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Chest wall deformities: diagnosis, classification, and treatment approaches

Ayşe Uğurum Yücemen[1], Bülent Mustafa Yenigün[2], Cihan Genco Çetinkanat[3]

[1]Department of Thoracic Surgery, Sivas Numune Hospital, Sivas, Turkiye
[2]Department of Thoracic Surgery, Faculty of Medicine, Ankara University, Ankara, Turkiye
[3]Department of Thoracic Surgery, Citrusmed Ltd. Co., Ankara, Turkiye

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Corresponding Author: Cihan Genco Çetinkanat, genco.cetinkanat@citrusmed.com.tr

ABSTRACT

Chest wall deformities encompass a range of structural abnormalities that may be congenital or acquired, impacting various aspects of the thoracic structure and function. This document provides an extensive review of primary chest wall deformities, including pectus excavatum (PE), pectus carinatum (PC), Poland syndrome (PS), Jeune syndrome (JS), and sternal cleft (SC). PE, the most prevalent deformity, is characterized by the depression of the anterior chest wall and is often linked to genetic syndromes such as Marfan and Noonan. Its clinical implications, such as exercise intolerance and cardiovascular effects, necessitate careful evaluation through physical examination, imaging, and pulmonary function tests. For severe cases, the Nuss and Ravitch surgical procedures offer corrective options, each with specific indications, benefits, and complications. PC is typified by the protrusion of the sternum and is generally considered a cosmetic issue. However, in severe cases, patients may experience functional impairments. Conservative treatment, including a dynamic compression system, is suitable for mild to moderate cases, while severe cases may require surgical intervention via the modified Ravitch or Abramson procedures. PS involves unilateral absence or hypoplasia of the pectoralis major muscle and may affect rib structures. Treatment focuses on both functional and cosmetic correction, with surgical options including custom implants and reconstructive techniques. JS, primarily congenital, leads to a restricted thoracic volume, posing a significant respiratory risk. Surgical expansion of the thorax is critical in severe cases, utilizing methods like rib grafting and vertical expandable titanium rib placement. SC, a rare condition with incomplete fusion of the sternum, requires early surgical intervention to prevent complications like respiratory distress and mediastinal shift. Across these conditions, individualized treatment plans are essential, balancing surgical benefits with risks, and considering each patient's unique anatomical, functional, and psychosocial needs. The complexity of chest wall deformities necessitates multidisciplinary management to optimize outcomes and enhance quality of life for affected individuals.

Keywords: Pectus excavatum, pectus carinatum, Poland syndrome, Jeune syndrome, sternal cleft, thoracic surgery techniques

INTRODUCTION

Chest wall deformity can be seen in one-third of live births. There is a possibility that the deformity will progress during growth. Nuss stated that patients lose their chance of treatment because of the belief of pediatricians that "There is no need to worry, it will resolve itself over time" for these deformities.[1]

Chest wall abnormalities may be either congenital or acquired. They are categorized into three main groups. The major conditions are sternal depression (pectus excavatum) and sternal protrusion (pectus carinatum), caused by an imbalance in the development of the costochondral region. The second group includes anomalies that disrupt the normal development of the chest wall, including bifid sternum, pentalogy of Cantrell, and Poland syndrome (PS). The last group includes defects that develop due to trauma.[1-3]

PECTUS EXCAVATUM

Pectus excavatum (PE) is a malformation of the anterior chest wall marked by variable degrees of sternal depression. Its prevalence is 1 in every 400 to 1000 live births[4,5], with a higher incidence in males than females.[6] The deformity typically manifests in the early years of life and tends to worsen over time.[7]

Several hypotheses exist regarding the pathogenesis of PE, but none have received conclusive proof.[8] It is thought that the deformity may arise from an imbalance in the development of the costochondral regions.

PE is generally sporadic[9], nevertheless, genetic factors likely play a significant role, as familial inheritance has been sometimes shown.[10,11] PE is also linked to connective tissue disorders (e.g., Marfan syndrome), genetic syndromes (e.g., Noonan syndrome, Turner syndrome), and neuromuscular diseases (e.g., spinal muscular atrophy).[6,12,13]

Diagnosis

The most common symptoms in patients with PE include exercise intolerance, chest pain, poor endurance, and shortness of breath. As a result of distortion and displacement of the heart, stroke volume may decrease, and tachycardia may occur.[14] When evaluating these patients, consider the severity of the deformity, its impact on the cardiopulmonary system, and psychosocial factors.

During the physical examination, the degree of sternum depression is determined by inspection. The deformity may be either centralized or asymmetric. In asymmetric cases, the sternum is often rotated, and breast hypoplasia can be observed in female patients. 10–39% of patients with PE report having scoliosis.[5] Patients with respiratory complaints or those being considered for surgery should undergo pulmonary function testing. Patients with suspected heart issues should undergo a cardiac evaluation, which includes exercise testing, electrocardiography, and echocardiography. Patients who are not candidates for surgery should be followed up periodically, especially during adolescence.

For imaging, anteroposterior and lateral chest radiographs are typically obtained. In patients with moderate or severe PE and/or cardiopulmonary symptoms, computed tomography (CT) is used to assess the severity of the deformity. However, it is noted that symptom severity correlates poorly with the degree of deformity seen on CT.[15]

The Haller index (HI), also known as the pectus severity index, defines the depth of the defect. It is calculated by dividing the lateral diameter (A-B) of the chest by the distance between the sternum and vertebra at the point of maximum depression (C-D) on inspiratory CT (**Figure 1a**).[16] A normal HI is equal to or less than 2.5.[17] PE correction index (PCI), a horizontal line is drawn from the front of the vertebra. Then, the maximum distance (AB) between the inner edge of the most anterior part of the rib cage and the horizontal line is measured. The minimum distance (CD) between the inner edge of the most depressed point of the sternum and the horizontal line is measured (**Figure 1b**). The percentage of the patient's missing chest depth is calculated using the formula ((maximum distance - minimum distance)/maximum distance)x100.[18]

Patients with moderate to severe PE or respiratory symptoms should undergo pulmonary function testing. Although many patients with PE report respiratory symptoms, less than one-third have abnormal pulmonary function. There is little link between symptoms and pulmonary function.[19,20] Patients with moderate to severe PE or significant respiratory symptoms should undergo exercise testing, as it is more sensitive in detecting cardiopulmonary abnormalities.[9,21] Cardiovascular abnormalities are prevalent in individuals with PE and related

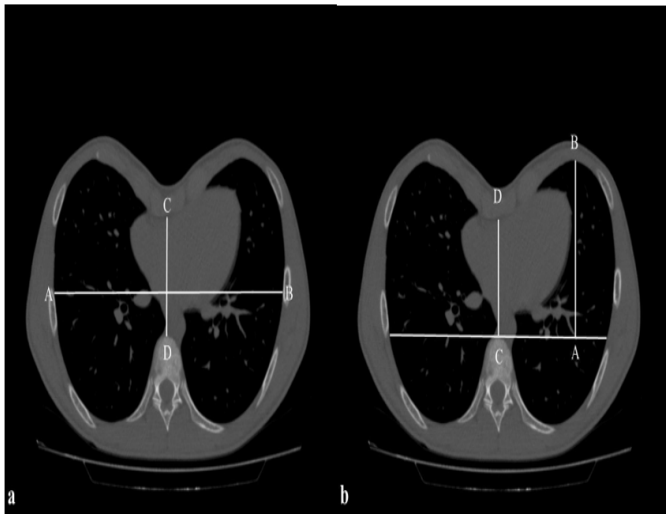

Figure 1. (**a**) Haller index: AB/CD. (**b**) Pectus excavatum correction index: ((AB-CD)/AB)x100

disorders.[12] Functional systolic murmurs may be observed in 18% of cases, while mitral valve prolapse occurs in 7-20% of instances.[5,6]

Kelly's criteria state that surgery is indicated for symptomatic and severe cases of PE if at least two of the following criteria are met: HI>3.25, cardiac and/or pulmonary compression demonstrated by CT or ECHO, restrictive breathing pattern in pulmonary function tests, presence of mitral valve prolapse or arrhythmia, history of unsuccessful corrective surgery, and severe discomfort with body image.[22–24]

Treatment

Depending on the severity of the malformation, treatment may be conservative or surgical. Surgeons frequently recommend surgical intervention prior to the complete ossification of the chest wall, ideally during late childhood or early adolescence.[23] In mild cases, or preoperatively in severe cases, vacuum bell therapy may be applied. Studies have shown that applying a vacuum bell can reduce the defect.[25-27]

The Ravitch procedure is an open technique. The intervention is performed by a transverse or vertical incision/ sternotomy incision on the sternum. The pectoralis major muscle is dissected to reveal the sternum. The defective cartilages (except for the second and third) are bilaterally resected subperichondrially (**Figure 2**). If the bony segment is affected, subperiosteal resection is also performed. Intercostal bands are freed, and the xiphoid process is resected. The sternum is mobilized from the xiphoid to the angle of Louis. The posterior manubrium and anterior corpus receive transverse osteotomy and wedge resection. The removed segments are placed in the osteotomy lines, bringing the sternum into its normal anatomical position. Kirschner wires are passed beneath the sternum and fixed to both sides of the chest wall.[28]

The most commonly used technique is the minimally invasive repair of PE (MIRPE), also known as the Nuss procedure. The points where the chest begins to depress inward are marked bilaterally as thoracic entry and exit at the most depressed point of the deformity. In addition, intercostal spaces at the intersection of the same line with the anterior axillary line bilaterally are marked as the incision site. The distance between both anterior axillary lines is measured to determine the length of the bar. The aluminum bar used as a mold is given the

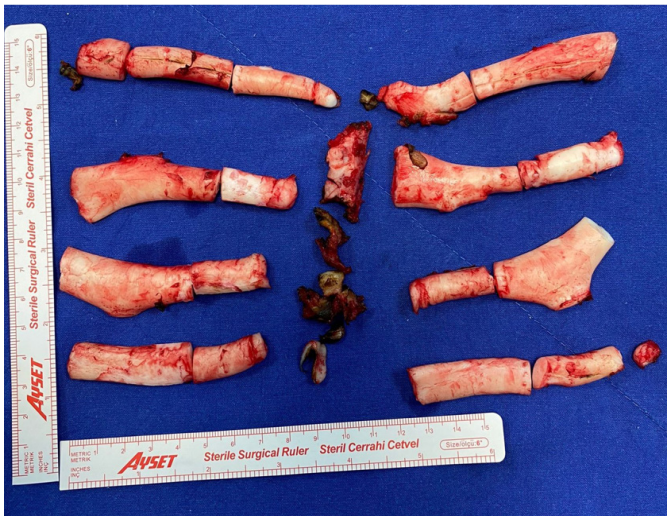

Figure 2. The intraoperative view of subperichondrially resected abnormal cartilages. (Resource: Department of Thoracic Surgery, Ankara University Faculty of Medicine, Ankara, Turkiye)

desired shape of the rib cage. Incisions are made at the marked intercostal spaces along the bilateral anterior axillary lines. A thoracoscope is inserted through the right hemithorax, and then intrathoracic air is infused. A tunnel is created over the ribs. A thoracoscope assists in passing the introducer through the parasternal intercostal space from the right hemithorax, advancing it over the pericardium, and removing it from the opposite hemothorax. The chest wall is shaped by lifting the introducer from both ends. The nylon type attached to the introducer is pulled and passed through the tunnel. Then the nylon type is removed from the introducer and attached to the pectus bar. The nylon-type fixed bar is passed through the tunnel with its convexity facing backward. The pectus bar is rotated 180 degrees and attached to a stabilizer on the left side. The bar is fixed to the ribs with absorbable monofilament pericostal sutures on the right side. The bar is removed between 2-4 years.[24]

Postoperatively, patients who receive the Nuss surgery generally report greater pain compared to those who undergo the Ravitch operation.[29] Secondary thoracic dystrophy is the most serious complication after the Ravitch procedure.[30] Pneumothorax and recurrence are other potential complications.[31]

During MIRPE, rare intraoperative complications include injuries to the heart, major vascular structures, and lungs, as well as severe bleeding. Early postoperative complications include spontaneous pneumothorax, pneumothorax requiring drainage, temporary Horner syndrome due to epidural analgesia, wound infection, pneumonia, pericarditis, and pleural effusion. Late complications (after one month) include bar displacement, overcorrection, allergic reactions to the metal bar, severe wound infections, pericardial effusion and pericarditis, and persistence of the deformity.[6,32-36]

PECTUS CARINATUM

Pectus carinatum (PC), also referred to as pigeon chest, is characterized by the protrusion of the sternum and costal cartilage.[37] It is more common in males than females.[4] Abnormal costal cartilage and sternum growth are believed to be the pathogenesis of PC, although the exact cause is unknown.[38,39] PC may be associated with various genetic syndromes and disorders affecting connective tissue.[40]

Classification is based on the anatomical region where the protrusion is most prominent. In chondro-gladiolar deformity, the middle and lower parts of the sternum show protrusion along with the costal cartilages, which may be symmetrical or asymmetrical. The costo-manubrial deformity, also known as pectus arcuatum (PA), is characterized by protrusion of the manubriosternal junction and adjacent costae.[41] The mixed type exhibits both PC and PE.

There is a special and rare form of the costo-manubrial variant called Currarino Silverman. Premature fusion and ossification of the manubriosternal joint and sternal segments cause this condition. The distal third of the sternum is short and thick, the 2nd to 5th ribs are angulated, and there is a high arcuatum appearance on the chest.[42]

Diagnosis

Typical PC deformity is usually recognized in early adolescence and worsens over time.[43] Patients are usually asymptomatic; however, symptoms such as respiratory problems and exercise intolerance may be observed in severe cases.[41] In untreated cases with isolated PC deformity, morbidity or mortality due to deformity is usually not observed.[44]

The anteroposterior diameter of the chest is enlarged, and chest wall mobility is typically restricted during physical examination.[41] The type and symmetry of the deformity are determined. It is checked whether the deformity improves when pressure is applied to the deformity. Direct radiographs are used to assess the severity of the deformity, and CT is performed on patients for whom surgery is planned.

Patients with PC typically have normal cardiovascular function. However, genetic syndromes associated with PC may increase the risk of mitral valve prolapse and aortic root dilatation, thereby recommending evaluation with ECHO.[45]

Treatment

PC is considered more of a cosmetic problem, and therefore there are no precise criteria for treatment. The patient's age, the severity, and the progression of the deformity determine the treatment options and timing.

Conservative treatment can be applied in mild and moderate cases. This treatment uses a dynamic compression system to correct the deformity by applying pressure to the rib cage before bone development is complete.[46] This method is applied for 12-18 months at daily intervals. Disadvantages include the fact that it cannot be used in adult patients and difficulty in patient compliance with treatment.[47] In order to increase treatment compliance, treatment can be started in the early period of puberty. This method is not effective in cases of P.[48]

Surgical treatment may be more effective in patients with severe PC, PA, moderate or severe sternal asymmetry, loss of chest wall elasticity, or in cases that do not comply with conservative treatment.[49]

The modified Ravitch procedure serves as the open surgical method for PC treatment. This procedure involves the resection of costal cartilages with abnormal angulation, while maintaining the perichondrium intact. A wedge osteotomy is performed on the protruding part of the sternum, and the resected portion is utilized as a graft to remodel the sternum. Absorbable or synthetic materials may also be used as grafts.[50]

The minimally invasive repair method is known as the Abramson procedure or modified Nuss procedure. In this method, an aluminum bar of appropriate size is shaped by applying pressure to the sternum at the point where the sternum is most protruding. The pectus bar is also shaped according to the aluminum bar. The intercostal space is reached through incisions made in the bilateral mid-axillary lines on the line where the protrusion is most prominent. Subperiosteally, two consecutive ribs are prepared, and fixation plates are fixed with pericostal wires. In this method, the pleural cavity is not entered. A chest tube with a trocar is passed through the tunnel opened in the presternal region. The trocar is removed, and the pectus bar is passed through the tube. The drain is retracted through the tunnel together with the bar. The bar is fixed to stabilizers on both sides and presses the sternum. The bar is usually removed after two years. Complications of this procedure include skin adhesion to the bar, seroma, bar breakage, infection, pain, and pneumothorax.[51]

POLAND SYNDROME

The absence of the sternocostal head of the pectoralis major muscle and hypoplasia or absence of the pectoralis minor muscle characterizes PS. The syndrome may also include aplasia or hypoplasia of the second to fifth costal cartilages, hypoplasia or absence of the breast and nipple, and ipsilateral brachydactyly and syndactyly. The presence and severity of the components are variable.[4]

PS is seen in 1/10.000 to 1/100.000 live births.[52] Most cases are sporadic, with rare reports of familial cases.[53,54] Although its etiology is still unclear, there is a hypothesis that it may have developed due to damage to the subclavian artery.[54]

In 2018, Romanini et al.[55] proposed a classification of PS to precisely identify patients, as the phenotype is significantly distinct. Type 1, also called the minimal form, has only pectoral muscle deficiency. Upper extremity or costal anomalies accompany the pectoral muscle defect in type 2, the partial form. Type 2a classifies upper extremity anomalies, while type 2b classifies costal anomalies. Type 3 represents the complete form, where both upper extremity and costal anomalies accompany pectoral muscle deficiency.[55]

In 2016, Romanini et al.[56] proposed the TBN classification to grade the severity of the defect, taking into account thoracic defects separately. T stands for thoracic defects, B for breast defects, and N for nipple-areola complex defect.

Diagnosis

These patients may have respiratory distress due to paradoxical respiratory movement, lung herniation, rarely spontaneous pneumothorax, bullous emphysema, and congenital diaphragmatic hernia.[57]

The most characteristic feature of PS is a partial or complete absence of the pectoralis major muscle.[54] Ultrasound is used to evaluate the pectoralis major muscle, breast tissue, and costae. Direct radiographs can be taken for costal anomalies; CT and 3D reconstructed CT are preferred in patients with planned surgery.[55]

Furthermore, for surgical planning in patients, a detailed evaluation of the internal thoracic artery is required.[58] ECHO and abdominal USG are recommended in terms of other accompanying pathologies.[55]

Treatment

Thoracic defects in PS are repaired for functional and cosmetic reasons.[54] Studies have shown that performing surgeries during the growth period improves quality of life more.[59] Surgical intervention may be performed in the presence of asymmetric depression in the chest wall, paradoxical respiration, poor protection of the heart and lungs, the presence of breast hypoplasia or aplasia in female patients, and cosmetic concerns.[53]

A midline anterior thoracic incision is made for anterior chest wall defects; in addition to this short incision, additional incisions can be made in the axillary line for costal anomalies. This incision allows for the resection of the PC-forming cartilages on the opposite side of the PS. Sternal osteotomy can be added if necessary. Cases with a single costal anomaly do not require surgery, whereas Gore-Tex mesh is used in cases with two costal anomalies and titanium bars are used in cases with two or more costal anomalies. Gore-Tex mesh should be placed between the titanium bar and the skin. A tissue expander or pectoral/breast prosthesis can be placed over this mesh layer. In recent years, CT-scanned and 3D-printed models of the rib cage have been used through these surgical incisions. If PS is accompanied by PE and PC, the classic PC or PE treatment is used.[55,60]

JEUNE SYNDROME

Jeune syndrome (JS) may be congenital or acquired. Acquired JS develops due to the loss of cartilage growth centers following excessive resection during prepubertal chest wall reconstruction surgeries. Congenital JS, also known as asphyctic thoracic dystrophy, is an autosomal recessive polychondrodysplasia. The incidence is between 1/100.000 and 1/130.000. In this syndrome, the costae are short and horizontal, and the costochondral joints are irregular. The thorax is narrowed and has reduced elasticity, resulting in decreased thoracic volume and reduced lung ventilation.

The severe form is characterized by pulmonary hypoplasia and pulmonary hypertension and presents soon after birth. The moderate form is seen in the first years after birth. Both forms are associated with respiratory failure and pneumonia. In the mild form, patients may have low exertional capacity, and their respiratory problems tend to decrease as they reach adolescence. There are also patients with normal lung function.[61,62]

Diagnosis

Other characteristic features of the syndrome include short arms and short legs. Kidney, liver, pancreatic, and ocular abnormalities may occur.[61,62] The diagnosis is made with clinical and radiologic findings. CT with 3-dimensional reconstruction is useful for surgical planning. ECHO and cardiologic evaluations are essential. Pulmonary hypertension and cor pulmonale may result from respiratory failure, making surgery contraindicated.

Treatment

Surgery for JS should be performed soon after birth or in early childhood. However, for stable patients and mild forms,

it is thought that waiting a few years to allow the costae to develop may increase the success of surgery. The aim of surgical treatment is to increase thoracic volume by expanding the thorax. The presence of recurrent respiratory infections, respiratory distress, growth retardation, and pulmonary hypertension determines the indication and timing of surgery.

There are various approaches to surgery.[63,64] One technique achieves thoracic expansion by placing retractors or grafts after a median sternotomy. The lateral thoracic expansion (LTE) technique involves performing a thoracotomy and splitting the 4th to 9th ribs at their midpoints. The ribs to be operated on are dissected free from the surrounding muscular tissue. The 5th and 6th, as well as the 7th and 8th ribs, are then cut and approximated towards the midline, where they are fixed with titanium bars. This method provides two longer, oblique ribs instead of two horizontal ones.[65] Davis recommends conducting a second surgery on the contralateral hemithorax after 6 months[65], but some authors prefer bilateral surgery in the same session.[66] Another technique, vertical expandable titanium rib (VEPTR), allows for vertical rib cage expansion. In this procedure, a titanium bar is placed vertically and secured to the ribs with locking cradles at both the top and bottom. The apparatus adapts to the patient's growth by sliding along the ribs.[67]

STERNAL CLEFT

Sternal cleft (SC) is a partial or complete fusion defect of the two halves of the sternum.[53] SC appears in 1 in 50.000–100.000 live births.[68] The etiology of SC is unknown. There are 4 main types, including superior and inferior SC, subtotal, and total SC. Superior SC with cleft mandible and median SC are rare types. The most common forms are superior and subtotal.[53] Inferior SC is frequently associated with Cantrell pentalogy.[69] Sternal defects are classified as thoracic ectopia cordis, cervical ectopia cordis, thoracoabdominal ectopia cordis (Cantrell pentalogy), and SC.[70]

Diagnosis

USG can diagnose SC during the intrauterine period, and magnetic resonance imaging (MRI) confirms the diagnosis. Fetal ECHO should also be performed for the presence of concomitant Cantrell's pentalogy.[69] Direct radiography, CT, and 3D reconstructed CT are used in patients with a surgical plan.

Treatment

Surgical intervention in the treatment of SC may be performed for reasons such as paradoxical respiratory movement and related complications (mediastinal shift, right ventricular overload, arrhythmia), dyspnea, frequent respiratory tract infections, vulnerability of the heart and great vessels to trauma, risk of dermopericardial fistula development, possibility of enlargement of the defect over time, and cosmetic discomfort caused by the external view of the heartbeat.[71] The most suitable time for surgery is the neonatal period, when the chest wall is most flexible and the risk of compression of the underlying structures is minimal.

In SC surgery, intervention is performed by making an anterior thoracic incision along the midline. A V-shaped incision is made in the intact sternum to enable primary repair in cases with partial SC (**Figure 3a**). The sternal halves are approximated using nonabsorbable sutures (**Figure 3b, 3c**). Approximation during primary repair may cause increased mediastinal pressure, which could compromise venous return and cause hemodynamic disturbances.[72,73] To minimize this risk, Torre et al.[72] recommend complete or partial removal of the thymus. In cases of large defects or in older patients where the sternal halves cannot be easily approximated due to poor compliance, multiple chondrotomies may be performed to ease the closure and reduce mediastinal pressure. Periosteal flaps, dissected from the sternal bars, are rotated inward and sutured at the midline to form the posterior wall of the newly reconstructed sternum. Any residual gap can be filled with cartilage grafts or, alternatively, synthetic prostheses. If primary repair cannot be achieved, materials such as Gore-Tex, or bone grafts can be used to bridge the gap.[10,74,75]

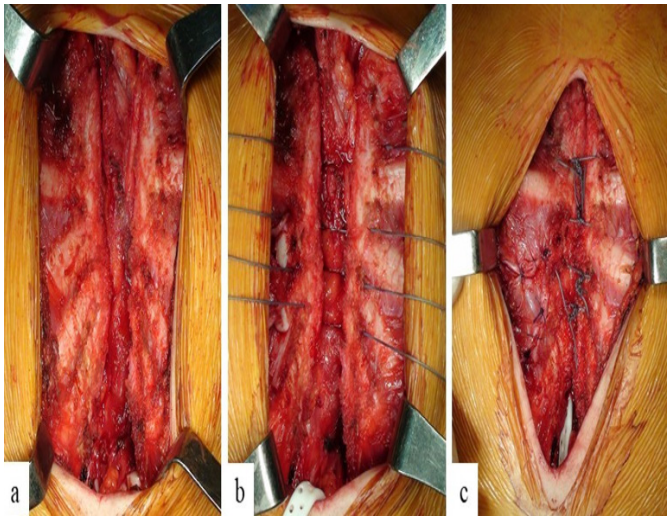

Figure 3. The intraoperative view of sternal cleft. (**a**) The view of seperated sternal halves. (**b**) Sternal halves were prepared for primary repair. (**c**) Both sternal halves were sutured with interrupted nonabsorbable sutures. (Resource: Department of Thoracic Surgery, Faculty of Medicine, Ankara University Ankara, Turkiye)

CONCLUSION

Chest wall deformities present with varying degrees of functional and psychological challenges. There is a weak correlation between the severity of the deformity and the symptoms. Therefore, each patient should be evaluated individually, and a treatment plan should be developed through a multidisciplinary approach, considering both conservative and surgical options. Recent advancements in minimally invasive techniques and prosthetic applications have contributed to reduced complications and improved cosmetic and functional outcomes.

ETHICAL DECLARATIONS

Referee Evaluation Process

Externally peer-reviewed.

Conflict of Interest Statement

The authors have no conflicts of interest to declare.

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Author Contributions

All of the authors declare that they have all participated in the design, execution, and analysis of the paper, and that they have approved the final version.

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