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CLINICAL FEATURES AND EARLY DIAGNOSTIC METHODS OF PECTUS EXCAVATUM IN CHILDREN

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Abstract: Pectus excavatum (PE), also known as funnel chest, is the most common congenital deformity of the anterior chest wall, characterized by a sunken sternum and adjacent costal cartilages. This article provides an in-depth clinical analysis of PE in children, examining its etiology, pathogenesis, classification, clinical manifestations, modern diagnostic approaches, and preventive measures. Emphasis is placed on the role of early diagnosis through physical examination and imaging techniques such as computed tomography (CT), spirometry, and echocardiography (ECHO). The study also evaluates the impact of PE on cardiopulmonary function and psychosocial development, offering insights for pediatricians, thoracic surgeons, and orthopedists.

Keywords: Pectus excavatum, funnel chest, chest wall deformity, Haller index, pediatric cardiothoracic disorders, early diagnosis, spirometry, computed tomography, echocardiography.

Introduction

Pectus excavatum (PE), commonly known as funnel chest, is the most prevalent congenital deformity of the anterior chest wall. It is characterized by a depression of the sternum and adjacent costal cartilages, resulting in a concave or sunken chest appearance. PE accounts for approximately 90% of all congenital chest wall deformities and is observed in approximately 1 out of every 300–400 live births [1]. The condition is more common in males, with a male-to-female ratio of around 3:1 to 4:1 [1], [2]. While some cases are noticeable at birth, the deformity often becomes more pronounced during adolescence due to rapid skeletal growth.

Although PE is often perceived as a cosmetic defect, several studies have demonstrated its potential impact on cardiopulmonary function and psychosocial well-being. In moderate and severe cases, the depressed sternum can lead to cardiac compression, decreased stroke volume, and restricted lung expansion, resulting in dyspnea, reduced exercise tolerance, and fatigue [3]. These physiological issues may significantly impair children's physical development and daily activities.

In addition to physical symptoms, psychological impacts of PE are well documented. Children and especially adolescents may experience low self-esteem, social withdrawal, anxiety, and depression due to their chest appearance [2], [4]. These psychosocial effects can be more debilitating than the physical limitations, emphasizing the need for a multidisciplinary approach to care.

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Etiologically, PE is considered multifactorial, involving both genetic predisposition and environmental influences. A positive family history is present in up to 40% of cases, and PE frequently coexists with connective tissue disorders such as Marfan syndrome and scoliosis [1], [5]. The primary underlying mechanism is thought to involve abnormal growth of the costal cartilages, which pushes the sternum inward.

In the past, PE was often underdiagnosed or dismissed as a benign anomaly. However, advancements in imaging techniques such as chest X-ray, computed tomography (CT), magnetic resonance imaging (MRI), and echocardiography have improved our ability to diagnose and quantify the severity of deformity. The **Haller Index**, which is calculated from axial CT images, remains the standard metric for determining surgical candidacy. A Haller Index greater than 3.25 is generally considered an indication for operative correction [3], [6]. Early diagnosis of PE, particularly during childhood or early adolescence, allows for better treatment outcomes. Non-surgical methods such as vacuum bell therapy can be considered in selected mild cases, while surgical correction—typically performed using the minimally invasive Nuss procedure—is most effective when the chest wall is still flexible, typically between the ages of 10 and 16 [4], [6].

This article aims to describe the clinical features of PE in children, explore modern diagnostic approaches, and emphasize the importance of early detection and intervention. By increasing awareness among pediatricians, thoracic surgeons, and general practitioners, we can promote timely diagnosis and management, thereby improving the long-term physical and psychological outcomes for affected children.

Etiology and Pathogenesis

Pectus excavatum (PE) is a congenital chest wall anomaly characterized by a posterior depression of the sternum and adjacent costal cartilages. The precise etiology of PE remains incompletely understood; however, increasing evidence suggests a multifactorial origin involving genetic predisposition, abnormal cartilage growth, and possible connective tissue defects. In up to 40% of cases, a positive family history is observed, indicating a potential hereditary pattern of inheritance, possibly autosomal dominant with variable penetrance. This familial clustering supports the hypothesis that genetic factors play a substantial role in the development of the deformity.

From a developmental biology perspective, the pathogenesis of PE is believed to involve dysregulation in the growth and structural integrity of costal cartilage. Histological analyses have demonstrated disorganized collagen fiber architecture and reduced chondrocyte density in affected cartilage, suggesting intrinsic cartilage weakness and altered biomechanical properties. These changes lead to excessive inward bending of the anterior chest wall, particularly during periods of rapid growth in adolescence. The abnormal cartilage growth does not affect all costal ribs equally, resulting in asymmetric or symmetric depression of the sternum.

Molecular and biomechanical studies have suggested that overgrowth of costal cartilage may not be the sole mechanism. Rather, impaired structural resilience and a lack of tensile support in the anterior thoracic framework may permit the sternum to collapse inward under the influence of negative intrathoracic pressure. Furthermore, the involvement of connective tissue anomalies has been well documented, particularly in syndromic cases. PE frequently coexists with connective tissue disorders such as Marfan syndrome, Ehlers-Danlos syndrome, and Noonan syndrome, which are characterized by abnormalities in fibrillin-1, collagen synthesis, or extracellular matrix regulation. These associations further support the notion that PE is not

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merely a localized skeletal deformity but may be a phenotypic manifestation of systemic connective tissue pathology.

On a molecular level, mutations in genes involved in cartilage development and matrix stability—such as **FBN1**, **COL1A1**, and **TGF-β** signaling pathways—are under investigation. Abnormalities in these genetic pathways may impair chondrogenesis or alter the mechanical integrity of cartilage and bone. Although specific genetic mutations responsible for isolated PE have not been definitively identified, ongoing whole-exome sequencing studies have identified several candidate genes.

Another contributing factor may include mechanical and intrauterine pressures during gestation that influence the shape of the fetal thoracic cage, though these theories remain speculative. Additionally, studies using animal models and biomechanical simulations have shown that certain thoracic postures and muscle imbalances during early childhood growth may exacerbate the progression of PE in predisposed individuals.

Notably, PE is not merely a static deformity but often progressive during growth, particularly around puberty. The flexible chest wall in pediatric patients is more vulnerable to deformation under unbalanced forces, and without intervention, the depression may deepen over time. This dynamic progression necessitates timely evaluation and intervention.

In summary, the etiology and pathogenesis of pectus excavatum are complex and multifaceted, involving a combination of genetic, molecular, structural, and possibly environmental factors. Advances in genetic and biomechanical research continue to elucidate the underlying mechanisms, which may ultimately guide the development of targeted therapies or early screening protocols for at-risk populations.

Diagnostic Methods

1. Clinical Examination

- Inspection of chest wall morphology.
- Measurement of chest dimensions and asymmetry.
- Auscultation for heart murmurs or abnormal breath sounds.
- Posture and spine assessment.

2. Radiological and Instrumental Investigations

- **Chest X-ray**: Initial screening, shows sternal depression and cardiac shift.
- **Computed Tomography (CT)**: Gold standard for anatomical evaluation and Haller index calculation.
- **Spirometry**: Assesses pulmonary function (FVC, FEV1). Reductions indicate restrictive lung impairment.
- **Echocardiography (ECHO)**: Detects right heart compression, tricuspid regurgitation, and decreased stroke volume.
- **MRI (optional)**: In complex or recurrent cases for soft tissue and vascular mapping.

3. Psychological Evaluation

- Screening tools for body image dissatisfaction and emotional impact.
- Clinical interviews and psychosocial counseling.

Clinical Study and Results

A prospective study was conducted in a pediatric thoracic surgery center in Tashkent (n=72, ages 5–16), and patients were classified based on Haller index:

Severity	No. of Patients	Major Clinical Findings

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Mild (2.5–2.9)	22	Cosmetic concern only
Moderate (3.0–3.4)	31	Dyspnea, fatigue, mild arrhythmias
Severe (≥3.5)	19	Cardiac compression, reduced FVC (<70%), abnormal ECHO

- **Average Haller index**: 3.2
- **Cardiac displacement**: Detected in 42% of patients
- **Pulmonary function impairment**: Observed in 58%
- **Psychosocial dysfunction**: Present in 64% (especially among adolescents)

Importance of Early Diagnosis

Timely identification of PE allows for:

- Preventive postural and physiotherapeutic interventions.
- Avoidance of long-term cardiopulmonary compromise.
- Improved cosmetic outcomes with conservative or surgical management.
- Enhanced psychosocial development during adolescence.

In mild cases, early intervention with physiotherapy and respiratory exercises can halt progression. Moderate to severe cases benefit from surgical correction during prepubertal or early pubertal years for optimal outcomes.

Management Options

1. Conservative Therapy

- **Physical therapy**: Breathing exercises, posture correction, chest wall mobilization.
- **Bracing**: Vacuum bell therapy (for compliant, younger patients).
- **Swimming and aerobic exercises**: To improve thoracic expansion.

2. Surgical Treatment

- **Nuss Procedure**: Minimally invasive bar placement to elevate the sternum (ideal for children aged 10–16).
- **Ravitch Procedure**: Open resection of abnormal cartilages and sternal realignment (reserved for complex deformities).
- **Postoperative care**: Pain management, physiotherapy, and bar removal after 2–3 years.

Discussion

The clinical burden of PE in pediatric populations encompasses more than physical deformity. It poses significant challenges to respiratory efficiency, cardiac performance, and mental health. Although many patients present with asymptomatic cases, the potential for progressive dysfunction makes early screening and structured follow-up essential.

Advancements in minimally invasive thoracic surgery, particularly the Nuss procedure, have revolutionized the treatment paradigm for PE. However, surgical outcomes are maximized only when performed at the appropriate age and with proper preoperative planning. Conservative treatments also hold promise in early stages, especially when combined with psychosocial support to address body image concerns. The interdisciplinary collaboration of pediatricians, surgeons, physical therapists, and psychologists is critical in holistic patient care.

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