Malformations of the Chest Wall

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Michele Torre, Giovanni Rapuzzi, Patricio Varela and Vincenzo Jasonni

26.1 Introduction

Chest-wall malformations (CWMs) have a relatively high incidence and an unsuspecting impact on the quality of life of patients. Besides a minority of symptomatic cases, most patients seek medical advice for psychosocial concerns. These concerns can be severe, and are usually due to: poor cosmetic appearance; aversion to undertaking sporting activities: aversion to being seen in public.

CWMs are often misdiagnosed or neglected by physicians, thereby resulting in a significant delay or mistakes in the diagnostic work-up or management. Since the introduction of the Nuss technique for pectus excavatum (PE) [1], however, interest from the scientific community about CWMs has increased dramatically.

CWMs are represented by a wide range of malformations; some are very well defined and others are part of a wide spectrum of deformities. However, the literature relating to their nomenclature and classification is confusing. In our opinion, the classification is of paramount importance because of the treatment implications. Other controversial issues are the treatment options: many surgical techniques or other therapeutic alternatives have been proposed (especially in the last decade), so it can be difficult for a pediatrician (or even a surgeon) to advise patients about the possible correction techniques. In this chapter, we describe the different types of CWMs, their diagnosis and treatment and classify them into four types according to their embryology (Table 26.1).

26.2 Type I: Cartilaginous Anomalies

26.2.1 PE

26.2.1.1 Epidemiology

PE is the most common thoracic malformation, with an incidence of about 1/300 live births, and represents 90% of all CWMs [2, 3]. PE is characterized by the presence of a sternal depression associated with a malformation of the lower condrosternal joints.

26.2.1.2 Etiology and Pathogenesis

In most cases PE occurs during or after the pubertal spurt (though it can occur in a neonate). Older patients can have a association with malformations of muscular connec-

M. Torre (✉)
Department of Pediatric Surgery
G. Gaslini Children's Hospital
Genoa, Italy
e-mail: micheletorre@ospedale-gaslini.ge.it

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Table 26.1 Modified version of the Acastello classification of chest-wall malformations according to their embryologic origin

Type I: cartilaginous	PE PC type 1 True PC type 2	
Type II: costal	Simple (1 or 2 ribs)	Agenesis, hypoplasia, sovrannumerary, bifid, fused, dysmorphic, rare (always complexes)
	Complex (≥3 ribs)	
	Syndromic (always complex)	Jeune, Jarcho-Levin, Cerebrocostomandibular, others
Type III: chondro-costal	PS	
Type IV: sternal	Sternal cleft (with or without ectopia cordis) CS syndrome	

PE, pectus excavatum; *PC*, pectus carinatum; *PS*, Poland syndrome; *CS*, Currarino–Silverman

tive tissue, such as the Marfan and Ehlers–Danlos syndromes [2–6]. The etiology of PE is not clear, and many hypotheses have been proposed [5]. Deficiencies in vitamins or nutrients are probably not involved, but disorders of connective tissue and genetic predisposition could have a role [6]. Overgrowth of costal cartilage could be the pathogenetic mechanism leading to the development of PE [2, 5]. Examination of costal cartilage has demonstrated type-II collagen in PE patients, as well as overexpression or downregulation of collagen genes, matrix metalloproteinases, tumor necrosis factor-alpha, and filamin [2]. Forty percent of cases are familial [5]. Moreover, PE patients can have siblings with other CWMs, such as pectus carinatum (PC). The main transmission pattern seems to be autosomal dominant, but families with autosomal recessive and X-linked patterns have been documented [7].

26.2.1.3 Clinical Features

Often, PE patients are tall and slim, with some degree of joint laxity, a kyphotic habit with rounded shoulders and a "pot belly" [4]. In fact, 15–50% of cases have associated spinal deformities such as scoliosis or kyphoscoliosis. These malformations are often mild and usually do not require treatment. Apart from prolapse of the mitral valve, associated cardiac anomalies are not common [6]. We observed an association with congenital diaphragmatic hernia or airway anomalies in infants and children with respiratory obstruction (e.g., hypetrophic tonsils). PE associated with airway anomaly is the only one that can ameliorate significantly or disappear during infancy [5]. In most cases, PE is mild at birth and worsens over time, progressing and causing symptoms (especially during the pubertal spurt). When the deformity is very deep, patients may manifest pulmonary symptoms (dyspnea, lack of stamina), cardiologic symptoms (palpitations), thoracic pain or psychological discomfort [1–5, 8]. Sternal depression can displace the heart leftward and cause compression of the right-heart chambers with different degrees of dysfunction on echocardiography. The inferior vena cava can also be compressed. Some degree of dysfunction can also be evident on lung function tests, with more dysfunction upon stress conditions than at rest. The most common pattern of PE is restrictive, but obstructive or mixed patterns are not uncommon, whereas asthma induced by exercise is rare. Esthetic factors can affect self-esteem and self-image in most of the patients, who are usually extremely shy and refuse to practice any activity that may involve exposing their chests (e.g., swimming, going to the beach).

Table 26.2 Classification of pectus excavatum according to morphology

- Grand Canyon (Fig. 26.1) refers to a deep and severe excavation with a long canal in the sternum. Thoracoscopic correction is extremely difficult, especially if the thorax is largely ossified and sternum extremely rotated. A higher prevalence of complications after correction is reported compared with the other types. In these cases, modified open procedures can be a valid option for correction.
- Punch or cup shape (Fig. 26.1): PE is localized (usually on the inferior part of the sternum) and is symmetric. In our opinion, correction can be very difficult at any age and sometimes the outcome is partially successful.
- Saucer type (Fig. 26.1) can be symmetric or asymmetric. It is the most common of all PE types and the thorax is usually quite flat, along the complete anterior chest.
- 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 (Fig. 26.1): The main feature of this type of PE (but sometimes this is an isolated malformation without associated PE) is the flaring chest at the level of the final ribs.
- Pectus excavatum-carinatum is a combined malformation with a sunken chest and cartilage protrusion beside the sternal edge.
- Superior PE is very rare and localized to the upper part of the sternum and cartilage ribs. The lower sternum is normal.

26.2.1.4 Diagnosis and Classification

Each PE patient is unique, so it is important to assess the severity to select the best treatment. The depth of the excavation can be measured by calipers [4] or a pulvimeter [2] but the most important index, the Haller index, can be evaluated by computed tomography (CT) [9]. This Haller index is the most widely accepted index, and is based on the division between lateral and anteroposterior thoracic diameters. Index values >3 or 3.25 indicate surgical correction. Another important feature is PE symmetry. Asymmetric excavations (usually more depressed on the right side due to a variable degree of sternal rotation) can compromise or influence the final result. In females with asymmetric PE, the sternum is usually rotated towards the right side and the right breast seems hypoplasic, mimicking the Poland syndrome (PS).

Evaluation of the shape of PE is crucial for determining the type of surgical approach and its prognosis. We can classify PE according to morphology as shown in Table 26.2 [2, 5, 10] and with reference to Figure 26.1.

26.2.1.5 Management

Patients are selected for surgical correction if they meet two or more of the criteria shown in Table 26.3 [5]. The ideal age for correction is controversial [3, 10]. Open and thoracoscopic procedures are feasible and elicit good results [11]. However, it is widely accepted that surgical correction should be done before complete ossification of the thorax. Surgical correction in the first years of life is probably unnecessary and can carry the risk of relapse [10] or severe postoperative complications such as acquired Jeune syndrome [12]. A good age for correction with the Nuss procedure is considered to be 9–15 years of life [10].

The main procedures for PE repair are the Ravitch procedure (and its modifications) and the Nuss procedure. The Ravitch procedure was first described in 1949 [13]. It comprises resection of all deformed costal cartilages. Subsequent modifications preserved the perichondrium to facilitate rib regeneration and fixation of the sternum to reduce the risk of a flail chest and recurrence. Initially, this procedure was done with a bone graft and later with a steel bar passed posterior to the sternum. Many means of sternal fixation have been proposed during the following years, some of them absorbable. In 1970, Wada introduced a totally new concept: he described a method involving a “sternal turnover” in which the sternum was completely detached and removed, rotated by 180°, and then sutured back to the ribs. Another approach, attempted in mild cas-

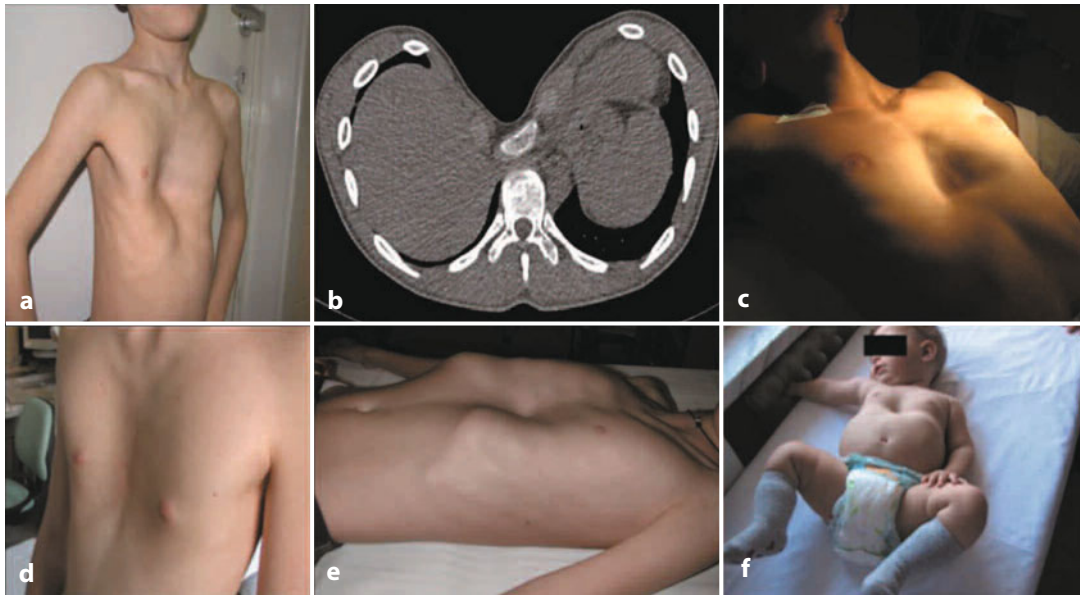

Fig. 26.1 First row: Grand Canyon-shapes PE (**a**). CT of the thorax showing a Grand Canyon-shaped PE with sternal rotation (**b**). Punch-shaped PE (**c**). Saucer-shaped PE (**d**). PE with flaring chest (**e**). Congenital punch-shaped PE (**f**)

Table 26.3 Criteria for surgical correction of pectus excavatum [5]

- symptoms (cardiologic, pulmonary or psychological);
- history of evolution;
- paradoxical movement of the chest wall;
- Haller index >3.25;
- cardiac compression and/or consequent cardiac disease;
- compression of the vena cava or pulmonary veins;
- significant restrictive disease on pulmonary function studies;
- history of failed previous repair;
- severe disturbance of body image.

Patients are selected for surgical correction if they meet two or more of these criteria.

es, and proposed for the first time in 1972 by Standford, was carried out by filling the concavity of PE with prosthetic material (e.g., Silastic®; Dow Corning Corporation) or other subsequent modifications (e.g., omental flap) [14].

In 1997, Nuss and colleagues proposed a revolutionary new method, and published it after 1 year [1]. The method involves implanting a retrosternal metallic bar which is bent and rotated at 180° to obtain immediate correction of the deformity. This metal bar is inserted through small lateral incisions and neither costal resections nor sternotomy are required. The long-term efficacy of the Nuss procedure is shown by the principle of thoracic cage remodeling determined by the retrosternal bar. Hence, the bar should remain for ≥3 years and is subsequently removed through an outpatient procedure. To avoid dislocation, the bar is fixed to the chest-wall muscles and stabilized *via* a lateral device that avoids slippage. Initially, the retrosternal tunnel for the bar was created blindly but, with the advent of thoracoscopy, the tunnel is now created under direct vision. In our opinion, thoracoscopic

aids are crucial because the bar must pass very close to the heart, which can be very closely attached to the sternum. Other, less frequently adopted conservative procedures have been described. They are based on suction devices (vacuum bell) [15] or magnetic forces [16], and attempt to correct PE without surgery, but evidence of efficacy is lacking.

26.2.1.6 Outcome and Follow-up

All major series have reported [3, 8, 10, 17] good results in >80–90% of cases depending on the severity and type of PE and the patient's age at correction. The largest series consisted of 1,215 patients and was reported by Nuss and colleagues [18]. With respect to the procedure, they reported a prevalence of 95.8% for surgeon satisfaction, 93% for patient satisfaction and 92% for parent satisfaction. There is ongoing debate between proponents of the Nuss procedure and proponents of the Ravitch procedure to define which is safer and can guarantee better results. However, the Nuss procedure is used more often because it guarantees minimal access and does not leave anterior scars.

The complications observed in open and minimally invasive procedures are: wound infections; hematomas; bar dislocations; pneumothorax requiring thoracic drainage; transient Horner syndrome; major bleeding; overcorrection/ mild correction [3, 8, 10, 18–19]. Complications specific to an extensive open procedure (particularly at early age) are floating sternum and acquired Jeune syndrome [12]. Complications specific to the Nuss procedure are pericarditis and allergy to nickel (component of the metal bar) [10]. During the Nuss procedure, very few heart lesions and deaths were reported; most of them occurred when thoracoscopy was not used [10, 20]. In case of infection due to the Nuss bar, this can be managed successfully conservatively [21]. Recurrence in 2–5% of cases has been reported [3, 18]. In our experience, in case of suboptimal results, one or more treatments with lipofilling into the residual excavation can improve the final outcome significantly, but published series are lacking. In females, Nuss or open procedures alone can correct the breast aspect but, if some degree of asymmetry persists, breast augmentation may be required, preferably after PE repair [22].

26.2.2 PC

26.2.2.1 Epidemiology

After PE, the second most frequent malformation is PC. The incidence of PC is estimated to be five-times less frequent than that of PE [2, 4] with a major predominance in male patients. PC comprises a protrusion of the sternum and chondrocostal joints (Fig. 26.2).

26.2.2.2 Etiology and Pathogenesis

The etiology of PC is not known, but the pathogenetic mechanism could be (as for PE)

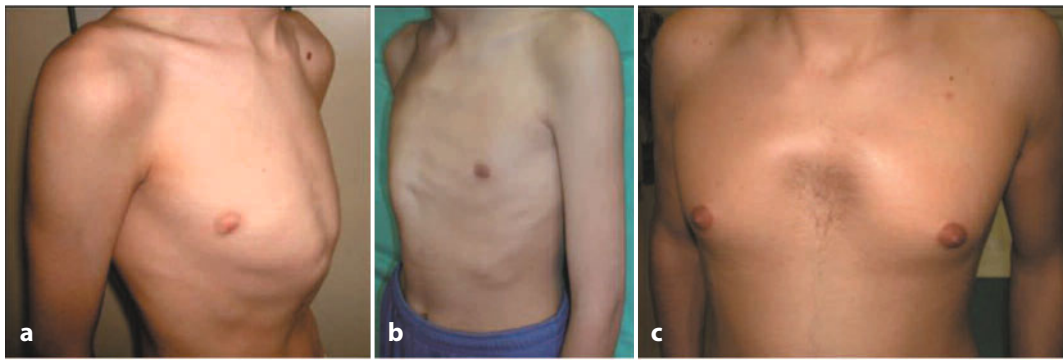

Fig. 26.2 Types of PC. Type 1 (inferior) PC (**a**). Unilateral PC (**b**). Currarino–Silverman syndrome (**c**)

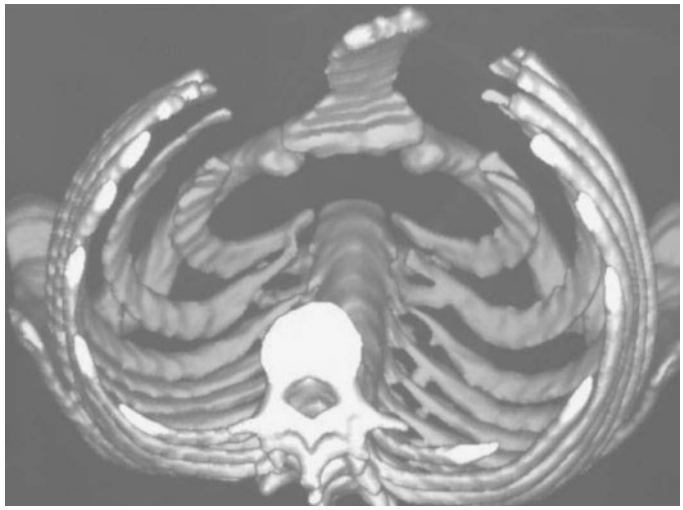

Fig. 26.3 CT reconstruction showing asymmetric unilateral PC

an overgrowth of the ribs. PC patients report anomalies of costal cartilage similar to those seen in PE patients [2]. Familial cases are not uncommon [2, 23] and it is possible to observe PC and PE cases in the same family [23]. Seldom associated with PC are connective-tissue disorders, Noonan syndrome and cardiac anomalies [6].

26.2.2.3 Clinical Aspects

PC usually has a later onset when compared with PE, occurring mainly during pre-puberty or puberty. However, in some cases, it is possible to observe infants or children with this anomaly. As with PE, PC protrusion rapidly increases during the growth spurt. The same symptoms as PE can be observed, with thoracic pain being more frequent than respiratory or cardiologic complaints [4]. In fact, cardiac and pulmonary functions are less involved than in PE [2], whereas psychological effects can be severe: they are the main indication for surgical correction.

26.2.2.4 Diagnosis and Classification

PC can be classified according to localization and symmetry into two types [4]:

- type 1: chondrogladiolar (Fig. 26.3) is the most frequent type and is more often symmetric. Sternal protrusion is located in the inferior or mid-sternum. The lower ribs can be depressed on lateral aspects;
- type 2: chondromanubrial is much less common. The sternum has a normal length and is not depressed in the lower third. This anomaly is probably due (similarly to inferior PC) to a cartilage anomaly.

A particular form of CWM is the Currarino-Silverman syndrome [24] (also called Pouter pigeon breast). Originally placed within the PC family [8], it instead must be distinguished from type-2 PC (though these variants share some features). This is a sternal malformation characterized by premature fusion and ossification of the manubriosternal joint and sternal segments. This results in a superior symmetric carinatum chest deformity with a short thick sternum excavated in the lower third (Fig. 26.2). The sternum on the lateral view is S-shaped. In our opinion, it should be part of sternal anomalies because of the sternal origin of the anomaly (see Table 26.1). Other types of PC described are:

- Lateral or unilateral PC [2]: asymmetric, consists of a protrusion of some costal cartilages on one side (Fig. 26. 3). The sternum can be rotated towards the opposite side.
- Reactive PC [25] is a complication of PE correction. It is more frequent in patients with connective-tissue disorders.

To best assess the severity and asymmetry of PC, some radiological indices have been proposed [26] that are measurable on CT but which in clinical practice are used less often than the Haller index is for PE. CT remains the "gold standard" imaging evaluation for PC.

26.2.2.5 Management, Outcome and Follow-up

As for PE, the standard correction is through costal excision surgery. Ravitch in 1952 was one of the first to describe the surgical technique for PC, whereas Howard introduced the principle of sternal osteotomy, which is usually required to correct the defect. There are many ways of carrying out an osteotomy; on the anterior sternal plate it can be done in a transverse approach, whereas in the case of asymmetric PC it can be undertaken in an oblique fashion. Recently, some modifications to the Ravitch procedure were proposed [27, 28]. They attempted to reduce its invasiveness as well as the extent of resection of muscle and cartilage. In case of the Currarino–Silverman syndrome, the best treatment is the open procedure, whereas alternatives (such as minimally invasive or conservative techniques) have been proposed recently for type-1 PC, with good results, as discussed below.

The orthotic brace system, proposed in 1992, gained popularity only recently [29]. This system is based on the principle of reshaping the thorax during puberty, thanks to its malleability (as in the Nuss procedure for PE), by applying dynamic compression upon it. Martinez-Ferro et al. [23] added to this system the possibility of measuring the pressure necessary for the correction and regulating it (dynamic compression system (DCS)). Good results were seen in a large proportion of patients, especially if the brace was used for most of the time during the day and night. A significant proportion of patients (13.8%) abandoned treatment, and some minor complications (hematomas, ulcerations, back pain) were reported in 12.5% of patients [23]. Moreover, this approach cannot be used in adults due to the ossification of the thorax, so it can be applied only to adolescent patients.

The intrathoracic compression (Abramson's) procedure [30] is based on same principle as the one for bracing. A metal bar is placed surgically in the presternal space through two lateral incisions under the pectoralis muscles and fixed to lateral stabilizers. It is like a "reverse Nuss procedure" and has an age limit. The advantage is obtaining the result immediately without wearing an external brace. In the series reported by Abramson et al., the results at 5 years were good; the bar is usually removed after ≥2 years [31].

Thoracoscopic cartilage resection [32] was described recently. It is undertaken under thoracoscopy and consists of cutting the anomalous costal cartilages unilaterally or bilaterally (according to the type of defect) without damaging the internal thoracic vessels. In severe cases, it can be associated with an intrathoracic compression procedure according to the Abramson procedure to better stabilize the sternum.

Thoracoscopic complete cartilage resection with perichondrium preservation (CCRPP) [33] has been reported by our research team. It is different from the procedures described above because cartilages are isolated laterally and medially to the internal thoracic vessels, up to the chondrosternal joints. Internal thoracic vessels are coagulated and cartilages excised completely, leaving the anterior perichondrium intact.

Minimally invasive submuscular dissection involves dissection of the pectoralis muscle and is carried out by insufflating CO_2 in the submuscular space. Resection of the ribs, sternal osteotomy, and insertion of trans-sternal steel struts are undertaken through a sagittal pre-sternal incision under endoscopic view. Recently, some technical variations have been reported [34]. The pre-sternal incision was abandoned and a major submuscular dissection was carried out with two lateral incisions between the anterior and middle axillary lines. These modifications should allow for the creation of a submuscular and presternal tunnel to implant a Nuss metal bar presternally.

However, special eight-hole stabilizers are required.

Minimal access treatment involves a bar being inserted (as in the Abramson procedure) through two lateral incisions above the sternum. However, the bar passes on both sides into the thoracic cavities; thoracoscopy can be used.

Reactive PC after the Nuss procedure can be corrected by withdrawing the bar. An open procedure is advised only in the case of failure or in other cases. Alternatively, a minimally invasive procedure can be attempted.

26.3 Type II: Costal Anomalies

26.3.1 Dysmorphic and Non-syndromic

Cartilaginous ribs are malformed and the consequence can be a unilateral or bilateral depression in the thoracic wall. The treatment of these malformations comprises excision of the anomalous ribs. A rare malformation belonging to this group is the "intrathoracic rib" (Fig. 26.4) and is classified into four types [35]:

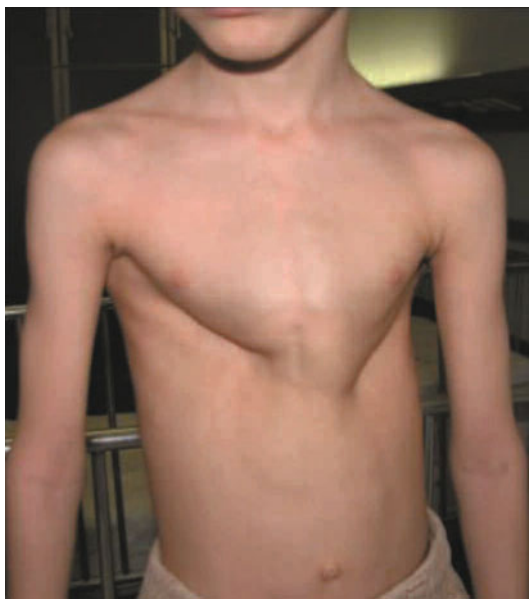

Fig. 26.4 Bilateral type-III intrathoracic rib

- type Ia is a supernumerary rib articulated with a vertebral body;
- type Ib is a bifurcated ("bifid") rib taking its origin close to the vertebral body;
- type II a bifid rib arising more laterally;
- type III is a non-bifid rib depressed into the thoracic cavity.

Another malformation is flaring chest, which comprises hypertrophy or fusion of the cartilaginous ribs in the lower costal margin. Open resection of all malformed ribs is an option. Other frequently observed malformations are cartilage rib asymmetries. They appear as an isolated protrusion of the cartilage ribs and, in most cases, the ribs are fused.

26.3.2 Syndromic Anomalies

26.3.2.1 Jeune Syndrome

Jeune syndrome (also known as asphyxiating thoracic dystrophy) is an autosomal recessive disorder. It was originally described by Jeune in 1954 in a pair of siblings. The frequency of the condition is rare, with an overall incidence estimated between 1/100,000 and 1/30,000 live births. Jeune syndrome is characterized by severe bone abnormalities. Usually, the affected patients have a long, narrow thorax with a reduced thoracic capacity, leaving the lungs with insufficient room to expand and grow. Both thoracic diameters (anteroposterior and lateral) are reduced, causing severe respiratory distress. Patients who develop respiratory symptoms during the first months of life have a poor prognosis, usually resulting in death during infancy.

All patients have small chests with short, wide and horizontal ribs (Fig. 26.5). Clinical and imaging aspects of this malformation vary between patients, and two variants exist:

- the severe variant represents ≈70% of cases. It is usually lethal during infancy. The thorax is extremely small, leaving the abdomen with a prominent aspect; respiratory failure is the rule.
- in the mild variant, the ribs are less affected (≈30% of patients), respiratory symp-

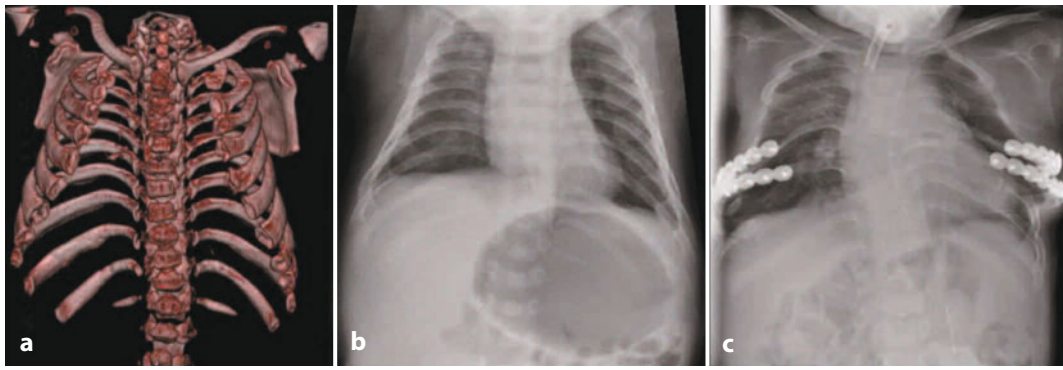

Fig. 26.5 Jeune syndrome. CT reconstruction showing short, wide and horizontal ribs with reduction of intrathoracic capacity (**a**). Chest radiograph demonstrating the disproportion between the thoracic and abdominal diameters and the rib anomalies mentioned above (**b**). The result of bilateral thoracic expansion (**c**)

toms are manageable and survival is prolonged. Renal or liver dysfunctions can be present, and can lead to death.

Surgical repair methods typically involved median sternotomy (with graft interposition), resulting in poor outcomes [36]. Other methods have been proposed: lateral thoracic expansion realized by rib incisions that are sutured in a staggered fashion or, more recently, a vertical expandable prosthetic titanium rib (VEPTR). Both of these methods seem to offer good results. Sometimes, the mild type of Jeune syndrome may not require treatment.

26.3.2.2 Cerebrocostomandibular Syndrome

Cerebrocostomandibular syndrome is such a rare entity that there is no clinical experience of it in the world. We diagnosed 1 case in the last decade: the main feature was a lack in development of the rib cage. Affected patients have only costal vestiges, the chest has a flail aspect, and mechanical ventilation is required from birth. In some cases, the thoracic cage agenesis can be unilateral. This defective costal development can also be associated with features of the Pierre–Robin anomaly. Cerebral maldevelopment or malfunction is also common [37].

26.3.2.3 Costal Agenesis

Costal agenesis is usually limited to some ribs and is not syndromic. These are also rare conditions. Lung herniation may occur, requiring thoracoplasty using the same technique used for Poland Syndrome (PS).

26.3.3 Rare type-II CWMs

There is a series of CWMs that are rarely observed and not included in standard classifications. They differ from each one from other so treatment must be personalized.

26.4 Type III: Chondrocostal Anomalies

26.4.1 Poland Syndrome (PS)

PS occurs in around 1/30,000 live births [38]. It is characterized by the absence or hypoplasia of the pectoralis major muscle, which is often associated with other ipsilateral abnormalities of the chest wall, breast and upper limb [5, 39]. The defect is essentially unilateral and, in most cases, right-sided, although very rare bilateral cases have been described [40, 41]. There is a male preponderance (2:1). Its etiology is not known, but the most accredited hypothesis is interruption of the vascular supply in subclavian and vertebral arteries during embryonic life, leading to different malformations in the corresponding districts. According to this, PS could be interpreted being a sequence.

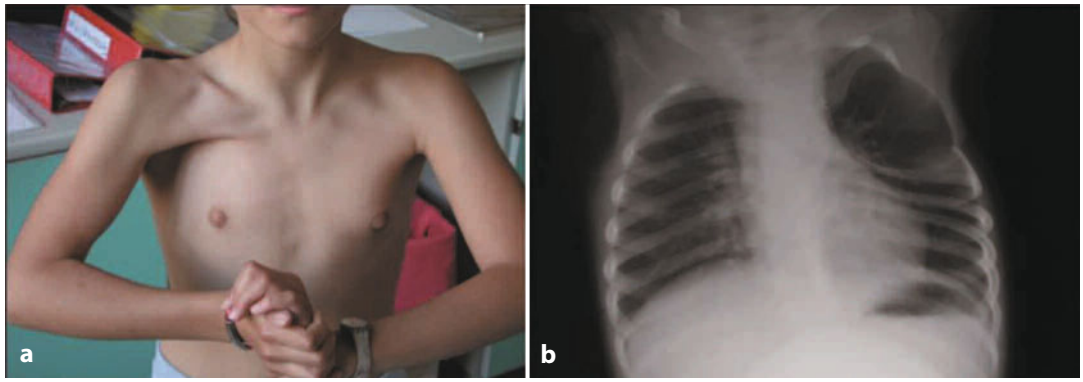

Fig. 26.6 Poland syndrome. Agenesia in the left major and minor pectoralis muscle (**a**). Agenesia in the left upper ribs with lung herniation (**b**)

PS is usually sporadic. However, in 4% of cases, there is familial recurrence, and in 8% of patients some upper-limb or thoracic anomaly (not classifiable as PS) is present in a family member [42]. Possible transmission with an autosomal dominant pattern has been hypothesized. Association with other anomalies (e.g., Moebius, Klippel–Feil syndromes, Sprengel anomaly) [41] has been reported.

The PS phenotype is extremely variable. The thoracic defect is usually evident at birth, but can pass undiagnosed until the child gets older. In fact, the pectoralis muscle deficiency causes an asymmetric aspect, more evident if there are associated costal anomalies (Fig. 26.6). In case of rib agenesis, particularly if multiple (the most affected ribs are the third and the fourth) with a wide defect, lung herniation and paradoxical respiratory movements are evident. Ribs can also be hypoplasic or anomalous (Fig. 26.6). Other CWMs such as PE or PC can be associated, but surgery is required in <10% of cases. A wide spectrum of defects ranging from a mild degree of breast hypoplasia to complete absence of mammary glands is a constant feature. Other uncommon anomalies are cardiac and renal defects, as well as scoliosis. Dextrocardia is reported frequently, always associated with left PS, and seems to be caused by mechanical factors during embryonic life in patients with multiple left rib agenesis [43]. Patients are asymptomatic, and usually do not suffer any limitation due to muscle defects. The upper limb is frequently involved, ranging from the classical symbrachydactyly to split hand or other defects.

The main indication to carry out a thoracoplasty is cosmetic. Usually, it is undertaken at puberty and is rarely necessary during infancy. There is no evidence of the utility of any surgical procedure to guarantee protection against thoracic traumatic injuries in children with rib agenesis. In case of surgical correction in the pediatric age, some options are available, from costal transposition, to a prosthetic repair [44, 45]. According to some authors [8], costal transposition and the consequent stabilization of the thorax can prevent the progression of thoracic deformity, but there is no consensus about this concept. Most authors [45] prefer to wait until puberty and later to correct, in one or more stages, the thoracic flail chest and pectoral defect. In female patients, the most frequent issue is breast and pectoral reconstruction. Correction with prostheses alone or associated with other surgical procedures (transposition of the latissimus dorsi muscle or rectal abdominal muscle; lipofilling; omental flap) has been advocated [45]. Each surgical approach must be tailored to the patient. In males, the same methods can be applied, but the indication for the surgical procedure needs to be evaluated case by case because the esthetic defect is less important. Recently, Martinez-Ferro described transposition of latissimus dorsi muscle flaps using a minimally invasive approach [46]. Usually,

the approach is multidisciplinary (pediatric, thoracic, plastic surgeons), thereby providing the best chance of a successful outcome.

26.5 Type IV: Sternal Anomalies

26.5.1 Sternal Cleft (SC)

SC is a rare idiopathic defect in fusion of the sternum. Acastello et al. found that SC accounts for 0.15% of all CWMs [47]. Also, the HOXB gene might be involved in the development of SC. In our opinion, the clearest classification has been proposed by Shamberger and Welch [48] and involves four types:

- Thoracic ectopia cordis: the heart is ectopic, uncovered by skin, and usually has intrinsic anomalies (majorly when anterior) and kephalic ectopia. The sternal defect can be superior, inferior, central (rare) or total. Abdominal-wall defects can be associated. The thorax is hypoplasic, so surgical correction is usually not sufficient to save life (though isolated survival after surgery has been reported).
- Cervical ectopia cordis is the rarest type. The heart is cranial, sometimes with the apex fused with the mouth. Maxillofacial anomalies are common. The prognosis is always poor.
- Thoracoabdominal ectopia cordis: the heart is covered by a thin membranous or cutaneous layer. The sternal defect is inferior. The heart (located into the thorax or in the abdomen) is not rotated as in the types described above, but intrinsic anomalies are common. This type of anomaly is, in general, found as part of the Pentalogy of Cantrell [49]. If surgically repaired, the prognosis can be good.
- SC is the most common of this group of CWMs, comprising a congenital malformation of the anterior thoracic wall, arising in a deficiency in the midline embryonic fusion of the sternal halves. The incidence is unknown, but it is more common in females [8].

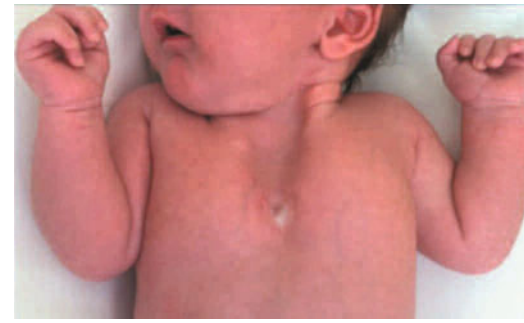

Fig. 26.7 Total sternal cleft in a newborn (note the congenital connectival nevus)

SC can also be classified as "partial" or "complete" (Fig. 26.7). The partial deformity can be located superiorly or inferiorly. The inferior variety is less common and is often associated with a thoracoabdominal ectopia cordis, whereas the more frequent upper partial cleft can be an isolated abnormality. Sternal clavicular joints are displaced laterally, but the clavicles have a normal length. Thoracic viscera bulge in the midline across the defect, which is more evident during forced expiration. A complete form (also described) is much less common. There is a total lack of fusion; it produces an even bigger paradoxical movement than partial cleft and can cause respiratory distress.

Moreover, SC is frequently associated with other defects (82%) [50]. These must be carefully looked for before any surgical procedure because they can lead to major complications. Some of them are evident on physical examination (e.g., maxillofacial hemangiomas) [51], cleft lip or cleft palate, PE, connectival nevi [50], supraumbilical raphe, or gastroschisis. Other defects must be ruled out, such as cardiac defects, aortic coarctation, eye abnormalities, posterior fossa anomalies, and hidden hemangiomas (cerebral or subglottic in particular).

In some cases, SC is also part of clinical conditions. These include: posterior fossa malformations–hemangiomas–arterial anomalies–cardiac defects–eye abnormalities–sternal cleft and supraumbilical raphe syndrome

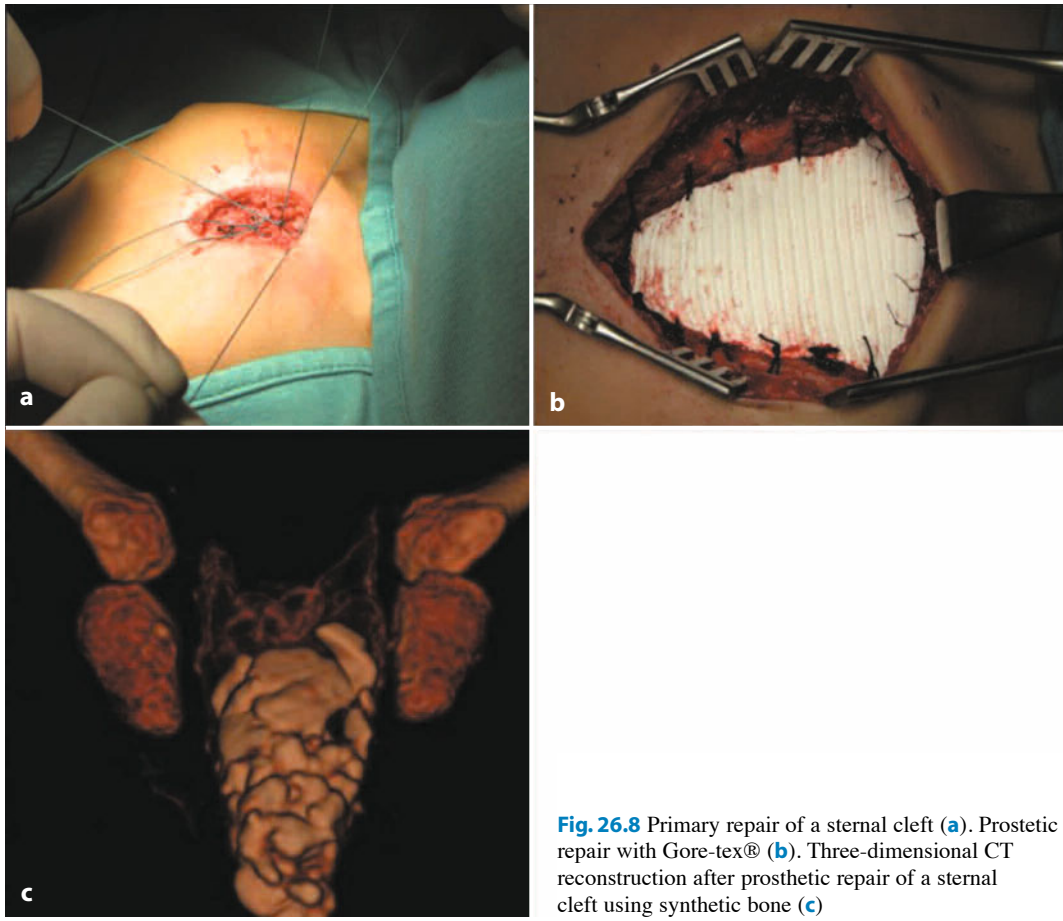

Fig. 26.8 Primary repair of a sternal cleft (**a**). Prostetic repair with Gore-tex® (**b**). Three-dimensional CT reconstruction after prosthetic repair of a sternal cleft using synthetic bone (**c**)

(PHACES) [52]; sternal malformation/vascular dysplasia; midline fusion defects; pentalogy of Cantrell.

The treatment of SC is surgical. Surgery is crucial: to re-establish the bony protection of the mediastinum; to prevent paradoxical visceral movement; to allow normal growth of the thoracic cage; for cosmetic reasons. Ideally, it should take place during the neonatal period or in the first months of life [8, 53, 54]. The reason for preferring an early surgical approach is that primary closure is easier and there is no need for a big procedure (which may be necessary at older ages). In fact, later in life, primary closure (Fig. 26.8) could require: sternoclavicular disarticulation; sternal isolation; inferior sternal osteotomy and medialization of the neck muscles after separation of their sternoclavicular attachments laterally [47]. Moreover, because it can carry the risk of circulatory impairment due to cardiovascular compression, in some cases primary sternal suture may not be possible and prosthetic or autologous closure [54] may be preferable because it is less invasive (Fig. 26.8). Sometimes, partial thymectomy can be useful to reduce the pressure on thoracic vessels [53]. Several prosthetic materials have been described for SC repair. We closed an upper cleft in one 8-year-old female with artificial bone tissue and had an excellent outcome. Complications are not common, but PE can occur later in life in patients who undergo surgery for SC. In case of prosthetic repair, there is an increased risk of infections and recurrence.

26.6 Other Anomalies

26.6.1 Postoperative Surgical Deformities

This category includes cases in which thoracic deformities develop after correction of other CWMs. We have experience of a few cases of these types of anomalies (Fig. 26.9). They can be due to multiple resections of cartilage during open surgery for a young patient with PE or due to previous thoracotomy (Fig. 26.9) or sternotomy (Fig. 26.10) for other reasons and finally result, after many years, in a thoracic deformity that requires surgical revision. As discussed above, the optimal age for PE repair is controversial [3, 10]. Repair in early childhood is easier but carries a high risk of causing restrictive growth patterns in the chest wall. Acquired Jeune syndrome is associated with open repair in young children, usually <4 years of age, with extensive resection of ≥5 ribs [12] and damage to the cartilage growth centers [55]. For these reasons, most authors postpone open surgical repair until after the patient is >10 years of age [3,10].

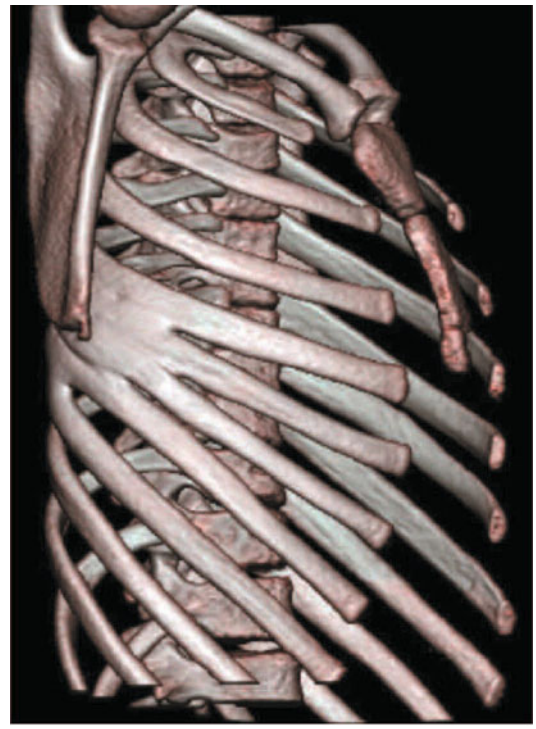

Fig. 26.9 Post-thoracotomy deformity. The ribs from the fourth to eighth rib are fused, causing scoliosis and reduced lung-function volumes. This female underwent three previous right thoracotomies for complicated esophageal atresia

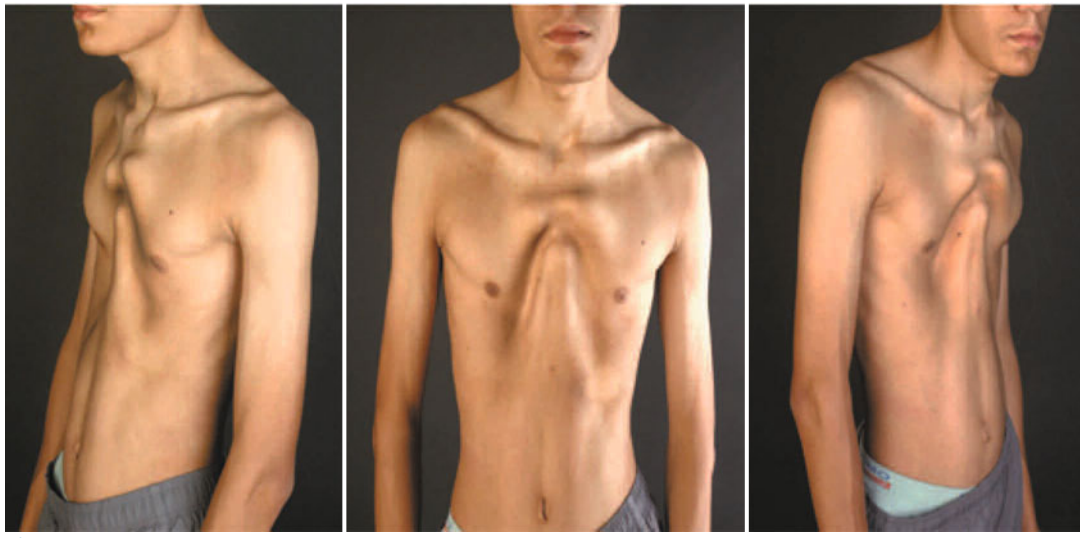

Fig. 26.10 Post-sternotomy deformity in a boy who underwent surgical treatment for tetralogy of Fallot

26.7 Conclusions

CWMs are a wide spectrum of anomalies. The etiological and genetic implications of CWMs are poorly characterized. Precise identification of a single malformation, its embryological classification, and an accurate diagnostic assessment are the first fundamental steps. Identification of familial cases, possible associated syndromes and anomalies, clinical symptoms and psychological implications, must be considered. Among the therapeutic armamentarium, classical methods and new approaches allow the most appropriate method for the individual patient to be chosen. This choice is according to the experience and preference of the surgeon but is achieved by tailoring the treatment to individual clinical and psychological needs. A multidisciplinary approach is advisable to manage CWMs in all their complexity.

References

1. Nuss D, Kelly RE, Croitoru DP, Katz ME (1998) A 10 year review of a minimally invasive technique for the correction of pectus excavatum. J Pediatr Surg 33:545–552
2. Fokin AA, Steuerwald NM, Ahrens WA et al (2009) Anatomical, histologic and genetic characteristics of congenital chest wall deformities. Semin Thorac Cardiovasc Surg 21:44–57
3. Lopushinsky SR, Fecteau AH (2008) Pectus deformities: A review of open surgery in the modern era. Semin Pediatr Surg 17:201–208
4. Colombani P (2009) Preoperative assessment of chest wall deformities. Semin Thorac Cardiovasc Surg 21:58–63
5. Kelly RE (2008) Pectus excavatum: historical background, clinical picture, preoperative evaluation and criteria for operation. Semin Pediatr Surg 17:181–193
6. Kotzot D, Schwabegger AH (2009) Etiology of chest wall deformities – genetic review for the treating physician. J Pediatr Surg 44:2004–2011
7. Creswick HA, Stacey MW, Kelly RE et al (2006) Family study on the inheritance of pectus excavatum. J Pediatr Surg 41:1699–1703
8. Acastello E (2006) Patologias de la pared toracica en pediatria. Editorial El Ateneo, Buenos Aires
9. Haller JA, Kramer SS, Lietman SA (1987) Use of CT scans in selection of patients for pectus excavatum surgery: a preliminary report. J Pediatr Surg 22:904–906
10. Nuss D (2008) Minimally invasive surgical repair of pectus excavatum. Semin Pediatr Surg 17:209–217
11. Aronson DC, Bosgraaf RP, van der Horst C et al (2007) Nuss procedure: pediatric surgical solution for adults with pectus excavatum. World J Surg 31:26–29
12. Haller JA, Colombani PM, Humphries CT et al (1996) Chest wall constriction after too extensive and too early operations for pectus excavatum. Ann Thorac Surg 61:1618–1624
13. Ravitch MM (1966) Atyipical deformities of the chest wall – absence and deformities of the ribs and costal cartilages. Surgery 59:438–449
14. Grappolini S, Fanzio PM, D'Addetta PGC et al (2008) Aesthetic treatment of pectus excavatum: a new endoscopic technique using a porous polyethylene implant. Aesth Plast Surg 32:105–110
15. Schier F, Bahr M, Klobe E (2005) The vacuum chest wall lifter; an innovative, nonsurgical addition to the management of pectus excavatum. J Pediatr Surg 40:496–500
16. Harrison MR, Estefan-Ventura D, Fechter R (2007) Magnetic mini-mover procedure for pectus excavatum: development, design and simulations for feasibility and safety. J Pediatr Surg 41:81–86
17. Kelly RE, Shamberger RC, Mellins R et al (2007) Prospective multicenter study of surgical correction of pectus excavatum: design, perioperative complications, pain, and baseline pulmonary function facilitated by internet-based data collection. J Am Coll Surg 205:205–216
18. Kelly RE, Goretsky MJ, Obermeyer R et al (2010) Twenty-one years of experience with minimally invasive repair of pectus excavatum by the Nuss procedure in 1215 patients. Ann Surg 252:1072–1081
19. Park HJ, Lee SY, Lee CS (2004) Complications associated with the Nuss procedure: analysis of risk factors and suggested measures of prevention of complications. J Pediatr Surg 39:391–395
20. Moss L, Albanese C, Reynolds M. (2001) Major complications after minimally invasive repair of pectus excavatum: case reports. J Pediatr Surg 36:155–158
21. Van Renterghem KM, von Bismarck S, Bax NM et al (2005) Should an infected Nuss bar be removed? J Pediatr Surg 40:670–673
22. Rapuzzi G, Torre M, Romanini Mvet al (2010) The Nuss procedure after breast augmentation for female pectus excavatum. Aesthetic Plast Surg 34:397–400
23. Martinez-Ferro M, Fraire C, Bernard S (2008) Dynamic compression system for the correction of pectus carinatum. Semin Pediatr Surg 17:194–200
24. Currarino G, Silverman FN (1958) Premature obliteration of the sternal sutures and pigeon-breast deformity. Radiology 70:532–540
25. Swanson JW, Colombani PM (2008) Reactive pectus carinatum in patients treated for pectus excavatum. J Pediatr Surg 43:1468–1473
26. Egan JC, Du Bois JJ, Morphy M et al (2000) Compressive orthotics in the treatment of asymmetric pectus carinatum: a preliminary report with an objective radiographic marker. J Pediatr Surg 35:1183–1186

27. Del Frari B, Schwabegger AH (2011) Ten-year experience with the muscle split technique, bioabsorbable plates, and postoperative bracing for correction of pectus carinatum: The Innsbruck protocol. J Thorac Cardiovasc Surg 141:1403–1409
28. Fonkalsrud EW, Anselmo DM (2004) Less extensive techniques for repair of pectus carinatum: the undertreated chest deformity. J Am Coll Surg 198:898–905
29. Kravarusic D, Dicken BJ, Dewar R et al (2006) The Calgary protocol for bracing of pectus carinatum: a preliminary report. J Pediatr Surg 41:923–926
30. Abramson H (2005) A minimally invasive technique to repair pectus carinatum. Preliminary report. Arch Bronconeumol 41:349–351
31. Abramson H, D'Agostino J, Wuscovi S (2009) A 5-year experience with a minimally invasive technique for pectus carinatum repair. J Pediatr Surg 44:118–124
32. Kim S, Idowu O (2009) Minimally invasive thoracoscopic repair of unilateral pectus carinatum. J Pediatr Surg 44:471–474
33. Varela P, Torre M (2011) Thoracoscopic cartilage resection with partial perichondrium preservation in unilateral pectus carinatum: preliminary results. J Pediatr Surg 46:263–266
34. Schaarschmidt K, Lempe-Sellin M, Schlesinger F (2011) New Berlin-Buch "reversed Nuss£, endoscopic pectus carinatum repair using eight-hole stabilizers, submuscular CO2, and presternal Nuss bar compression: first results in 35 patients. J Laparoendosc Adv Surg Tech A 21:283–286
35. Kamano H, Ishihama T, Ishihama H et al (2006) Bifid intrathoracic rib: a case report and classification of intrathoracic ribs. Intern Med 45:627–630
36. Philips JD, van Aalst JA (2008) Jeune's syndrome (asphyxiating thoracic dystrophy): congenital and acquired. Semin Pediatr Surg 17:167–172
37. Drossou-Agakidou V, Andreou A, Soubassi-Griva V et al (1991) Cerebrocostomandibular syndrome in four sibs, two pairs of twins. J Med Genet 28:704–707
38. Freire-Maia N, Chautard EA, Opitz JM et al (1973) The Poland syndrome – clinical, and genealogical data, dermatoglyphic analysis, and incidence. Hum Hered 23:97–104
39. Poland A. (1841) Deficiency of the pectoralis muscles. Guys Hosp Rep 6:191
40. Baban A, Torre M, Bianca S et al (2009) Poland syndrome with bilateral features: case description with review of the literature. Am J Med Genet 149A:1597–1602
41. Karnak I, Tanyel FC, Tunçbilek E et al (1998) Bilateral Poland anomaly. Am J Med Genet 75:505–507
42. Baban A, Torre M, Costanzo S et al (2011) Familial Poland anomaly revisited. Am J Med Genet A Nov 22. doi: 10.1002/ajmg.a.34370 (Epub ahead of print)
43. Torre M, Baban A, Buluggiu A et al (2010) Dextrocardia in patients with Poland syndrome: Phenotypic characterization provides insight into the pathogenesis. J Thorac Cardiovasc Surg 139:1177–1182
44. Shamberger RC, Welch KJ, Upton J III (1989) Surgical treatment of thoracic deformity in Poland's syndrome. J Pediatr Surg 24:760–765
45. Moir CR, Johnson CH (2008) Poland's syndrome. Semin Pediatr Surg 17:161–166
46. Martinez-Ferro M, Fraire C, Saldaña L et al (2007) Complete videoendoscopic harvest and transposition of latissimus dorsi muscle for the treatment of Poland syndrome: a first report. J Laparoendosc Adv Surg Tech 17:108–113
47. Acastello E, Majluf F, Garrido P et al (2003) Sternal cleft: a surgical opportunity. J Pediatr Surg 38:178–183
48. Shamberger R, Welch K (1990) Sternal defects. Pediatr Surg Int 5:156–164
49. Cantrell JR, Haller JA, Ravitch MM (1958) A syndrome of congenital defects involving the abdominal wall, sternum, diaphragm, pericardium, and heart. Surg Gynecol Obstet 107:602–614
50. Torre M, Rapuzzi G, Carlucci M et al (2011) Phenotypic spectrum and management of sternal cleft: literature review and presentation of a new series. Eur J Cardiothorac Surg 2012 41:4–9
51. Fokin AA (2000) Cleft sternum and sternal foramen. Chest Surg Clin N Am 10:261–276
52. Metry D, Heyer G, Hess C et al (2009) Consensus statement on diagnostic criteria for PHACE syndrome. Pediatrics 124:1447–1456
53. Torre M, Rapuzzi G, Guida E et al (2008) Thymectomy to achieve primary closure of total sternal cleft. J Pediatr Surg 43:e17–e20
54. De Campos JR, Filomeno LT, Fernandez A et al (1998) Repair of congenital sternal cleft in infants and adolescents. Ann Thorac Surg 66:1151–1154
55. Robicsek F, Watts LT, Fokin AA (2009) Surgical repair of pectus excavatum and carinatum. Semin Thorac Cardiovasc Surg 21:64–75