Pectus Excavatum: A Review of Diagnosis and Current Treatment Options

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Osteopathic medicine places a special emphasis on the musculoskeletal system, and understanding how chest wall structure may influence function is critical. Pectus excavatum is a common congenital chest wall defect in which the sternum is depressed posteriorly. Patients may present with complaints of chest wall discomfort, exercise intolerance, and tachycardia. The medical implications, diagnosis, and treatment options for patients with pectus excavatum are reviewed.

Musculoskeletal disorders are commonly encountered by osteopathic physicians in primary care. Many patients with chest wall malformations may present with complaints of positional and exertional chest pain, exercise intolerance, and tachycardia. Understanding how chest wall structure may influence function is critical. Pectus deformities, including excavatum and carinatum, are among the most common congenital chest anomalies, with an overall incidence of pectus excavatum (PE) in 23 of 10,000 births. Pectus excavatum is more likely to occur in men than in women; however, female adolescents and women may be underdiagnosed because breast tissue could disguise the defect.

In patients with PE, the anterior chest wall is depressed into the thoracic cavity. Depending on the severity, PE can manifest with minor symptoms and cosmetic issues or with disabling cardiopulmonary symptoms. The inward deformation of the sternum may cause right-sided heart compression and restrictive pulmonary deficits. With the increase in awareness of pectus deformities and the information available, more patients with uncorrected PE are seeking evaluation and treatment.

The cause of PE may stem from unbalanced overgrowth in the costochondral regions. Studies have shown that patients with asymmetric PE had shorter ribs on the more severely depressed side of the defect. Up to 43% of patients with PE have a family history of the condition. Pectus excavatum is thought to be of multifactorial inheritance, but the exact genes implicated are unknown. Pectus excavatum can be associated with scoliosis and connective tissue disorders such as Marfan syndrome, Ehlers-Danlos syndrome, and Noonan syndrome. One study found scoliosis incidence as high as 21% in patients with PE.
Patient Presentation and Evaluation

When a patient presents with PE, an evaluation should be completed to understand the severity of the condition and determine treatment options. A thorough workup is indicated in patients with symptoms and can include history and physical examination; thoracic imaging (eg, inspiratory and expiratory computed tomography, magnetic resonance imaging, electrocardiography, echocardiography); blood chemistry analysis (eg, basic metabolic profile, complete blood cell count, lactate dehydrogenase [LDH] level); cardiopulmonary exercise testing through measurement of maximum oxygen consumption ($V_{O_2}$); and pulmonary function tests.

History and Physical Examination

Although PE can be detected at birth or in early childhood, many patients may not present until early adolescence.\textsuperscript{4,14} The malformation can be symmetric or asymmetric, with asymmetry associated with rotation of the sternum to the more depressed side.\textsuperscript{15} Because patients with PE may exhibit a marfanoid habitus,\textsuperscript{13} assessment of Marfan syndrome’s clinical features should be performed. Patients may have phenotypic findings consistent with Marfan syndrome but not meet all diagnostic criteria.\textsuperscript{16} Referral to a genetic specialist may be indicated for definitive assessment. If scoliosis is present, it should be assessed.\textsuperscript{13} Poor posture can also be associated with severe PE.\textsuperscript{13}

The cardiopulmonary effects of PE cannot be accurately assessed by measurement of the depth of the defect alone.\textsuperscript{4} Wide, flat PE may cause cardiac compression despite the mild appearance. Cardiac auscultation may reveal a murmur, and imaging of the chest wall can detect a leftward shift of the heart. This shift can be caused by distortion and compression of the heart or valve abnormalities.\textsuperscript{17,18}

Patients may exhibit a variety of symptoms (Figure 1), which are often described as progressing with age. Many patients may not have symptoms until adolescence or adulthood.\textsuperscript{4,14} Adult patients may also have notable progression of symptoms with aging as the chest wall becomes less compliant and as compensatory mechanisms decrease.\textsuperscript{14,19} Psychosocial issues and body image are factors that cannot be underestimated. For example, adolescent patients can be susceptible to psychological stress and trauma from peer scrutiny.\textsuperscript{20} In our practice experience, we had an adolescent patient who sustained a near-fatal, self-inflicted shotgun wound to the chest after being bullied by his classmates because of his appearance in having PE. Surgical correction of PE may improve symptoms in these patients.\textsuperscript{21}

Thoracic Imaging

Initial evaluation should include imaging, particularly noncontrast computed tomography or magnetic resonance imaging.\textsuperscript{12,21} These imaging modalities allow visualization of the malformation and can reveal cardiac compression, cardiac displacement, and the presence of atelectasis or tracheobronchial compression.\textsuperscript{22} Computed tomographic images should be obtained on both inspiration and expiration because the severity of the defect may substantially worsen when

<table>
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<th>Symptom</th>
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<td>Dyspnea with exercise</td>
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<td>Loss of endurance</td>
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<td>Chest pain</td>
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<td>Palpitations</td>
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<td>Exercise-induced wheezing</td>
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<td>Upper respiratory tract infections or asthma</td>
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<td>Fainting or dizziness</td>
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<td>Exercise intolerance</td>
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<td>Early postprandial satiety</td>
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<td>Inability to keep up with peers</td>
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<td>Anxiety and body image disturbance</td>
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Figure 1.
Commonly reported symptoms of pectus excavatum.
in which \( a \) is the minimum distance between the anterior spine and the posterior surface of the sternum, and \( b \) is the maximum distance between the anterior spine and most anterior internal rib. It yields a percentage that the chest would need to be corrected to achieve normal dimensions, with a normal level being 10% or less. The correction index better assesses patients with a more barrel-shaped chest who have a Haller index score that is falsely low.

### Electrocardiography

A 12-lead electrocardiogram should be ordered for all patients. Abnormalities in electrocardiogram results, most commonly right bundle branch block and signs of atrial and ventricular hypertrophy, are frequently seen in patients with PE. Sinus arrhythmias and premature ventricular beats have also been reported.

### Echocardiography

Although transthoracic imaging may be limited in patients with severe anomalies, echocardiography should be performed to rule out substantial anatomic abnormalities. This imaging is critical for patients who are suspected of having Marfan syndrome to assess aortic dimensions and mitral valve function. Anatomical abnormalities in the mitral valve leaflets and right ventricular morphologic changes, including a rounded apex, trabecular hypertrophy, structural changes in the moderator band, and sacculations of the ventricular wall, have also been reported. Transesophageal echocardiogram can better assess the right side of the heart in patients with PE but is invasive. Right-sided heart compression can be notable, with associated diastolic dysfunction and decreased outflow. Additionally, the right atrium and outflow tract can be compressed, reducing atrial filling and consequently decreasing the ejection fraction. The outflow of the right ventricle can be notably increased after surgical correction of the PE defect and can result in an increase in both right and left ventricular output.
Blood Chemistry Analysis
One analyte has been found to be consistently abnormal in patients with PE: LDH. In patients with PE, LDH can be elevated, but it decreases after correction. This elevation may be due to internal organ compression; however, because LDH is present in almost all body tissues, it is a nonspecific marker.

Cardiopulmonary Exercise Testing
To quantify the degree to which PE is affecting a patient’s ability to exercise and consume oxygen, cardiopulmonary exercise testing should be performed. Cardiac limitations resulting from the malformation can be demonstrated by an abnormally low maximum anaerobic $V_{O_2}$ level during exercise testing. A study found that during exercise, patients with PE were unable to reach the stroke volumes of control patients at any intensity of exercise. Accordingly, cardiac output was decreased, which led to a limitation in peak exercise capacity. Exercise ability and maximum $V_{O_2}$ have been shown in some studies to be statistically significantly improved after repair of PE.

Pulmonary Function
In general, pulmonary function is not substantially abnormal in most patients with PE. In severe cases, compression of the sternum and decreased thoracic volume may cause a decrease in forced vital capacity, tidal volume, and mixed $V_{O_2}$ saturation. Additionally, a study found that a majority of patients had a normal forced expiratory volume in 1 second/forced vital capacity ratio, suggesting that PE causes restrictive, but not obstructive, lung disease.

Management of PE
Pain is the presenting complaint in many patients with PE. The use of anti-inflammatory medications, osteopathic manipulative treatment, acupuncture, and physical therapy is recommended for pain management. A clinically significant improvement in pain and quality of life may be achieved among patients receiving acupuncture and osteopathic manipulative treatment for musculoskeletal pain.

Selected younger patients with PE can undergo conservative treatment with a vacuum bell, which is now approved in the United States. This device has a suction cup that the patient applies to the chest for a minimum of 1 hour daily using a hand pump to provide suction. Patients in other countries have reported correction of the malformation and elevation of the chest with the vacuum bell; 10% of these patients selected surgical repair after using the device.

Surgical procedures can offer benefits to patients with severe anomalies, including improved cardiopulmonary function, higher self-esteem, and resolution of many symptoms (eg, dyspnea, fatigue, palpitations, chest pain, increase in exercise tolerance).

Indications for Surgical Intervention
Surgical procedures are recommended for patients with severe PE as noted by a Haller Index score of more than 3.25 or a correction index score of more than 20%. Evidence of cardiopulmonary disability includes decreased peak anaerobic $V_{O_2}$, restrictive pulmonary disease, rightsided cardiac compression on echocardiogram or thoracic images, and a shift of the heart into the left side of the thorax. Some insurance companies require medical evidence of cardiopulmonary disability for coverage.

In general, the age recommendation for surgical correction is the mid-adolescent years. Repair during this stage allows for patients to complete their growth and decreases the risk of recurrence. Pectus repair in adults can also be successful.

In the past, the Ravitch procedure was advocated for managing PE. This procedure involved various degrees of removal of the costal cartilage through an anterior incision (Figure 3A). Anterior support with a metal strut or mesh was also performed by some surgeons. This intervention at too young of an age (6 years or
Some surgeons still advocate the Ravitch procedure for older adults and asymmetric anomalies. However, in children and adolescents, the Ravitch procedure has generally been replaced with minimally invasive procedures such as the Nuss procedure.

In the 1990s, the Nuss technique for PE repair was developed using an intrathoracic bar placed behind the sternum to elevate and support the depression. The bar is secured with sutures around the ribs or horizontal stabilizers and removed in 2 to 3 years. An outpatient procedure is performed to remove the bar from the side pocket incisions. Initial criticisms of the Nuss procedure were made by Ravitch proponents owing to the potential for cardiac injury. However, modifications, including use of video thoracoscopy and sternal elevation to allow passage of the bars under direct visualization, have substantially decreased surgical risks.

Bar rotation or malpositioning continues to be a potential postoperative complication of the Nuss procedure. In an experienced surgeon’s hands, this risk should be less than 5%. In general, a short hospitalization of 3 to 5 days with recovery to normal activity by 4 to 6 weeks is expected in most patients. Minimally invasive repair in patients aged 50 years or older is also feasible with experienced pectus surgeons. Several large series have reported successful correction using the Nuss procedure in patients aged 30 to 70 years.

Pectus excavatum can recur after any type of surgical repair, and the risk varies based on the patient’s age, type and severity of PE, and the presence of underlying connective tissue disorders. Removal of the support bars 2 to 3 years after the Nuss procedure has decreased recurrence rates to 2% to 5%. Quality of life and self-esteem have been shown to improve with surgical repair. When surveyed after either the Ravitch procedure or Nuss procedure, patients exhibited an increase in health-related quality of life, with minimal differences in patient satisfaction between the 2 surgical procedures.
Conclusion

Pectus excavatum is a common chest wall malformation that is gaining increasing awareness. Osteopathic physicians must understand how the chest wall structure influences patients’ symptoms and function. Medical implications to the musculoskeletal and cardiopulmonary symptoms can be severe in some cases. For milder cases, osteopathic manipulative treatment techniques can offer clinically significant improvement in chest wall discomfort.15

Author Contributions

Dr Jaroszewski provided substantial contributions to conception and design, acquisition of data, or analysis and interpretation of data; all authors drafted the article or revised it critically for important intellectual content; Dr Jaroszewski gave final approval of the version of the article to be published; and Dr Jaroszewski agrees to be accountable for all aspects of the work in ensuring that questions related to the accuracy or integrity of any part of the work are appropriately investigated and resolved.

References


Figure 4.
The Nuss procedure for pectus excavatum involves placement of stainless steel bars as braces behind the sternum (A) to elevate and support the depression. These bars are then removed in 2 to 3 years. Complications with bar rotation and intrathoracic positioning can occur as seen in the chest radiograph (B) and computed tomographic image (C). Image A used with permission of Mayo Foundation for Medical Education and Research, all rights reserved.


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