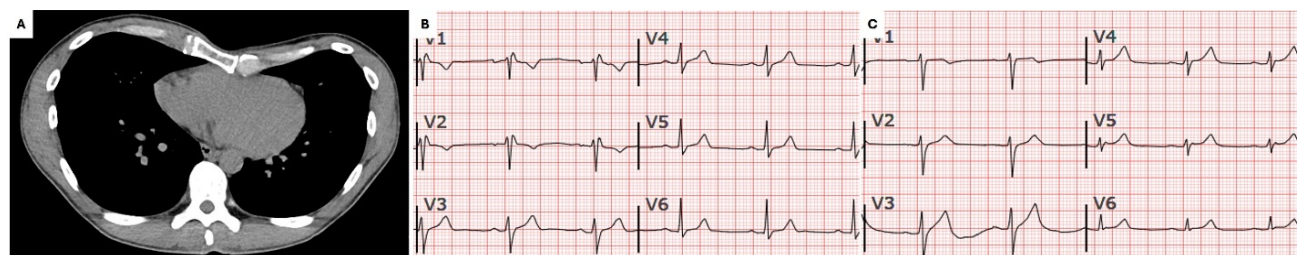


Figure 6. Computer tomography (CT) scan of a patient with severe pectus excavatum, Haller Index of 18 and correction index of 44% illustrating right asymmetry with the left side of the chest appearing incongruently larger than the right (orange lines) with sternal torsion of 31 degrees (red acute angle).



exercise test (CPET).³⁵ Once PE correction was done, these ECG changes were found to normalize in many patients (Figure 7).³⁴

Figure 7. Effect of cardiac compression on electrocardiogram (ECG) findings. (A) Pre-operative CT scan of a pectus excavatum patient showing cardiac compression. (B) Preoperative ECG of the same patient showing normal sinus rhythm, ventricular conduction revealing incomplete right bundle branch block (RBBB). (C) Post-operative ECG showing resolved ECG changes in V1-V2 and V5-V6.



Echocardiogram

Given the association with connective tissue disorders, transthoracic echocardiogram (TTE) is important for structural information such as aortic dimensions and valve function.² Due to the deformity and distortion of the heart, it can be difficult to get traditional acoustic windows and obtain complete visualization of the right heart chambers.³³ A “normal” TTE does not preclude the presence of compression.⁷ In addition to the lateral decubitus position, hemodynamic changes may be identified when patients move from a supine to a sitting or leaning forward position due to an increase in compression on the right ventricular outflow tract.⁷ An intraoperative transesophageal echocardiogram monitoring surgical correction can be useful to document preoperative cardiac compression and confirm complete postoperative resolution which is key to successful repair.⁶

Cardiopulmonary Exercise Testing

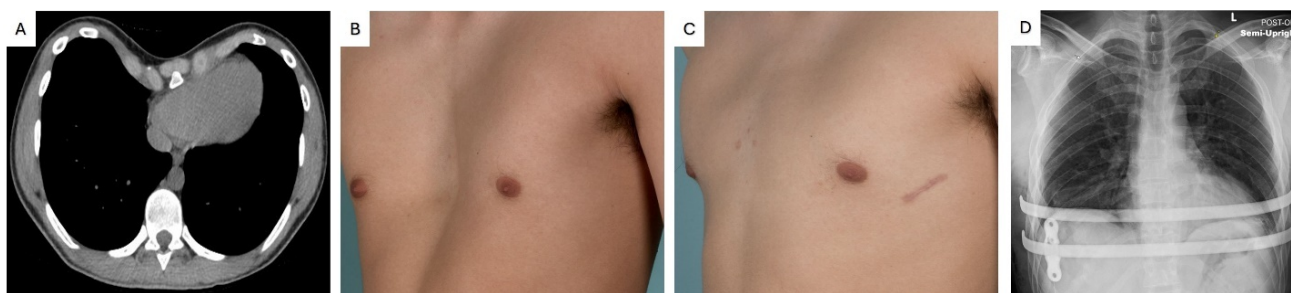
CPET is useful to assess patients with exercise intolerance and can quantify exercise capacity and oxygen consumption. A reduced peak maximal oxygen consumption (VO_2) and oxygen pulse can indicate cardiac impairment from the depression.⁴ Several studies have confirmed the improvement of key CPET parameters following surgical correction of PE, but it is still a non-specific tool and the level of exercise a patient is performing can significantly affect the test.^{1,4} A normal VO_2 , especially in an athletic patient, does not negate surgical intervention.⁴

Indications for Surgery

There is no established set of indications for surgical intervention, however, most insurance providers require patients meet two or more of the following criteria for consideration of medical coverage²:

1. HI greater than 3.25 or a CI greater than 20%¹⁸
2. Cardiopulmonary symptoms²
3. Evidence of cardiac compression or restrictive lung disease^{2,36}
4. Psychosocial issues secondary to the deformity^{23,24}
5. Recurrence of PE following previous surgical repair³⁷

Figure 8. Pre-operative CT of a patient with severe pectus excavatum (Haller Index = 3.85) (A) and clinical photograph of patient preoperative and (B), (C) postoperative after minimally invasive repair of pectus excavatum (MIRPE). (D) Post-operative chest roentgenogram showing two Nuss bars after pectus repair with a medial stabilizer on the lower bar.



The optimal age for surgical treatment is debated. In some countries such as South Korea and China, repair of children as young as 3 years old is performed.³⁸ In the United States, Canada and Western Europe, the mean age for PE repair has increased over the years, and most centers recommend surgery be deferred until puberty and early teenage years, when patients are closer to skeletal maturity.³⁹ Although repair is technically “easier” in younger patients because of their more malleable chest wall, an older age for pectus repair is now recommended due to the high percentage of very young patients that experienced regression after implant removal.^{38,40} Additionally, placing multiple bars for chest wall support and leaving implants in longer have been advocated to decrease the risks of regression.⁴⁰ The question remains as to “how old is too old for PE repair?”. More centers are repairing adult patients well into their 30-40’s utilizing minimally invasive surgical techniques.⁴¹ The rigidity and weight of the adult chest wall increases the difficulty and complications of repair.⁴²

Older patients and those with more complex deformities require highly experienced surgeons. It is estimated that the learning curve for operating on older and more complicated patients is double that of adolescents.⁴³ The definition of what qualifies as an experienced surgeon varies. For adolescent patients, a learning curve of 25 cases has been reported to yield average outcomes; however, for excellent outcomes with a complication rate of <10%, at least 59 cases need to be performed by one surgeon.^{44,45} Centers performing around 70 cases annually report lower complication rates and shorter hospital stays.⁴³

Surgical Repair

A variety of surgical techniques have been used over the years for repair of PE however the two most common methods are the open Ravitch repair and the MIRPE (or “Nuss”).^{2,40}

The open Ravitch repair involves resection of the deformed costal cartilage of the defect and performing an osteotomy at the sternum. Metal struts, mesh, and plating may be used to support the repair.²

The MIRPE procedure is the standard of care for pediatric and adolescent patients and increasingly used for re-

pair of adult PE.^{2,26} MIRPE has advantages when compared to the open Ravitch procedure, including less bleeding and shorter hospital stay.⁴⁴

Under thoracoscopic visualization, curved bars are inserted through lateral incision between the ribs and behind the sternum. The bars, like braces, are removed after 2-4 years.³⁶ Since the introduction of the original MIRPE procedure (*Figure 8*), technique modifications have been implemented which have reduced the risk of complications.^{2,36} Recurrence rates as low as 2% are reported when performed at experienced centers.^{25,37,46}

Pain has been a major postoperative concern following PE repair, with various strategies such as intravenous opioids, epidurals, subcutaneous catheters, and local blocks being employed to reduce postoperative pain.⁴⁷ More recently, intercostal nerve cryoablation has become the recommended method of postoperative pain control by many large volume pectus centers.^{48,49} The modality allows for freezing of the chest wall nerves that is temporary with return of nerve function within 4-6 weeks. Superiority has been reported regarding lower narcotic use and shorter hospital stay with the use of intercostal cryoablation.^{47,49}

Non-Surgical Options

The vacuum bell is a suction cup which is positioned over the PE deformity and connected to a hand pump so the patient controls pressure to reduce the defect. Daily use for a prescribed amount of time has been shown to reduce the severity of PE defects. The device is most effective in patients younger than 11 years old, with some avoiding surgical correction through its use. It was reported that 44% of patients had complete correction of their pectus deformity after consistent use of the device (mean of 21.8 months). Potential adverse effects include sternal pain, skin irritation, and hematoma formation. Other noninvasive recommendations for associated issues such as chest pain and back pain include physical therapy, the use of a posture brace, and regulated exercise.⁵⁰

Conclusion

Primary care physicians are often the first to encounter and diagnose pectus cases and may encounter them in the postoperative period while implants are still in place. Pectus excavatum can have physiological and psychological implications potentially affecting quality of life. Diagnosis with surgical referral of appropriate patients may facilitate timely and effective intervention. Maintaining a high index of suspicion is recommended, particularly for patients with visually mild deformities or in female patients.

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Conflicts of Interest

Dawn Jaroszewski discloses consulting and IP/royalties through Mayo Clinic Ventures with Zimmer Biomet, Inc. and Atricure Inc. Other authors report no conflict of interest.

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