Congenital Thoracic Deformities

9

Giovanna Riccipetitoni, Sara Costanzo, and Francesca Destro

9.1 Introduction and Classification

Congenital deformities of the chest wall or chest wall malformations (CWMs) comprise a complex spectrum of anomalies which range from those that do not impact on the health and quality of life of the patient to those which are life-threatening [1–3].

CWM occurs when there is anomalous skeletal development and/or formation of the thoracic cavity. The formation of the thoracic cavity occurs after the intraembryonic cavity has formed (around the 4th week of gestation). At the beginning of the 5th week of gestation, the lung buds rapidly grow caudally and laterally at the pericardioperitoneal canals inducing development of the thoracic wall and pleuropericardial membranes. The sternum also develops during the 6th week of gestation. The complex development of the sternum can be simplified into three stages: formation, chondrification, and ossification [4]. Formation begins with the condensed mesenchyme where paired parallel mesenchymal bands migrate from the lateral plates and fuse in the midline by the 10th week of gestation. Chondrification begins immediately once the sternal plates fuse. Ossification begins approximately during the 6th month of gestation. It occurs in isolated centers starting in the manubrium, subsequently in the middle body during the 7th month, and finally in the lower body during the first postnatal year. The xiphoid process will undergo ossification between the 5th and 18th years of life, and the multiple ossification centers do not coalesce until after puberty.

Thoracic deformities may also be acquired, established postnatally as secondary to other conditions that disrupt the thoracic wall. Acquired chest wall deformities typically follow prior chest surgery (e.g., costal fusions caused by thoracotomies carried out in any period of childhood, sternal diastasis secondary to an infection of a sternotomy wound) or a posterolateral diaphragmatic hernia repair (Bochdalek) or spine deformities (i.e., kyphoscoliosis, hemivertebrae, wedge vertebrae, defects of vertebral segmentation). Secondary CWM may also follow fetal procedures carried out for congenital pulmonary malformations, such as thoracentesis or thoracoamniotic shunt [5].

Chest wall can be markedly deformed, by various degrees of anteroposterior flattening, in patients affected by bronchopulmonary dysplasia (BPD), a common complication of preterm birth (Fig. 9.1). The reason of this phenomenon is not completely explained, although the combination of many factors, such as the long-standing lung function abnormalities, the presence of

G. Riccipetitoni (✉) · S. Costanzo · F. Destro
Pediatric Surgery Department, Ospedale dei Bambini V. Buzzi, Milano, Italy
e-mail: giovanna.riccipetitoni@asst-fbf-sacco.it

© Springer Nature Switzerland AG 2019

M. Lima, O. Reinberg (eds.), *Neonatal Surgery*, https://doi.org/10.1007/978-3-319-93534-8_9

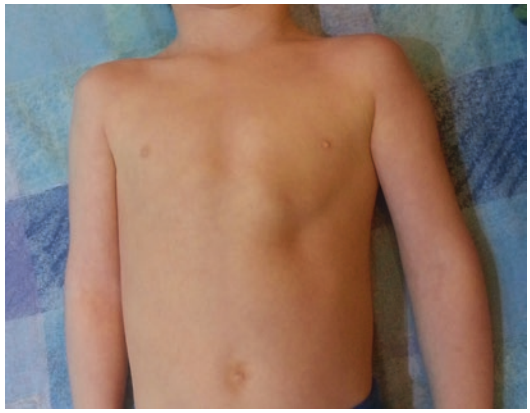

Fig. 9.1 Unilateral costal deformity in an ex-preterm child with bronchopulmonary dysplasia

demineralized and sometimes fractured ribs, and chronic sternal retraction, which is common in respiratory distress of any cause in infants, may contribute to chest flattening [6, 7].

A straightforward and complete classification of CWM was proposed by Eduardo Acastello in 2006, distinguishing CWM into five types, according to the origin of the anomaly [8, 9] (Table 9.1):

1. Cartilaginous
2. Costal
3. Chondrocostal
4. Sternal
5. Clavicle-scapular

It is difficult to accurately determine the overall incidence of CWM. This is due to multiple factors, among which their broad spectrum and the poor record of some mild deformities which do not even come to the medical attention. However, the specific incidence of the majority of them is clearly established in the published literature and will be detailed in the subsequent analysis of each malformation. Type I deformities are the most frequent, comprising more than 90% of the entire spectrum.

CWMs are rarely diagnosed prenatally. Three-dimensional ultrasound offers a way to visualize fetal skeletal dysplasias such as Jeune syndrome. Other CWM can be indirectly suspected on the basis of other anomalies detected, for example, the presence of hypoplasia or aplasia of one forearm or one hand in Poland's syndrome or the visualization of an ectopic heart in sternal anomalies [10–12].

Table 9.1 Acastello classification of chest wall malformations

Type I	Cartilaginous anomalies	Pectus excavatum
		Pectus carinatum
Type II	Costal anomalies	Simple
		Complex
		Syndromic (always complex)
Type III	Chondrocostal anomalies	Poland syndrome
		Thoracopagus conjoined twins
Type IV	Sternal anomalies	Sternal cleft
Type V	Clavicle-scapular anomalies	Clavicular
		Scapular
		Combined

A detailed evaluation is required for each CWM patient that comes to the medical attention [13]. It should start with an interrogation about personal and family history, since familial cases can be detected in up to 30% of patients. Great relevance has to be paid to the presence of associated malformations, which can affect vital organs (e.g., heart and lungs) and might allow to diagnose a syndromic form (Marfan, prune belly, etc.). The moment of detection, its evolution, and possible related symptoms must be always documented.

The physical examination of the patient has to be accurate. Inspection in the different positions (standing, in front and lateral view, in dorsal decubitus) will provide the most relevant data, in order to classify the type of deformity, its symmetry or asymmetry, the degree of alteration (mild, moderate, or severe), functional compromise, secondary deformities (kyphosis, scoliosis), and postural defects (scoliotic attitude).

The determination of thoracic measurements has a fundamental value in standardizing medical observations. The thoracic measurements to be evaluated are:

1. Thoracic circumference: measured during expiration on a line that crosses the nipples on the anterior aspect of the thorax and just below the scapular angles in the posterior region.

2. Intermamillary distance: it measures the distance between the two nipples and the distance from the half-sternal line to each nipple separately and serves to assess the symmetry or asymmetry of the chest.
3. Thoracic index: it is the ratio between the anteroposterior and the lateral diameters of the thorax (measured at the level of the nipples) multiplied by 100.

Imaging studies are necessary to complete the diagnostic work-up [14]. Chest radiograph in double view (front and profile) can provide basic information about the type and the degree of alteration and changes produced; it can identify, for example, the form of dysmorphic ribs or alterations in the shape of the thoracic cage; it is also useful for long-term postoperative follow-up. Second-level imaging techniques, such as computed tomography with or without three-dimensional reconstruction or magnetic resonance imaging, are usually indicated to better define the anatomy of the malformation.

The level of cardiopulmonary impact of each malformation is variable and depends on the particular characteristics of each pathology. Every patient with a CWM should undergo cardiac evaluation, comprising at least electrocardiographic exam and cardiac ultrasound; the specialist will determine the following steps, after evaluating the presence of congenital heart diseases and their possible functional repercussion.

A pulmonology evaluation has to be always requested, although a proper respiratory functional assessment is usually not feasible before 3 years of age.

The orthopedic evaluation completes the assessment of CWM children, given the high incidence of other skeletal anomalies, such as congenital scoliosis and vertebral deformities, in this group of patients.

With regard to treatment, not all CW malformations require surgical correction. Each pathology presents its particular indications to treatment, and it is fundamental to identify the right timing for it, to achieve the best esthetic and functional results [15].

9.2 Pectus Excavatum

Pectus excavatum (PE), also termed funnel chest, is characterized by the presence of a variably deep sternal depression associated with a malformation of the lowest chondrosternal joints. PE is the most frequent thoracic malformation, with an incidence of 1/100–1/1000 live births, and accounts for around the 90% of all CWMs. It occurs more frequently in males than females by a 5:1 to a 3:1 ratio. About 95% of cases occur in Caucasian patients, whereas Asian, African American, and Hispanic patients represent only the minority of PE cases. PE is most often congenital and noted in the first year of life (86%) (Figs. 9.2 and 9.3), while in the remaining cases, it appears later during development; in this last group of patients, there is a frequent association with malformations of the muscular connective tissue, such as Marfan and Ehlers-Danlos syndrome. Cases of spontaneous resolution during growth are very uncommon, and the more typical course is worsening of the depression, either gradual or more dramatic during phases of rapid vertical growth and puberty.

9.2.1 Etiopathogenesis

The etiology of PE is not clear, and many hypotheses have been proposed. The overgrowth of costal cartilages could be the pathogenetic mechanism leading to the development of PE. Another mechanism proposed is an abnormal tethering of the sternum to the diaphragm posteriorly. This theory is supported by a 33% incidence of acquired PE in patients after repair of posterolateral congenital diaphragmatic hernias (Bochdalek). Collagen type II disorders have been demonstrated in the costal cartilages in PE, as well as overexpression or downregulations of some genes playing a role in the metabolism of cartilage and connective tissues, as collagen genes, matrix metalloproteinases, tumor necrosis factor-alpha, and filamin [16, 17]. Although there is no confirmed chromosomal abnormality, there is a genetic predisposition supported by a familial recurrence in up to 40% of the cases [18]; more

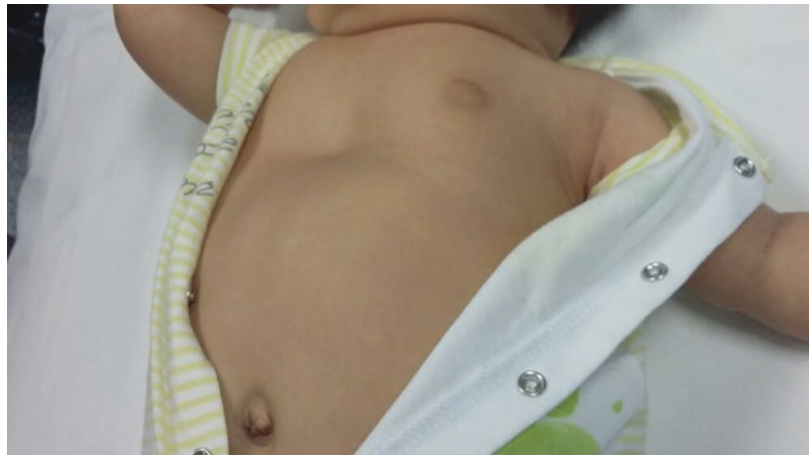

Fig. 9.2 Neonate with pectus excavatum [*Courtesy by Dr. Michele Torre, G. Gaslini Children's Hospital, Genoa, Italy*]

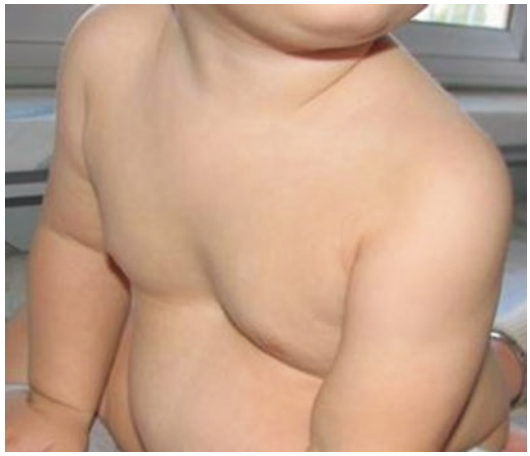

Fig. 9.3 Infant with pectus excavatum [*Courtesy by Dr. Michele Torre, G. Gaslini Children's Hospital, Genoa, Italy*]

rarely we can observe the presence of other CWM (such as PC) in a PE family. Four different possible patterns of inheritance have been suggested: autosomal dominant, autosomal recessive, X-linked recessive, and complex.

9.2.2 Pathophysiology

In cases of severe malformations, there can be physiological repercussions. Many studies have tried to elucidate the implications of PE on the respiratory and cardiac function [19]. Thoracic CT and MRI scans often demonstrate a leftward displacement of the heart and a compression of the right ventricle or atrium, deformed by the rotation and posterior displacement of the sternum, with different degrees of dysfunction on the echocardiogram. Mitral valve prolapse and mitral valve regurgitation are frequent, due to the deformation of the mitral valve annulus [20].

Pulmonary function tests (PFT) can be altered, more on stress conditions than on rest [21]. The most common pattern of PE is a restrictive one, but also obstructive or mixed patterns are not uncommon. A possible explanation for ease of fatigability in PE patients can be a less efficient mechanism of breathing [22, 23]. Using motion analysis technology, PE patients were determined to have significantly impaired chest wall motion at the area of the pectus defect and increased abdominal contributions to respiratory activity [24]. Other studies support the theory that exercise capacity is limited as a result of reduced filling of the right heart by the compressive effects of PE.

PE can be observed in some neonates with congenital diaphragmatic hernia or children with respiratory obstruction (bronchomalacia, hypertrophic tonsils). These particular cases of PE are the only ones that can improve during infancy. PE is otherwise usually mild at birth and progresses over the years, especially during preadolescent and adolescent age.

9.2.3 Clinical Evaluation

A complete history has to be collected, including questions regarding onset and progression of chest wall deformity and investigating the possible presence of symptoms, associated conditions, and familial cases. Most young children with PE are asymptomatic, but, as they become more active, particularly in their pre- and early teenage years, exercise intolerance and lack of endurance can appear.

The morphology of the PE should be described in detail [25, 26]. The deformity may be described as symmetric or asymmetric; asymmetry is often associated with sternal rotation that has to be underlined too.

PE can be morphologically classified as follows:

- Grand Canyon: It is a severe form of PE, with a deep long canal in the sternum, which is usually extremely rotated. This type is often asymmetric.
- Punch or cup shape: Localized deformity, usually on the inferior part of the sternum. It is more often symmetric. It has been observed that the punch type is the most common variant of PE (67%), more commonly symmetrical (80%), to the right of the midline (80%), and involving the lower sternum (99%).
- Saucer shape: It is a diffuse depression involving the complete anterior chest, where the thorax is usually quite flat. It can be symmetric or asymmetric.
- Transversal PE: The depression is transversal and below the sternum.
- Eccentric PE: The sternal depression is eccentric to the midline. It is the highest degree of asymmetric PE.
- PE with flaring chest: The lower ribs are flaring at each side of the depressed sternum.
- PE-PC: It is a combined malformation with a sunken chest and unilateral or bilateral protrusion of the cartilages beside the sternum edge.
- Superior PE: Very rare PE, localized in the upper part of the sternum; lower sternum is normal.
- Photographic documentation with different angles (frontal, left and right lateral, left and right oblique) is very helpful for the follow-up of these patients.

A thorough chest and cardiac physical exam completes the first part of the evaluation of these children [27].

9.2.4 Tests and Imaging

CT or MRI of the chest, PFT, and cardiac evaluation including electrocardiogram and echocardiogram are a routine part of the evaluation for a patient with PE [28].

There are several numeric indexes, usually calculated on CT and MRI, that have been developed over time to help quantify the severity of PE [29–31]. The most relevant are:

- Haller index (HI): described in 1987 by J. Haller et al., [32] it is calculated dividing the transverse maximum diameter of the rib cage by the anteroposterior diameter at the deepest point of the deformity. The cutoff point for PE patients is >3.25.
- Sternal depression index (SDI): it is the ratio between the maximal internal sagittal diameter of the left side of the chest and the minimal distance between the anterior surface of the vertebral column and the posterior border of the deepest portion of the sternum. An SDI of <2.4 is associated with mild sternal deformity, moderate sterna deformity is associated with an SDI of 2.4–2.9, and an SDI of >2.9 is conclusive of severe sternal deformity.
- Asymmetry index (AI): it is the ratio between the anteroposterior diameter of the right and the left part of the rib cage, multiplied by 100. Chest wall is considered asymmetric if AI is <−0.05 or >0.05.
- Others: correction index (CI), eccentricity index (EI), and others.

PE causes displacement of the heart into the left hemithorax. In order to measure the extent of cardiac compression, specific indexes, calculated

on CT or MRI, have been calculated, such as the cardiac compression index (CCI) and the cardiac asymmetry index (CAI).

9.2.5 Management

The optimal age for repair is 10–14 years old because at this time the rib cage is more malleable, thus allowing for rapid recovery, better results, and a lower recurrence rate as the bar remains in place during musculoskeletal maturation [33, 34]. Fixing PE in the first years of life is probably unnecessary, and it could carry the risk of relapse or postoperative severe complications as acquired Jeune syndrome [35].

9.3 Pectus Carinatum

Pectus carinatum (PC) is a protrusion of the sternum and chondrocostal joints (Fig. 9.4). It is the second most common CWM, and its incidence is estimated to be five times less frequent than PE in North America, although in other countries, it is much more common, for example, in Argentina, where it has been reported to comprise 55% of chest wall deformities. There is a strong male predominance (4:1). Most cases of PC are sporadic; however, familial incidence has been reported in about 26% of cases; in some families, it is possible to observe both PC and PE cases.

9.3.1 Etiopathogenesis

The etiology is unknown, although the origin of PC is considered to be similar to that of PE, related to abnormalities of connective tissue development, giving its frequent association with other skeletal abnormalities such as scoliosis (12%), and some genetic predisposition.

PC can be either an isolated condition or part of a syndrome (i.e., Poland syndrome) or connective tissue disorder.

9.3.2 Classification

PC can be classified into the following types:

– Type 1, inferior or chondrogladiolar: It is the most frequent type; the protrusion is located in the inferior sternum, with maximum prominence at the sterno-xiphoidal junction, which can be very noticeable (“pyramidal” or “keel chest”); it

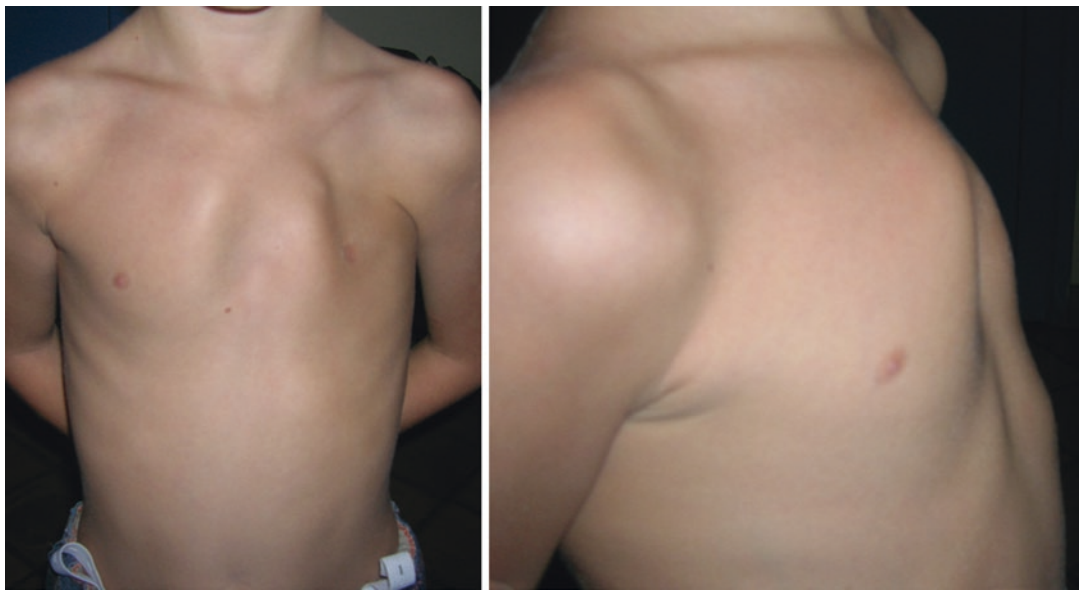

Fig. 9.4 Asymmetric pectus carinatum in frontal and lateral view

is usually symmetric. This type of PC can be associated with lateral depressions of the ribs.
– Type 2, superior or chondromanubrial: The protrusion is localized above the intermammary line. A variant of this form is called *Currarino-Silverman syndrome* or pouter pigeon breast, characterized by a high symmetric carinatum chest deformity with a short thick sternum with a depression in the lower third [36]. Its aspect is of a superior PC with an inferior PE, and the sternum is typically S-shaped on a lateral view.

A rarer form called lateral PC has also been described: always asymmetric by definition, it consists in the protrusion of some costal cartilages, besides the chondrosternal joints, on one side, with often concomitant rotation of the sternum (30–60–90°) toward the opposite side.

9.3.3 Clinical Presentation and Imaging

In contrast to PE, PC usually appears later in life. Only about one-sixth of the patients show a carinate deformity within the first year of life, while in almost half of them, PC is diagnosed noted during prepuberty or puberty. The deformity, which may be mild at birth, often worsens rapidly during the growth spurt.

Most PC patients are asymptomatic, although those affected by a severe anomaly may complain of some degree of thoracic pain. Cardiac and pulmonary functions are usually less affected than in PE. Only the variant "pouter pigeon breast" can be associated with congenital heart disease in 18% of cases.

Imaging studies include posteroanterior and lateral chest radiographs, although CT scan remains the gold standard radiologic evaluation for PC. Some radiological indexes, measurable on CT scan, have been proposed, but in clinical practice, they are less used than those calculated for PE.

9.3.4 Management

PC is usually not treated during the first years of life. Its management, either conservative through an orthotic brace system [37, 38] or surgical with different techniques [39], is deferred until at least preschool age or early teenage years, respectively.

9.4 Costal Anomalies

Costal anomalies represent the 3.2% of thoracic wall malformations. They are divided into simple and complex.

Simple malformations involve one or two ribs, up to three nonconsecutive ones, with isolated malformations. The effects on the thoracic cage are limited.

Complex malformations compromise large sections of thorax affecting the respiratory dynamic.

Ulterior division is shown in Table 9.2.

Table 9.2 Classification and frequency of costal anomalies

Costal anomalies	Simple	Unique (45.3%)	Agenesis Hypoplasia Supernumerary Bifid Fusion Dysmorphic
		Double (2.6%)	
		Combined (10.7%)	
	Complex	Strange (5.5%)	
		Fusions (29.4%)	
		Syndromic (6.5%)	Jeune Jarcho-Levin Cerebrocostomandibular Others

9.4.1 Simple Costal Anomalies

- *Agenesis and costal hypoplasia*: They are rare isolated malformations or, more frequently, part of a syndrome (Poland, trisomy 13, cerebrocostomandibular). Patients are asymptomatic, and the management is conservative with radiological and clinical evaluation during growth.
- *Supernumerary cost*: It identifies the presence of more than 24 ribs. The extra rib usually corresponds to the cervical vertebra, and it is rudimentary, unilateral, or bilateral. It is uncommon and can be part of a syndrome. Most patients (70%) are asymptomatic. In symptomatic cases, signs and symptoms do not depend on cost size and can be related to vascular or nervous involvement (pain for arterial spasms and paresthesias). The physical examination shows a tumor in the supraclavicular gap. The thorax X-ray confirms the diagnosis. Asymptomatic patients do not require treatments. Surgery is performed in case of symptoms and consists of resection of the extra rib.
- *Bifid cost*: Bifurcation of the distal end of a rib. It is uncommon and usually isolated. In most cases it is a radiological finding. Sometimes there is a tumor of the costal wall ad pain in the deformed area caused by cartilaginous deformity. Thorax X-ray permits the diagnosis. Surgery is reserved for symptomatic patients (pain) or for esthetic reasons, and it consists of excision of the bifid cost together with the altered cartilage.
- *Costal fusion*: It is characterized by the union of two ribs and it is usually an incidental finding. Thorax X-ray shows the ribs involved and the level of the fusion. When necessary the evaluation is completed with CT (Fig. 9.5). In the evaluation of patients, it is important to detect the presence of malformations, the vertebral fractures, and the degree of scoliosis. The need for surgery is established on the base of the curvature progression.
- *Dysmorphic cost*: Alteration of the costal morphology with widening of the anterior end of the costal arch, a spur, or an irregularity of the entire costal length. Patients present with a small tumor on the anterior or lateral thoracic wall with localized pain (Fig. 9.5).

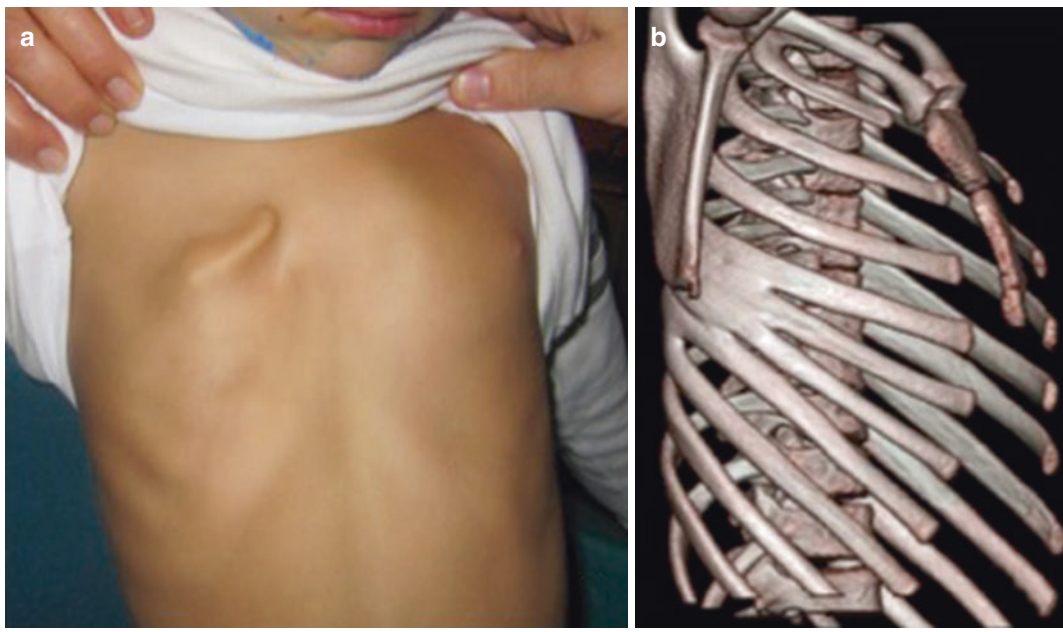

Fig. 9.5 Example of costal anomalies: patient with a dysmorphic cost (**a**) and 3D CT reconstruction of costal fusions (**b**) [*Courtesy by Dr. Michele Torre, G. Gaslini Children's Hospital, Genoa, Italy*]

9.4.2 Double Costal Anomalies

Double malformations include two unique malformations homolateral or contralateral. In case of agenesis of two ribs, there is a paradoxical mechanism during breathing with symptoms onset. Clinical evaluation together with thorax X-ray during growth spur is important during follow-up. Surgery consists of rib spit and placement of autologous costal graft/prosthetic mesh and muscular flap.

9.4.3 Complex Costal Anomalies

– *Strange malformations*: Malformations that do not follow a specific pattern are defined strange. They are extremely rare and each patient corresponds to a unique case. We can have costal hypoplasia, fusion, or intercostal space widening with lung hernia. The right side is affected more than the left one. Hydrocephalus and myelomeningocele are identified in 55% of cases. Clinical manifestations depend on the extent of the deformity. Children with severe deformity change their anatomical thoracic configuration with risk of constrictive thoracic disease. The involvement of cardiopulmonary function is always possible. Neonates may have respiratory distress requiring mechanical ventilation. After this period, symptoms arise during breast-feeding, and they are related to the presence of pulmonary sequelae (dysplasia). A small number of patients are asymptomatic, but the thorax is always abnormal. Thorax X-ray identifies the degree of parietal alteration. The spine should be investigated, as well. When required, surgery consists of costal block removal.
– *Costal fusion*: Costal fusion is a congenital malformation characterized by the union of three or more ribs in any spinal segment. It may cause varying degrees of deformity on the chest wall or spine. It is rare and associated to vertebral anomalies in 90% of cases. The thoracic deformity may be evident soon after birth. Restrictive respiratory symptoms are typical of older patients. Thorax X-ray permits to identify location and features of the malformation. It is important to evaluate the spine praecox. Surgery is indicated in case of severe scoliosis or other deformities.

9.4.4 Syndromic Costal Anomalies

In this group the costal anomaly is part of a specific syndrome.

9.4.4.1 Jeune Syndrome

It is also known as asphyxiating thoracic dystrophy, a congenital malformation (autosomal recessive inheritance) with small thorax, short limbs, and pelvic dysplasia. Fortunately many children do not develop respiratory asphyxia and survive the neonatal period.

The incidence is 1/100.000–130.000 without sexual preponderance.

Patients present with narrow bell-shaped thorax and prominent abdomen (Fig. 9.6); ribs are short, and costochondral junctions do not pass the anterior axillary lines. Costal cartilages have a rosary shape and clavicles are fix and horizontals. This reduces the respiratory movements and leads to an abdominal/diaphragmatic respiration. Hypoventilation causes hypoxia and respiratory distress. The severity is variable and sometimes it causes neonatal demise.

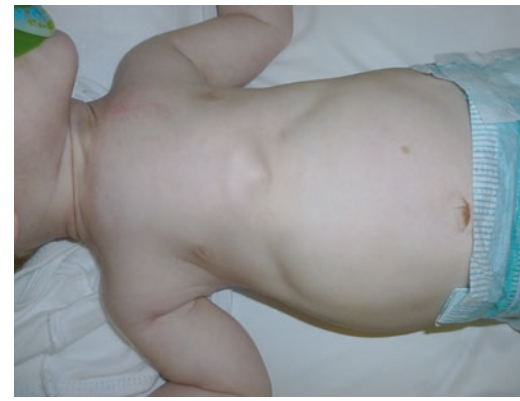

Fig. 9.6 Neonate with Jeune syndrome [*Courtesy by Dr. Michele Torre, G. Gaslini Children's Hospital, Genoa, Italy*]

Renal and hepatic alterations are common: progressive nephropathy from the second year of age due to glomerular sclerosis or tubular cystic dysplasia and hepatic fibrosis or cirrhosis for ductal anomalies. Pancreatic cysts are rare. The pelvis is small and radiological abnormalities tend to decrease during growth. In neonates there is a praecox ossification of femoral head and other bones resulting in growth retardation. Congenital heart disease, retinal degeneration, and dolichocephaly are less frequent associations.

Two forms are identified based on the clinical picture.

Type I, Major Form (70%)

The major form includes patients with small, rigid, and narrow thorax and abdominal respiration. In these cases, respiratory symptoms have early onset (severe neonatal distress), and patients require mechanical ventilation soon after birth. Mortality is high in the first year of age. Together with ventilator support, patients need surgery to enlarge the thorax increasing the lung capacity. The correction should be praecox to avoid long mechanical support and improve survival. Primary and stage repairs have been described starting from the first months of life. Among the surgical options, there is the median sternotomy with bone graft or prosthetic patch interposition. The aim is to enlarge the thoracic cavity with median sternotomy and to keep the two sternal segments open with different rigid material. The sternotomy is performed in the neonatal period and the defect is closed with a prosthetic patch. Once patients are stable (infancy), the patch is replaced by homologous grafts. Grafts include methyl methacrylate and bone grafts. Titanium patches have been used for staged procedures. Vertical expandable titanium rib (VEPTR) improves chest wall movements: they are attached to the ribs and to transverse spinal processes and progressively lengthened. The procedure may lead to scoliosis and patients require long-term follow-up (Fig. 9.7).

Despite the immediate relief from symptoms, surgery doesn't seem to affect long-term results and mortality. The only improvement is prenatal ultrasound diagnosis that allows for consultation and leads to an increment of pregnancy termination.

Type II, Minor Form (30%)

In these patients costal malformations are intermediate and symptoms are limited or absent. Radiological abnormalities tend to reduce over time. Physical examination is sufficient to suspect the syndrome. Thorax X-ray shows the small and narrow thoracic cage with bell-shaped configuration. Ribs and clavicles are horizontals. The

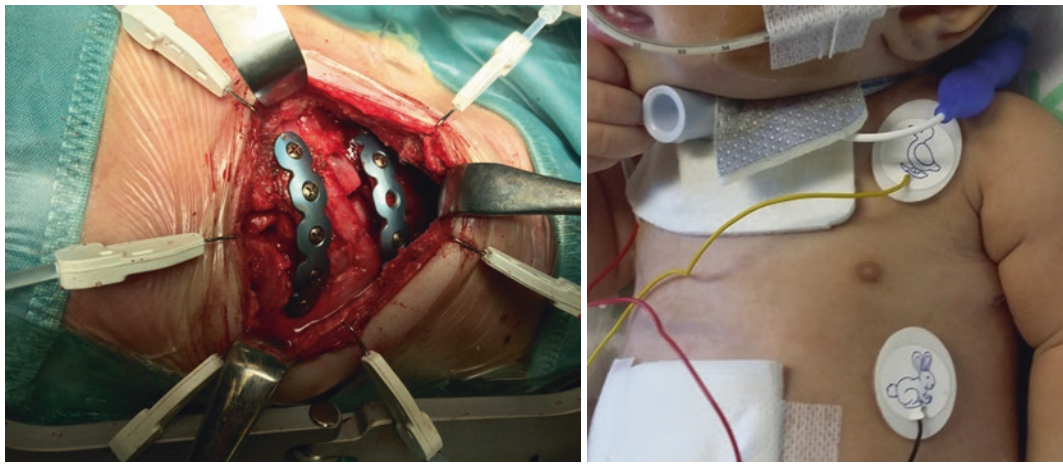

Fig. 9.7 Jeune syndrome: intra- and postoperative pictures [*Courtesy by Dr. Michele Torre, G. Gaslini Children's Hospital, Genoa, Italy*]

latter are shaped as bicycle handlebars and are high. These alterations are well detected on CT scans with 3D reconstruction. Pelvis X-rays show small pelvis and irregular acetabular roof (trident). Iliac wings are small and squared.

Some authors associate the measure of the thoracic perimeter with prognosis: a perimeter <28 cm correlates with bad prognosis. The measure of the thoracic cage might be responsible for pulmonary hypoplasia detected in some patients. The clinical spectrum runs from mild to severe forms where the rigidity of the chest does not allow proper expansion.

Type 2 form does not always require treatments being patients asymptomatic or mildly affected.

9.4.4.2 Jarcho-Levin Syndrome

It is called costovertebral dysplasia or hemivertebral syndrome, and it is characterized by a short thorax with vertebral and costal abnormalities. The inheritance is autosomal recessive with mild male preponderance.

The hemivertebra typically involves all or almost all the spine with fusion or absent vertebral bodies. Vertebral anomalies deform the thoracic cage and lead the posterior costal arches to be fused at the costovertebral junction. Shape and number of ribs are abnormal. The effect is a short and crab-shaped thorax.

Neonates have respiratory distress that progress into respiratory failure, and most of them die within the first year of age. The association with congenital heart disease and urological abnormalities (hydronephrosis, ureteral and urethral stenosis) is frequent. Rarely there are gastrointestinal malformations. Thorax X-ray shows the crab shape due to the short dorsolumbar column. There hasn't been any attempt of surgical correction to date.

9.4.4.3 Cerebrocostomandibular Syndrome

This syndrome is also called the one with segmented rib. It is rare and characterized by microcephaly, micrognathia, and costal anomalies. The type of inheritance has not been defined yet, but it is responsible for the altered cartilage development. There are only costal vestiges. The typical costal anomaly is the aplastic segment at the posterior costal arch with fibrosis, muscular elements, and calcifications. The extension of the costal defect is variable as it is variable to the number of affected ribs. The thorax is short and flat, and the deformity worsens the glossoptosis and the tracheal cartilage hypoplasia. The effect is a severe respiratory distress. Forty percent of patients die within the first months of life. Between the survivors, 50% have moderate mental retardation. Described associations are vertebral malformations, scoliosis, feet deformities, and hip dislocation.

9.4.4.4 Others

Rare type II congenital thoracic deformities include deformities that have been described but do not fit in any standard classification. They have different features and require personalized treatment and management.

9.5 Poland Syndrome

Named after Sir Alfred Poland who described it in 1841 [40], Poland syndrome (PS) is a rare congenital anomaly, occurring in 1:20,000–30,000 live births. Its main diagnostic criterion is the hypoplasia or agenesis of the pectoralis major muscle, although its phenotype can be extremely variable and frequently combined with other ipsilateral abnormalities of the chest wall, breast, and upper limb. PS is almost always unilateral, right-sided in two-thirds of cases; very rare bilateral cases have been described [41]. There is a male preponderance with a 2:1 male to female ratio. It is mainly sporadic, with around 4% of familial cases described [42].

9.5.1 Etiopathogenesis

The etiology of PS is unknown. The most accredited hypothesis regards a possible interruption of the vascular supply in subclavian and vertebral arteries during embryonic life, determined by both genetic and environmental factors, leading

to different malformations of the corresponding districts [43]. The occurrence of familial cases has raised the hypothesis of a possible genetic etiology with different inheritance patterns, although a specific gene has not been identified yet [44].

9.5.2 Clinical Presentation and Assessment

PS phenotype is extremely variable. Partial or total deficit of the pectoralis major muscle is present in 100% of patients, and the thoracic defect is usually evident at birth (Fig. 9.8a) [10].

Other clinical features are [45]:

- Anomalies of other chest wall muscles: Pectoralis minor results to be affected in more than 90% of cases in some series; less frequently an involvement of serratus anterior, latissimus dorsi, trapezius muscle, rhomboid muscle, and rectus abdominis can also be detected.
- Chest wall anomalies: Rib dysmorphisms, hypoplasia, and agenesis and anomalies like a PE or PC or both can occur; in case of rib agenesis, particularly if multiple (most frequently the third and the fourth ribs), lung herniation and paradoxical respiratory movements can be present.
- Breast hypoplasia or aplasia: Breast involvement regards the majority of PS patients, particularly significant in females, and ranges

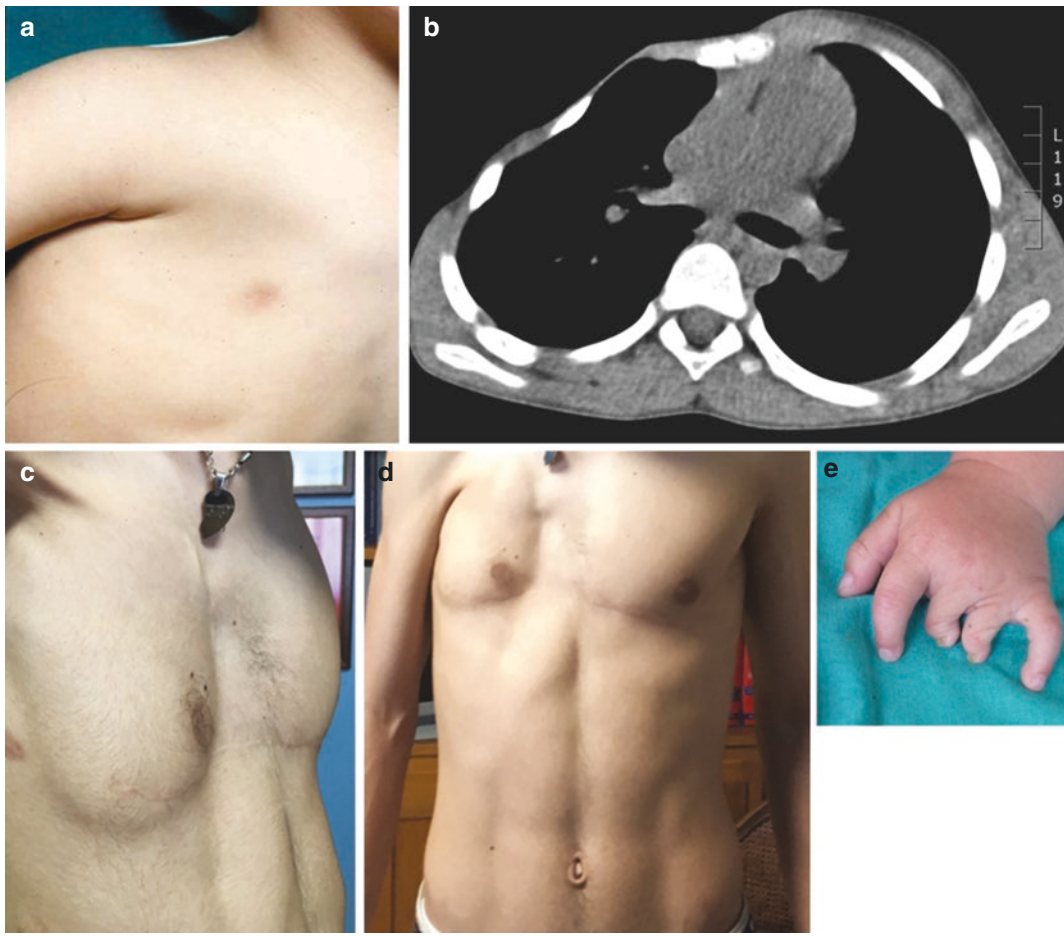

Fig. 9.8 Poland syndrome: chest wall asymmetry in a patient with right form (**a**); CT picture of a right PS (**b**); postoperative pictures of the same patient at long-term follow-up (**c**, **d**); brachydactyly in PS (**e**)

from varying degrees of breast hypoplasia to complete absence of the breast (amastia) and nipple (athelia), usually with hypoplasia of the subcutaneous tissue of the region (Fig. 9.8b).
- Upper limb anomalies [46, 47]: Shortness of the fingers (brachydactyly) (Fig. 9.8e), joined fingers (syndactyly), and a combination of both (brachysyndactyly) are frequently seen hand anomalies, although others can be present, affecting more than half of PS patients.
- Other skeletal deformities: Scoliosis and other spine deformities, Sprengel deformity (congenital elevated small scapula), etc.
- Cardiac/renal anomalies: They are uncommon and usually mild. Dextroposition, always reported in cases of left PS with rib anomalies, seems to be caused by mechanical factors during embryonic life in patients with multiple left rib agenesis [48].
- Other syndromic conditions, such as Moebius and Klippel-Feil syndromes, have been reported in association with PS [49].

The extreme variability of the phenotype imposes a multidisciplinary approach to all children with PS. Diagnostic work-up includes the evaluation of all the organs and systems that can be affected and should be completed as early as possible. Conversely, surgical correction is almost never necessary in the first years of life; nevertheless, since it may require multiple procedures and stages over the years [50] (Fig. 9.8c, d) (hand surgery, restoration of the structural integrity of the rib cage, improvement in the appearance of the chest, breast implants, etc.), a correct information and presentation of all the surgical possibilities should be early offered to the families.

9.6 Sternal Anomalies

Sternal defects are rare and include relatively benign anomalies, such as the partial sternal cleft, and major defects resulting in ectopia cordis. Four main defects can be identified: cervical ectopia cordis, thoracic ectopia cordis, thoracoabdominal ectopia cordis, and cleft or bifid sternum. The heart is displaced in ectopia cordis [51].

9.6.1 Thoracic Ectopia Cordis

Thoracic ectopia cordis presents with naked heart (entirely bare heart outside the thorax) and no overlying somatic structures (pericardium, skin, etc.). The heart has an anterior superior apex. Intrinsic cardiac anomalies are frequent. Upper abdominal wall defects (omphalocele, diastasis recti, eventration of the abdominal viscera) might be associated, as well. The sternum may be partially or completely split with the heart protruding through the defect. This condition differs from ectopia cordis in which the heart is in an orthotopic intrathoracic position, has a normal anatomy, and is covered by normal skin.

The lack of midline somatic tissues and the presence of a small intrathoracic cavity make the repair difficult with risk of heart failure. The correction (primary or stage repair) should be performed early in the first days of life. Different surgical approaches have been reported including the use of skin flaps, prosthetic meshes, rib grafts, and pectoral muscle flaps. Diaphragmatic mobilization and pericardial division from the anterior attachments of the chest wall might help in the closure. In all successful cases, the creation of a partial anterior cavity surrounding the heart avoids heart failure. The presence of intrinsic cardiac lesions and associated abdominal defects severely affects the prognosis, more than the surgical technique chosen for the correction. Postoperative complications include infection and extrusion of the graft.

9.6.2 Cervical Ectopia Cordis

Cervical ectopia cordis is distinguished from the thoracic ectopia cordis on the base of the superior heart displacement. The heart protrudes at the base of the neck, and it is often fused with the mouth, and there are many craniofacial

anomalies or other fetal deformities. The prognosis is bad and surgical repair is very difficult.

9.6.3 Thoracoabdominal Ectopia Cordis

Thoracoabdominal ectopia cordis presents with heart covered by thin and pigmented skin. It is associated with sternal cleft. There is no severe anterior heart rotation seen in thoracic ectopia cordis, but the heart is displaced within the thorax with diaphragmatic and pericardial defect below it or within the abdomen. Described associations are somatic defects, diaphragmatic anomalies, intrinsic cardiac malformations, and abdominal wall defects (omphalocele, diastasis recti). This ectopia cordis is often part of the Cantrell pentalogy (a cleft lower sternum, a half-moon anterior diaphragmatic defect due to failure of development of the septum transversum, absence of the parietal pericardium, adjacent or completely separate omphalocele, ventral hernia or diastasis recti, and in most patients a major form of congenital heart disease, most commonly tetralogy of Fallot or diverticulum from the left ventricle). Surgical correction is possible, and long-term survival rate is more frequent than that of other ectopia cordis. The first step is skin closure to avoid infections and mediastinitis and omphalocele excision. The correction is required early with primary closure or prosthetic mesh closure. The rectus muscles are distant with difficulties in obtaining a good primary closure.

9.6.4 Sternal Cleft or Bifid Sternum

The sternal cleft (SC) is caused by a defect in the fusion process of mesenchymal cells that starts at around the 6th week of gestation. The effect is an anomaly in which the heart is in the orthotopic position in the thoracic cavity but the sternum is clef or partially fused over the heart (Fig. 9.9). The skin coverage is normal and the pericardium is intact. The condition is idiopathic and accounts for 0.15% of all chest deformities with a female preponderance (Table 9.3).

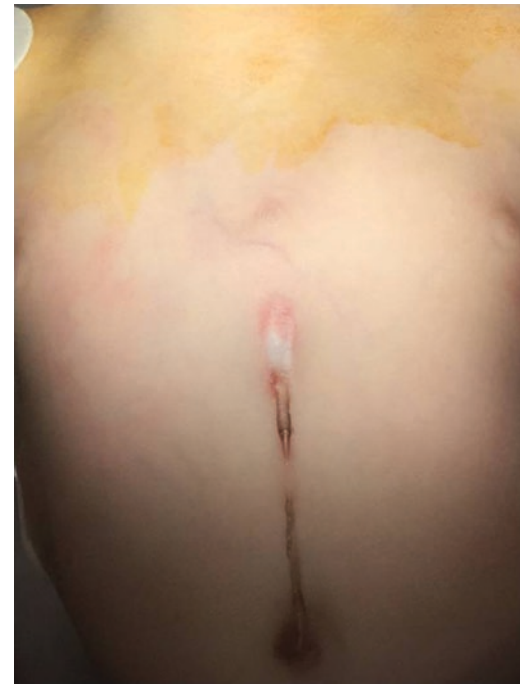

Fig. 9.9 Newborn with sternal cleft

Table 9.3 Sternal cleft types and frequency

Sternal cleft types	
Partial superior form	67%
Complete form	19.5%
Partial inferior form	11%
Sternal foramen	2.5%

Sternal clefts are classified into complete or partial [52]. The partial form can be superior or inferior: the partial superior SC is usually isolated and relatively easy to repair; the partial inferior SC is often associated with complete sternal fissure resulting in ectopia cordis. Other associated defects are vascular dysplasia, PHACES syndrome, midline fusion defects, and pentalogy of Cantrell (heart, pericardium, diaphragm, anterior abdominal wall defects) (Fig. 9.10).

PHACE(S) syndrome is a neurocutaneous disorder of unknown etiology. The acronym refers to the commonest features of PHACE: posterior fossa malformations, large facial hemangiomas, cerebral arterial anomalies, cardiovascular anomalies, and eye anomalies. When ventral developmental defects such as sternal clefting or

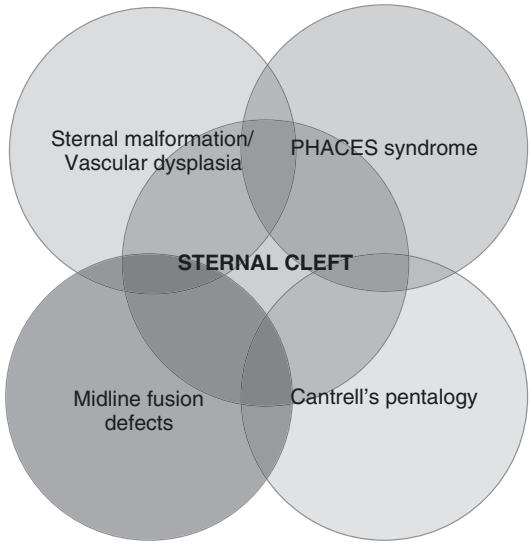

Fig. 9.10 The relationship between sternal cleft and other rare syndromes/associations (*From Torre et al. Phenotypic spectrum and management of sternal cleft: literature review and presentation of a new series. Eur J Cardiothorac Surg. 2012;41(1):4–9*)

supraumbilical raphe occur, the PHACES acronym may be used. The hallmark feature of PHACE is the presence of one or more large facial infantile hemangiomas that occupy at least one facial segment. Cerebral vascular anomalies are probably the most common extracutaneous feature. Given that several organ systems are involved, a multidisciplinary approach to disease surveillance and treatment is advised. In particular the assessment of affected babies should include the endoscopic evaluation of upper airways and cerebral MRI.

Intrinsic cardiac defects are rare. The presence of a fusion defect in the middle part of the sternum is called sternal foramen. SC determines paradox movements during respiration with risk of mediastinal viscera damage. Therefore the correction in neonatal age should be recommended. The difficulty in repairing the defect is related to the fact that the chest is small and cannot accommodate the heart with the risk of cardiovascular failure.

Some neonates are asymptomatic but present a paradoxical midline thoracic bulging due to the protrusion of viscera during expiration.

Older patients present respiratory symptoms such as dyspnea, cough, respiratory distress, and recurrent respiratory tract infections. Asymptomatic patients usually have partial defects and require repair to provide protective coverage for the heart (high risk of trauma-related injuries).

Several associations are seen, the most common being the bandlike scars from the umbilicus to the inferior part of the defect (supraumbilical raphe) that represent only a cosmetic anomaly, cardiac defects (22%) and vascular anomalies (9%).

Early repair in infancy is important because the chest is more flexible and primary closure is the preferred treatment. The primary closure can be challenging or impossible due to a stiff thorax. Alternatives include the use of prostheses (prosthetic materials such as Gore-Tex or Gore, DualMesh, calcium phosphate cement (Fig. 9.11), polyester, or various autologous grafts such as bone graft interposition, muscle flap interposition), partial or total thymectomy [53], cartilage resection, sliding chondrotomies, and clavicle dislocation.

In most series the primary repair is performed within the 3rd month of life. The first year of age is considered a favorable age for repair.

The preoperative evaluation should exclude associated anomalies that can lead to major complications and should define the thoracic anatomy. Some anomalies are evident (maxillofacial hemangiomas, cleft lip or palate, pectus excavatum, precordial skin tags, supraumbilical raphe, gastroschisis, connectival nevus of the anterior thoracic wall). Other defects (cardiac anomalies, aortic coarctation, eye abnormalities, posterior fossa anomalies, hidden hemangiomas) require investigations. Preoperative examinations include the study of the chest with X-ray and CT (Fig. 9.12) and cardiologic evaluation (electrocardiogram and echocardiography). Neuroradiologic imaging and ophthalmologic exams are performed in selected cases. Genetic evaluation completes the screening. Prior to surgery, laryngo-tracheo-bronchoscopy might identify subglottic hemangiomas.

For surgery, the midline vertical incision extends from the jugular notch to the end of the defect (Fig. 9.11). The dissection proceeds down-

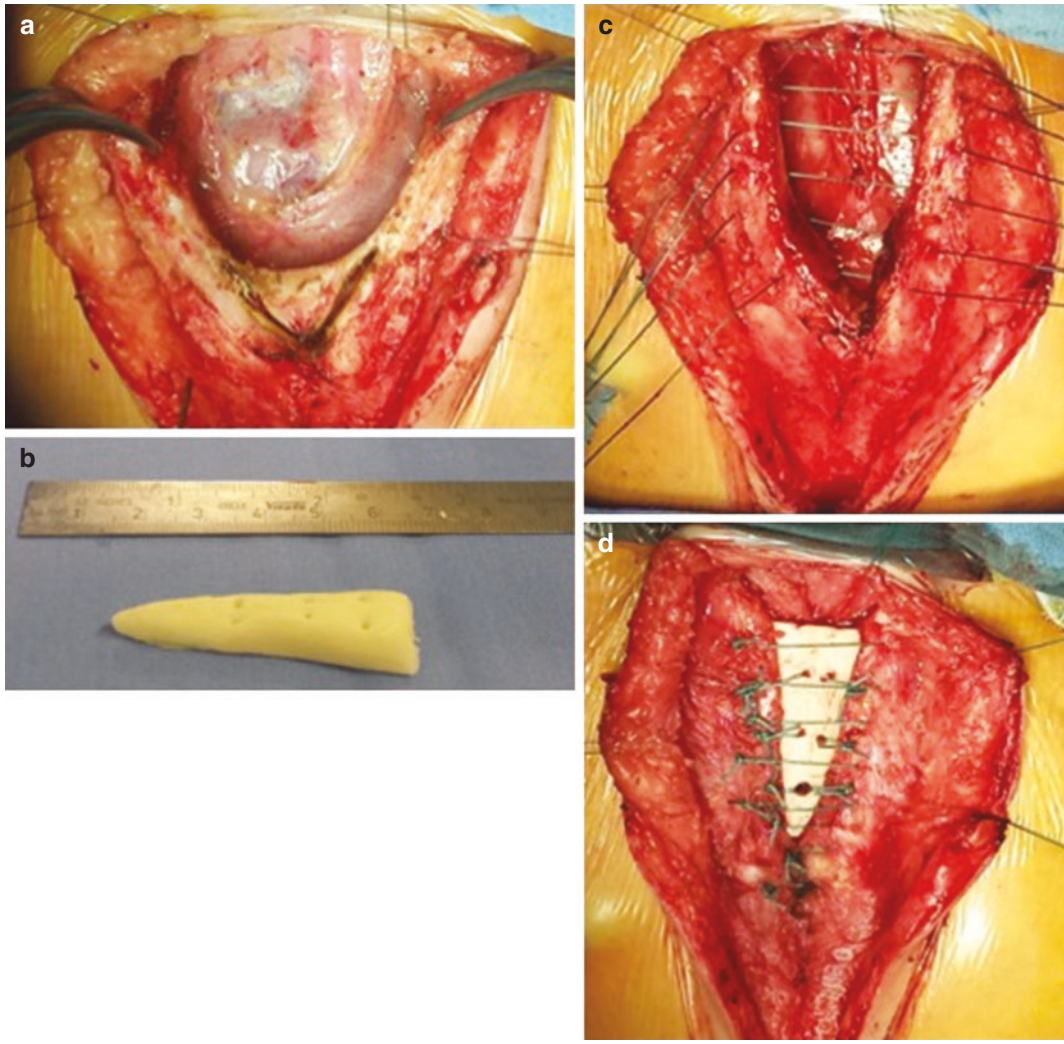

Fig. 9.11 Sternal cleft (**a**) repair (**c**, **d**) with a prosthesis in calcium phosphate cement (**b**)

ward to expose the sternal bars. Vertical strap neck muscles are divided at their insertion on the sternal upper margins and the two sternal halves are freed from the underlying pleura and pericardium. A U-shaped incision is performed in the inferior part of partial SC to facilitate the closure. The sternal bars are approximated on the midline and closed with nonabsorbable sutures when possible (Fig. 9.13).

Reported complications are pericardial or pleural tears during sternal dissection, retrosternal seromas, and pneumothorax. In females, care should be taken not to injure the mammary gland. Patients with severe associated anomalies have unfavorable events related to the underlying disease. No recurrences have been described.

The required follow-up is a close one since patients can develop other congenital wall malformations (e.g., pectus excavatum).

9.7 Clavicle-Scapular Anomalies

Clavicle-scapular malformations represent 0.5% of all the CWM. They are divided into malformations of the clavicle, malformations of the scapula, and combined malformations.

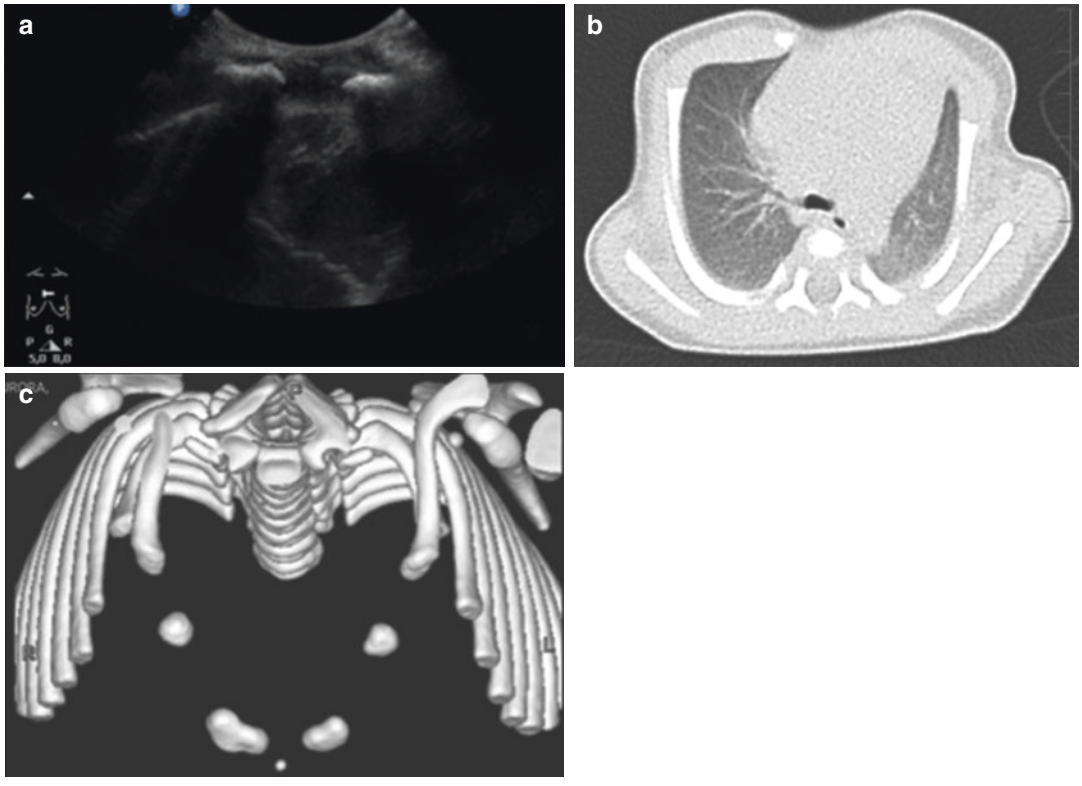

Fig. 9.12 (**a**) US showing "V-shaped" sternum with proximal diastasis; (**b**, **c**) CT evaluation of the sternal deformity: diastasis of the middle and proximal portion of the sternum, costal hypoplasia

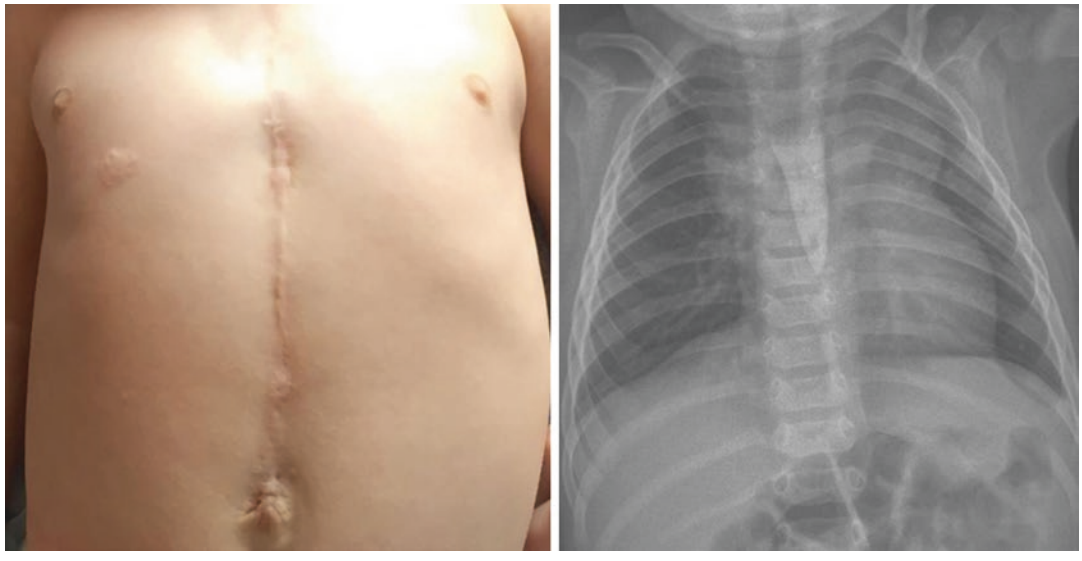

Fig. 9.13 Postoperative pictures after sternal cleft repair with a prosthesis in calcium phosphate cement

9.7.1 Clavicular Malformations

They originate from alterations in the growth or structure of the clavicle that can be aplastic, hypoplastic, or dysmorphic, on one or both sides. Clavicular malformations can be isolated (simple) or part of a syndromic association. They usually cause very little functional alteration, except when associated with other malformations of the scapula and/or upper ribs. Several syndromes, such as pseudohypoparathyroidism (Albright), Holt-Oram, and Pierre-Marie syndrome, among others, can present clavicular abnormalities as part of their clinical picture, usually associated with other skeletal deformities. The chest of these children is narrow in its upper part, with decreased lateral diameter. Functionally, this facilitates the realization of atypical movements, such as the approximation of the shoulders in front of the thorax. The muscles of the area can also be involved, presenting developmental and insertional anomalies. In the neonatal period, respiratory disorders can appear due to the narrowness of the chest. Chest X-ray shows the absence or hypoplasia of one or both clavicles. Clavicular malformations can be associated with pectus carinatum or excavatum.

9.7.2 Scapular Malformations

These anomalies are characterized by alterations in development and position of the scapula with variable joint mobility. The deformities occur at birth and their severity is variable; they are usually unilateral. They are classified as simple, with hypoplastic or winged scapula, or syndromic. Syndromes that favors scapular winging are, for example, muscular dystrophies or other conditions with muscular hypoplasia such as Poland syndrome. It can be associated with PE, PC, scoliosis, vertebral, and rib deformities.

Sprengel deformity is characterized by an elevated anomalous position of one or both scapulae, with variable disorders of joint mobility. The deformity is the result of a failure in the descent of the scapula from its fetal position in the neck to its normal position on the posteroexternal face of the thoracic wall. It is more common in women (4:1 ratio). It occurs sporadically, but an apparent pattern of autosomal dominant inheritance has been sometimes noted. The affected scapula is hypoplastic, with decreased vertical diameter and apparent increase of its horizontal width. The muscles connected to the scapula can be altered, most commonly the trapezoid, the deltoid, the rhomboid, the elevator of the scapula, and the serratus anterior, so that the amplitude of shoulder movements may be limited.

Scapular malformations can also be part of the Klippel-Feil syndrome, in association with anomalies of the cervical spine, a characteristic short neck with movement limitations, dorsal and lumbosacral vertebral malformations, anomalies of the ribs, malformations of the upper and lower limbs, and rarely cardiovascular malformations.

Chest X-ray in front and profile projections and CT with 3D reconstruction of the chest wall are the imaging techniques of choice. MRI is carried out when intramedullary alterations are suspected. Differential diagnosis includes other causes of shoulder asymmetry, such as scoliosis, asymmetry of lower limbs, and obstetric paralysis of the brachial plexus.

The goals of surgical treatment are to improve the function and the esthetic appearance of the shoulder. When indicated, surgery is recommended between 9 and 18 months of life.

9.7.3 Combined Malformations

They are the most frequent variant and are characterized by the association of different types both clavicular and scapular defects. They can be unilateral or bilateral, and they comprise the simple subtypes of each group of malformations.

9.8 Associated Spinal Deformities

The normal development of the vertebral column includes the longitudinal and axial growth of the vertebrae [54] that requires:

1. The skeletal growth (the growth of the axis for muscular development).
2. The symmetrical growth of the spine in the longitudinal and axial plane, preventing the development of deformities (scoliosis). The scoliosis is the spinal lateral deviation of more than 10° with vertebral body rotation. According to their origin, the scoliosis can be classified into idiopathic, congenital, neuromuscular, osteopathic, and neoplastic.
3. Protection of neural elements, ensuring that the child reaches adulthood without neurological complications.
4. The development of physiological spinal curvatures, which allow proper balance in the standing position.

The spine is intimately associated with the thoracic cage and any alteration of these two elements is reflected in the other. Under normal conditions, the development of the thorax is a dynamic process that involves the sternum, the ribs, the spine, and the diaphragm. Its growth shows a higher velocity in the first years of life. Everything that interferes with the thoracic normal development will reduce the respiratory capacity, especially in younger ages.

Also the thoracic morphology changes with growth: the thorax is cylindrical with horizontal ribs at birth; it has an oval shape, with an oblique orientation of the ribs at 2 years, and it is rectangular at 10 years. It has been established that the growth of 50% of the thoracic volume happens in the first 10 years and the other 50% is completed at the age of 16. An alteration in the longitudinal or anteroposterior growth of the thorax, thoracic volume, and alveoli will produce respiratory failure syndrome.

Spinal and thoracic deformities can be divided as:

1. Primary
   (a) Anterior deformities (pectus excavatum, carinatum, etc.)
   (b) Connective tissue deformities (Marfan)
   (c) Costal fusion and strange malformations
2. Secondary
   (a) Of the wall: costal resections, costal fusions
   (b) Of the content: retraction after empyema, pulmonary resections, diaphragmatic palsy

Hemivertebra is a type of vertebral anomaly and results from a lack of formation of one half of a vertebral body. It can be a common cause of a congenital scoliosis. The curve progression and the ultimate severity of the curve depend on the type of hemivertebrae, the location, the number of hemivertebrae, and their relationship with each other. It falls under the spectrum of segmentational anomalies and can involve one or multiple levels. Recognized associations are many and include Aicardi syndrome, cleidocranial dysostosis, gastroschisis, Gorlin syndrome, fetal pyelectasis, Jarcho-Levin syndrome, OEIS complex, VACTERL association, and mucopolysaccharidosis.

References

1. Acastello E. Patologías de la pared torácica en pediatría. Buenos Aires: Edimed; 2011.
2. Al-Qattan MM. Classification of hand anomalies in Poland's syndrome. Br J Plast Surg. 2001;54(2):132–6.
3. Baban A, Torre M, Bianca S, Buluggiu A, Rossello MI, Calevo MG, Valle M, Ravazzolo R, Jasonni V, Lerone M. Poland syndrome with bilateral features: case description with review of the literature. Am J Med Genet A. 2009;149A(7):1597–602. https://doi.org/10.1002/ajmg.a.32922.
4. Baban A, Torre M, Costanzo S, Gimelli S, Bianca S, Divizia MT, Sénès FM, Garavelli L, Rivieri F, Lerone M, Valle M, Ravazzolo R, Calevo MG. Familial Poland anomaly revisited. Am J Med Genet A. 2012;158A:140.
5. Bavinck JNB, Weaver DD. Subclavian artery supply disruption sequence: hypothesis of a vascular etiology for Poland, Klippel-Feil and Mobius anomalies. Am J Med Genet. 1986;23(4):903–18.
6. Berdel AL, Henrich W. Antenatal sonographic features of Poland syndrome on 2- and 3-dimensional sonography. J Ultrasound Med. 2010;29(4):679–80.
7. Blanco FC, Elliott ST, Sandler AD. Management of congenital chest wall deformities. Semin Plast Surg. 2011;25(1):107–16.
8. Breysem L, Smet MH, Van Lierde S, Devlieger H, De Boeck K. Bronchopulmonary dysplasia: correlation of radiographic and clinical findings. Pediatr Radiol. 1997;27(8):642–6.

9. Cartoski MJ, Nuss D, Goretsky MJ, Proud VK, Croitoru DP, Gustin T, Mitchell K, Vasser E, Kelly RE Jr. Classification of the dysmorphology of pectus excavatum. J Pediatr Surg. 2006;41(9):1573–81.
10. Catena N, Divizia MT, Calevo MG, Baban A, Torre M, Ravazzolo R, Lerone M, Sénès FM. Hand and upper limb anomalies in Poland syndrome: a new proposal of classification. J Pediatr Orthop. 2012;32(7):727–31.
11. Coln E, Carrasco J, Coln D. Demonstrating relief of cardiac compression with the Nuss minimally invasive repair for pectus excavatum. J Pediatr Surg. 2006;41(4):683–6.
12. Colombani P. Preoperative assessment of chest wall deformities. Semin Thorac Cardiovasc Surg. 2009;21(1):58–63.
13. Creswick HA, Stacey MW, Kelly RE Jr, Gustin T, Nuss D, Harvey H, Goretsky MJ, Vasser E, Welch JC, Mitchell K, Proud VK. Family study of the inheritance of pectus excavatum. J Pediatr Surg. 2006;41(10):1699–703.
14. Currarino G, Silverman FN. Premature obliteration of the sternal sutures and pigeon-breast deformity. Radiology. 1958;70(4):532–40.
15. Daltro P, Fricke BL, Kline-Fath BM, Werner H, Rodrigues L, Fazecas T, Domingues R, Donnelly LF. Prenatal MRI of congenital abdominal and chest wall defects. AJR Am J Roentgenol. 2005;184(3):1010–6.
16. Edwards DK 3rd, Hilton SW. Flat chest in chronic bronchopulmonary dysplasia. AJR Am J Roentgenol. 1987;149(6):1213–6.
17. Feng J, Hu T, Liu W, Zhang S, Tang Y, Chen R, Jiang X, Wei F. The biomechanical, morphologic, and histochemical properties of the costal cartilages in children with pectus excavatum. J Pediatr Surg. 2001;36(12):1770–6.
18. Fokin AA, Steuerwald NM, Ahrens WA, Allen KE. Anatomical, histologic, and genetic characteristics of congenital chest wall deformities. Semin Thorac Cardiovasc Surg. 2009;21(1):44–57.
19. Haje SA, Harcke HT, Bowen JR. Growth disturbance of the sternum and pectus deformities: imaging studies and clinical correlation. Pediatr Radiol. 1999;29(5):334–41.
20. Haller JA, Colombani PM, Humphries CT, Azizkhan RG, Loughlin GM. Chest wall constriction after too extensive and too early operations for pectus excavatum. Ann Thorac Surg. 1996;61(6):1618–24.
21. Haller JA, Kramer SS, Lietman SA. Use of CT scans in selection of patients for pectus excavatum surgery: a preliminary report. J Pediatr Surg. 1987;22(10):904–6.
22. Kaplan KM, Spivak JM, Bendo JA. Embryology of the spine and associated congenital abnormalities. Spine J. 2005;5(5):564–76.
23. Kelly RE Jr, Mellins RB, Shamberger RC, Mitchell KK, Lawson ML, Oldham KT, Azizkhan RG, Hebra AV, Nuss D, Goretsky MJ, Sharp RJ, Holcomb GW 3rd, Shim WK, Megison SM, Moss RL, Fecteau AH, Colombani PM, Cooper D, Bagley T, Quinn A, Moskowitz AB, Paulson JF. Multicenter study of pectus excavatum, final report: complications, static/exercise pulmonary function, and anatomic outcomes. J Am Coll Surg. 2013;217(6):1080–9.
24. Kelly RE Jr, Quinn A, Varela P, Redlinger RE Jr, Nuss D. Dysmorphology of chest wall deformities: frequency distribution of subtypes of typical pectus excavatum and rare subtypes. Arch Bronconeumol. 2013;49(5):196–200.
25. Kilda A, Basevicius A, Barauskas V, Lukosevicius S, Ragaisis D. Radiological assessment of children with pectus excavatum. Indian J Pediatr. 2007;74(2):143–7.
26. Kim HC, Choi H, Jin SO, Lee JJ, Nam KW, Kim IY, Nam KC, Park HJ, Lee KH, Kim MG. New computerized indices for quantitative evaluation of depression and asymmetry in patients with chest wall deformities. Artif Organs. 2013;37(8):712–8.
27. Kim M, Lee KY, Park HJ, Kim HY, Kang EY, Oh YW, Seo BK, Je BK, Choi EJ. Development of new cardiac deformity indexes for pectus excavatum on computed tomography: feasibility for pre- and post-operative evaluation. Yonsei Med J. 2009;50(3):385–90.
28. Kolvekar SK, Simon N, Kolvekar T. Diagnosis in chest wall deformities. J Vis Surg. 2016;2:103.
29. Kotzot D, Schwabegger AH. Etiology of chest wall deformities—a genetic review for the treating physician. J Pediatr Surg. 2009;44(10):2004–11.
30. Lawson ML, Mellins RB, Paulson JF, Shamberger RC, Oldham K, Azizkhan RG, Hebra AV, Nuss D, Goretsky MJ, Sharp RJ, Holcomb GW 3rd, Shim WK, Megison SM, Moss RL, Fecteau AH, Colombani PM, Moskowitz AB, Hill J, Kelly RE Jr. Increasing severity of pectus excavatum is associated with reduced pulmonary function. J Pediatr. 2011;159(2):256–61.
31. Lees RF, Caldicott WJH. Sternal anomalies and congenital heart disease. Am J Roentgenol. 1975;124:423–7.
32. Merchant AM, Peranteau W, Wilson RD, Johnson MP, Bebbington MW, Hedrick HL, Flake AW, Adzick NS. Postnatal chest wall deformities after fetal thoracoamniotic shunting for congenital cystic adenomatoid malformation. Fetal Diagn Ther. 2007;22(6):435–9.
33. Nuss D. Minimally invasive surgical repair of pectus excavatum. Semin Pediatr Surg. 2008;17(3):209–17.
34. Nuss D, Kelly RE. Congenital chest wall deformities, Chapter 20. In: Holcomb III GW, Murphy JP, editors. Ashcraft's pediatric surgery. V ed. Philadelphia, PA: Saunders Elsevier; 2010. p. 249–65.
35. Martinez-Ferro M, Fraire C, Bernard S. Dynamic compression system for the correction of pectus carinatum. Semin Pediatr Surg. 2008;17(3):194–200.
36. Obermeyer RJ, Goretsky MJ. Chest wall deformities in pediatric surgery. Surg Clin North Am. 2012;92(3):669–84.
37. Park HJ, Lee IS, Kim KT. Extreme eccentric canal type pectus excavatum: morphological study and repair techniques. Eur J Cardiothorac Surg. 2008;34(1):150–4.
38. Parker DL, Mitchell PR, Holmes GL. Poland-Moebius syndrome. J Med Genet. 1981;18(4):317–20.

39. Poland A. Deficiency of the pectoral muscles. Guy's. Hosp Rep. 1841;6:191–3.
40. Rahmani R, Sterling CL, Bedford HM. Prenatal diagnosis of Jeune-like syndromes with two-dimensional and three-dimensional sonography. J Clin Ultrasound. 2012;40(4):222–6.
41. Redlinger RE Jr, Wootton A, Kelly RE, Nuss D, Goretsky M, Kuhn MA, Obermeyer RJ. Optoelectronic plethysmography demonstrates abrogation of regional chest wall motion dysfunction in patients with pectus excavatum after Nuss repair. J Pediatr Surg. 2012;47(1):160–4.
42. Romanini MV, Torre M, Santi P, Dova L, Valle M, Martinoli C, Baldelli I. Proposal of the TBN classification of thoracic anomalies and treatment algorithm for Poland syndrome. Plast Reconstr Surg. 2016;138(1):50–8.
43. Shamberger RC. Congenital thoracic deformities, Chapter 23. In: Puri P, editor. Newborn surgery. 2nd ed. London: Arnold; 2003. p. 239–46.
44. Shamberger RC, Welch KJ. Surgical correction of pectus carinatum. J Pediatr Surg. 1987;22:48–53.
45. Silbiger JJ, Parikh A. Pectus excavatum: echocardiographic, pathophysiologic, and surgical insights. Echocardiography. 2016;33(8):1239–44.
46. Stephenson JT, Du Bois J. Compressive orthotic brace in the treatment of pectus carinatum: the use of radiographic markers to predict success. J Pediatr Surg. 2008;43(10):1776–80.
47. Torre M, Baban A, Buluggiu A, Costanzo S, Bricco L, Lerone M, Bianca S, Gatti GL, Sénès FM, Valle M, Calevo MG. Dextrocardia in patients with Poland syndrome: phenotypic characterization provides insight into the pathogenesis. J Thorac Cardiovasc Surg. 2010;139(5):1177–82.
48. Torre M, Rapuzzi G, Carlucci M, Pio L, Jasonni V. Phenotypic spectrum and management of sternal cleft: literature review and presentation of a new series. Eur J Cardiothorac Surg. 2012;41(1):4–9.
49. Torre M, Rapuzzi G, Guida E, Costanzo S, Jasonni V. Thymectomy to achieve primary closure of total sternal cleft. J Pediatr Surg. 2008;43(12):e17–20.
50. Torre M, Rapuzzi G, Jasonni V, Varela P. Chest wall deformities: an overview on classification and surgical options. In: Guerreiro Cardoso PF, editor. Topics in thoracic surgery. Rijeka: InTech; 2012.
51. Vaccari CM, Tassano E, Torre M, Gimelli S, Divizia MT, Romanini MV, Bossi S, Musante I, Valle M, Senes F, Catena N, Bedeschi MF, Baban A, Calevo MG, Acquaviva M, Lerone M, Ravazzolo R, Puliti A. Assessment of copy number variations in 120 patients with Poland syndrome. BMC Med Genet. 2016;17(1):89.
52. Williams AM, Crabbe DC. Pectus deformities of the anterior chest wall. Paediatr Respir Rev. 2003;4(3):237–42.
53. Yiyit N, Işıtmangil T, Öksüz S. Clinical analysis of 113 patients with Poland syndrome. Ann Thorac Surg. 2015;99(3):999–1004.
54. Zhao L, Feinberg MS, Gaides M, Ben-Dov I. Why is exercise capacity reduced in subjects with pectus excavatum? J Pediatr. 2000;136(2):163–7.