CLINICAL VIGNETTE

A 19-Year-Old Woman with Pectus Excavatum

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Case Presentation

A 19-year-old girl presented for an evaluation of a "sunken chest" present since childhood. Past medical history includes anxiety, depression, and scoliosis. Exam includes BP of 118/83, heart rate of 98, oxygen saturation of 97% on room air and BMI of 24.9. Her exam was normal except for the sunken chest deformity. She was previously told that she had pectus excavatum. She noted symptoms of shortness of breath on exertion that have worsened over the last 3 years with decreased exercise tolerance, sharp substernal chest pains and palpitations with mild exertion. A transthoracic echocardiogram showed normal left ventricular systolic function with mildly depressed right ventricular systolic function. Chest CT confirmed pectus excavatum with a Haller index o 3.6. Pulmonary function tests showed a mild restrictive ventilatory defect. The patient was referred to cardiothoracic surgery to discuss corrective procedures. Given her symptoms she underwent minimally invasive Nuss procedure. She had an uneventful surgery and post op course and reported decreased symptoms as she recovered. Two years later the bars placed during surgery were removed.

Discussion

Pectus excavatum (PE) also known as "funnel or sunken chest," is the most common anterior chest wall deformity. It is characterized by sternal depression or invagination of the lower sternum and adjacent costal cartilages. While often asymptomatic, it can lead to physical and psychological sequelae, necessitating appropriate diagnosis and management.

The incidence of PE is 1 in every 400 to 1000 live births and is three to five times more prevalent in males than females.[1] There is no clear etiology of PE and is generally sporadic. Several theories range from abnormal cartilage remodeling to abnormal stress on the sternum and costal cartilages. It has also been associated with connective tissue disorders including Marfan syndrome, Ehlers-Danlos syndrome and osteogenesis imperfecta.[2] It has also been reported with neuromuscular diseases and genetic conditions. These include Noonan syndrome, Turner syndrome, neurofibromatosis type I and multiple endocrine neoplasia type 2b.[3]

Finding may be present at birth and become more noticeable during periods of rapid growth, typically during adolescence. Clinical features include cosmetic concerns predominantly among female patients to chest pain, decreased exercise tolerance and shortness of breath.[4] Clinical significance and referral for surgical correction depends on the severity of chest wall defect, cardiopulmonary morbidity and psychosocial concerns. Patients with moderate to severe symptomatic PE are usually evaluated with computed tomography (CT) scans to quantify severity, and pulmonary function testing to assess for restrictive disease and possibly exercise testing to evaluate for any cardiopulmonary limitations. Patients may also have associated scoliosis and may be tachycardic due to reduced stroke volume from distortion and displacement of the heart in the thoracic cavity.[5] A pectus severity index (PSI), also known as the Haller index describes the depth of the pectus defect, comparing the ratio of the lateral diameter of the chest to the sternum-to-spine distance at the point of maximal depression is calculated from the CT to determine the severity of the defect. A normal chest has a PSI of ≤ 2.5. Typical candidates for surgery usually have PSI of > 3.25.[6] Cardiac evaluation with echocardiogram in patients with severe PE may demonstrate right ventricular outflow obstruction and reduced right ventricular systolic function.[7] Corrective surgery is considered in patients with two or more of the following: PSI > 3.25, cardiac abnormalities (including compression, displacement, mitral valve prolapse or conduction abnormalities), restrictive respiratory disease or failed previous repair of PE.[8]

The Nuss procedure is the most common repair. It is considered "minimally invasive" reconstruction and is the most common PE repair. It is now preferred over traditional open repair techniques. The procedure involves placing a custom-contoured steel bar ("Nuss bar") behind the sternum attached laterally to the outer edge of the rib cage. The bar places outward pressure on the pectus deformity and typically removed after approximately two years. Patients report subjective improvement in exercise tolerance with subtle improvements in objective measures of cardiopulmonary function.[8]

PE is a common congenital chest wall deformity associated with potentially significant physical and psychosocial morbidity. Timely diagnoses and appropriate management can alleviate symptoms, improve quality of life, and prevent long-term complications.

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