Classification of Chest Wall Deformities

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Amulya K. Saxena

Introduction

Chest wall deformities (CWDs) encompass a wide range of anomalies which extend from the sternum to the vertebral column. Few patients with CWDs present clinically with functional respiratory impairment and noticeable symptoms, but these patients with deformities seek medical due to psychosocial concerns and their aversion to sports and public exposure. The variations and the complexities in the presentation and occurrence of CWDs, often leads to their misdiagnosis or neglect by medical care specialists, that results in inappropriate diagnosis which in turn delays the therapeutic management. Surgical repair using radical procedures and inconsistent reporting of outcomes has deterred the medical community, not directly involved in repairs, to opt for a conservative "wait and see" approach. However, with the introduction of the minimal access repair techniques as well as evolving options in conservative treatment for pliable CWDs, interest has been revived both in the scientific community and the general public with regards to treatment of the most common forms of CWDs- Pectus excavatum and Pectus carinatum and their combinations.

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The exact description of the various types of CWDs is challenging as some forms are well defined, whereas others are part of a larger spectrum of deformities or syndromes. Classification is of CWDs is important as correct diagnosis has direct implication on the treatment options. Complex classifications are difficult to understand; furthermore, since the management of CWDs involves multiple specialties a clear and concise classification for CWDs is of paramount importance to offer suitable therapy options.

Classification and Guidelines

The classification of CWDs is based on the morphological site of deformities extrending from the sternum to the vertebrae. This "Chest Wall Deformities Morphological Classification" divides CWDs in to 5 types based on the anatomic topography of the chest wall: Type I- sternum, Type 2- costal cartilage, Type 3- rib, Type 4- combined costal cartilage and rib, and Type 5- Costovertebral junction (Table 2.1).

Pectus excavtum, Pectus carinatum and their combinations represent the majority of the patients who are referred for evaluation to medical practitioners worldwide [1]. The trend with regards to these deformities (see Chap. 1) was initially associated with reporting of the deformities for over five centuries with the advent towards open surgical corrections in the 1930s. This was followed by introduction of the era of minimal

A.K. Saxena (ed.), *Chest Wall Deformities*, DOI 10.1007/978-3-662-53088-7_2

Table 2.1 Chest wall deformities morphological classification: deformities are classified based on the anatomical topography

Classification	Deformity (anatomical topography)	Characteristic presentation
Type-1	Sternum	Cleft sternum (+/– *Ectopia Cordis*) Currarino Silvermann syndrome
Type-2	Costal cartilage	Pectus excavatum Pectus carinatum
Type-3	Rib	Simple Complex (+/– syndrome)
Type-4	Combined costal cartilage and rib	Poland's syndrome VACTER syndrome
Type-5	Costo-vertebral junction	Osteogenesis imperfecta Syndromic

access correction in the late 1990s which was further followed by the development of conservative approaches in management of pliable CWDs from 2000's using vacuum bells for pectus excavtums and compressive braces for pectus carinatum [2, 3]. Based on these advances in management, a "Guideline for management of Pectus deformities" was proposed to offer an algorithm for patients with combinations of pectus deformities (Fig. 2.1) [1]. According to the guidelines: (A) Mild forms of pectus deformities in young children warrant a "wait and watch" approach, (B) Pliable pectus deformities in school age children and adolescent are suitable for conservative treatment (Vacuum bell-Pectus excavtaum or compression brace- Pectus carinatum), (C) Symmetric (or mild asymmetric) pectus deformities could be best managed by minimal access surgical procedures, and (D) Complex pectus deformities especially those with severe forms of asymmetry, combined formed (Pectus excavtum combined with Pectus carinatum), platythorax or cranial pectus carinatum require an open surgical correction. This guideline also offer flexibility to opt for a minimal access or open surgical procedure based on the severity of the deformity as well as the expertise of the surgeon.

Type 1- Sternum Deformities

Sternal clefts have an estimated incidence of 0.15 % of all CWDs and are formed when there is a defect in the fusion of the sternum [4]. It is presumed to be an embryonic developmental disorder; with the Hoxb gene implicated in the development of this deformity [5]. A classification proposed by Shamberger and Welch in 1990 divided sternal clefts into 4 types [6]:

- ***Thoracic ectopia cordis***: In this form of sternal cleft the heart is ectopic and devoid of a skin covering. The heart is generally located in an anterior position and has intrinsic anomalies. The sternal defect itself can be located cranial, caudal, central or may be total. Thoracic ectopia cordis can also be associated with omphaloceles. Since the thoracic cavity is hypoplasic, surgical correction is generally unsuccessful.
- ***Cervical ectopia cordis***: In these rare defects the heart is located more cranially and this condition is frequently associated with craniofacial anomalies.
- ***Thoracoabdominal ectopia cordis***: In this condition the heart is covered by a thin membranous or cutaneous layer within a caudal located defect. The heart which may be located either in the thorax or in the abdomen, is not rotated as in the previously types described, but is accompanied by intrinsic anomalies. This anomaly is a part of the Pentalogy of Cantrell.
- **Sternal cleft**: This is the most common among the types of sternal deformities and arises from a deficiency in the midline embryonic fusion of the sternal plates. Sternal clefts can be partial or complete. The partial deformities could have a cranial or caudal location. The caudal forms are often associated with a thoraco-abdominal ectopia cordis, whereas the cranial partial clefts exist as isolated malformations. Although the sternoclavicular joints are displaced laterally in sternal clefts, the clavicles retain a normal length. Sternal clefts can frequently be associated with other defects (maxillofacial hemangiomas, cleft lip or cleft palate, pectus excavatum, connectival nevi, supraumbilical raphe, or gastroschisis)

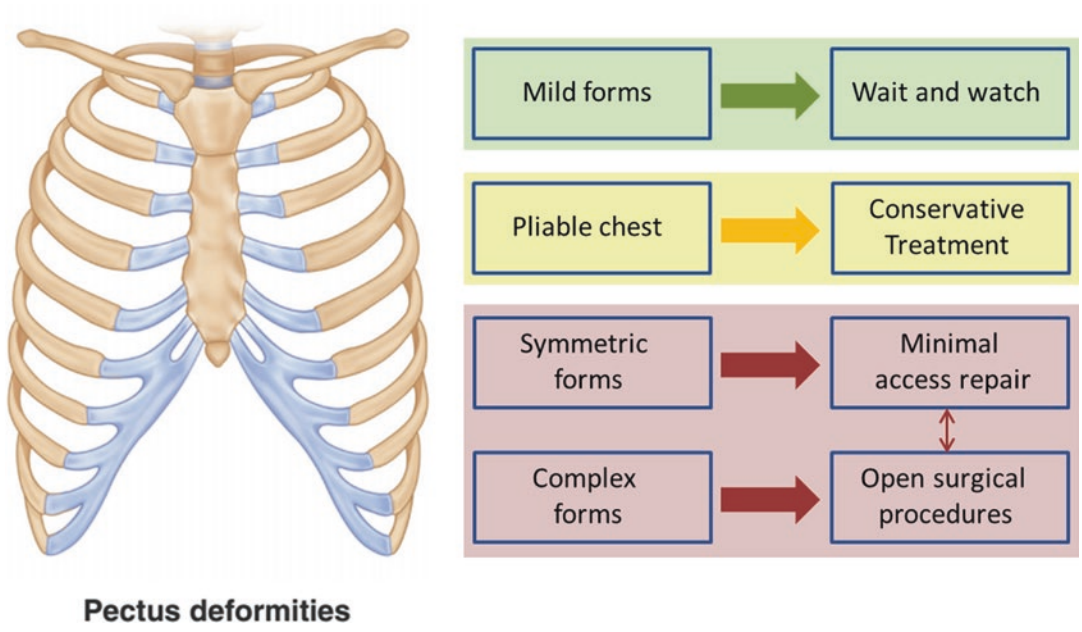


Fig. 2.1 "Guideline for management of Pectus deformities": (**a**) Mild forms in young children warrant a "wait and watch" approach, (**b**) Pliable pectus deformities in school age children and adolescent warrant conservative treatment (Vacuum bell-Pectus excavtaum or compression brace- Pectus carinatum), (**c**) Symmetric (*or mild asymmetric) pectus deformities should be offered minimal access repair options, and (**d**) Complex pectus (severe forms of asymmetry, combined forms, platythorax or cranial pectus carinatum) require an open surgical correction. Minimal access or open surgical procedure options offered are based on the expertise of the surgeon as well as the severity of the deformity

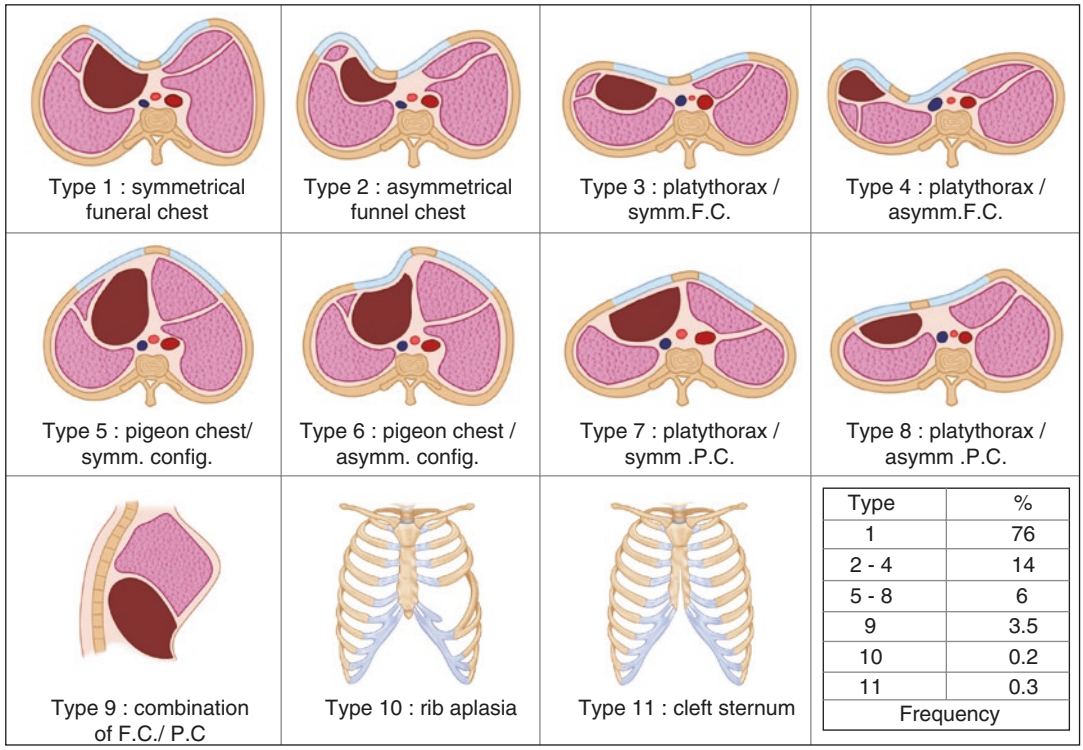
which must be carefully evaluated in the planning of any surgical procedure [7]. Other defects, such as cardiac defects, aortic coarctation, occular abnormalities, posterior fossa anomalies and obscured hemangiomas must be ruled out during evaluation of the patient.

Type 2- Costal Cartilage Deformities

Pectus deformities belong to the type of costal cartilage deformities which present a wide range of forms from pectus excavatum to pectus carinatum and their combinations. The patients are graded according to the Willital's classification, which is based on morphologic findings of the thorax and divides congenital chest wall deformities into 11 types – funnel chest (4 types), pigeon chest (4 types), combination of funnel and pigeon chest, chest wall aplasia and cleft sternum (Fig. 2.2) [8, 9] This classification allows a more accurate description and documentation of the deformities and takes into consideration the asymmetric presentation of pectus deformities along with inclusion of the rare forms.

Willital's classification of congenital chest wall deformities:

Type 1 Symmetric pectus excavatum within a normal configured thorax
Type-2 Asymmetric pectus excavatum within a normal configured thorax
Type-3 Symmetric pectus excavatum associated with platythorax
Type-4 Asymmetric pectus excavatum associated with platythorax
Type-5 Symmetric pectus carinatum within a normal configured thorax
Type-6 Asymmetric pectus carinatum within a normal congigured thorax
Type-7 Symmetric pectus carinatum associated with platythorax
Type-8 Asymmetric pectus carinatum associated with platythorax



Type	%
1	76
2 - 4	14
5 - 8	6
9	3.5
10	0.2
11	0.3
Frequency	

Fig. 2.2 Willital's classification of anterior congenital chest wall deformities [9]

Type-9 Combination of pectus excavatum and pectus carinatum
Type-10 Thoracic wall aplasia/hypoplasia
Type-11 Cleft sternum defects

- ***Pectus excavatum* (*PE*)**
  PE has an incidence of 1:100 to 1:1000 live births and accounts for 90% of the CWDs [10, 11]. It is characterized by the presence of a variably deep sternal depression due to uncontrolled growth of the lowest costal cartilages. PE is commonly congenital, but can appear during childhood or adolescence (Fig. 2.3). The etiology of PE is not clear with regards to the exact nature of the associated connective tissue disorders. Genetics studies on PE have shown the presence of an autosomal dominant transmission pattern, but autosomal recessive and X-linked patterns have also been associated with its development [12]. The overgrowth of costal cartilages has been presumed as the pathogenic mechanism leading to the development of PE [10, 13]. Collagen type II disorders have been demonstrated in the costal cartilages in PE [14], as well as overexpression or down regulation 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 have also been implicated [10].

  PE presents with various degrees of severity which is important in assessment of the condition (Figs. 2.4 and 2.5). Simple patient evaluation can be performed by measuring the depth of the deformity using a caliper (Fig. 2.6). Three areas of reference are measured between the sternum and the vertebral column: A-Manibrium-Vertebral distance, B-Corpus-Vertebral distance and C-Xiphoid-Vertebral distance (Fig. 2.7). These distances provide information with regards to the level of the length of the sternum in relation to the

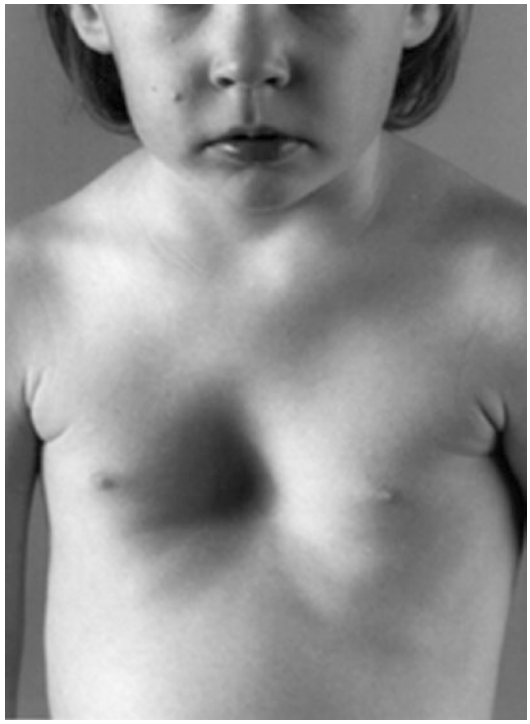


Fig. 2.3 Pectus excavatum in a 2-year old girl presenting with a symmetric deformity

Fig. 2.4 Asymmetric pectus excavtum in a 40-year old male patient with sternal rotation

vertebral column. Using a flexible ruler the contours of the thorax can be estimated (Fig. 2.8). These are then plotted at the point of maximum deformity to determine the variations in depth of the chest wall with regards to geometric proportions (Fig. 2.9).

However, Computerized Tomography (CT) scan are necessary to access the severity of the deformity and to calculate the Haller index to estimate the severity of chest depression in patients with PE (Fig. 2.10) (Haller index is the ratio of the transverse diameter and the anteroposterior diameter of the rib-cage. Haller index in a normal chest is around 2.5, but in pectus excavatum the index can reach 3.25 or even as high as 5.5) [15]. Another important feature in assessment is the symmetry. Asymmetric PE present with variable degrees of sternal rotation which are important to recognize and correct during repairs. Three-dimensional reconstruction images from CT can offer excellent images of the bony thorax with regards to the estimation of rib cage asymmetry and sternal rotation (Fig. 2.11). Another method to map the surface contours of the chest wall is the videostereoraster examination (Fig. 2.12). This examination can be performed preoperative and during follow-ups to compare the results of surgical repair.

- ***Pectus carinatum*** **(*PC*)**
  PC is less frequent than PE and has incidence of approximately 10–15 % [10, 16, 17] with a male predominance (Fig. 2.13). However, in some geographical areas of the world, PC is almost equal or more prevalent than PE [3, 18, 19]. The etiology of PC as in PE is also unknown, but the pathogenesis could be similar to PE which involves in an overgrowth of the costal cartilage [13]. Familial cases are not uncommon and cases of PC and PE within the same family have been reported [3, 10].

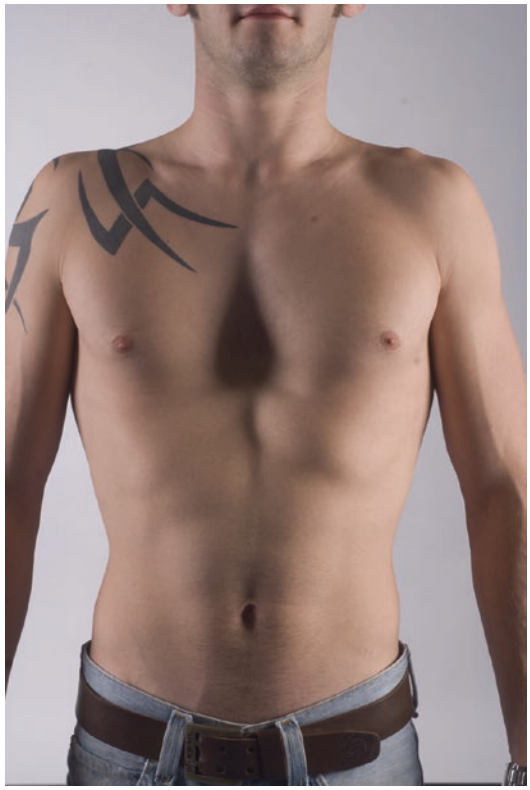


Fig. 2.5 Symmetric pectus excavatum in a 24-year old male patient

Connective tissue disorders and cardiac anomalies are rarely associated with PC [20]. Although PC usually appears later in life than PE, mainly during puberty, this deformity has also been observed in infants and children. With regards to estimation of the contours of the deformity, investigation that were performed in patients for PE- flexible rule and caliper measurement as well as CT can be performed for preoperative analysis (Fig. 2.14).

PC is been classified into 2 types according to the localization into the following types [17, 21]:

– ***Type 1- Chondrogladiolar***: The symmetric sternal protrusion is located in the inferior or mid sternum region. The lower ribs may be slightly or severely depressed on lateral side.
– ***Type 2-Chondromanubrial***: These have been frequently referred to as Currarino-Silverman syndrome or Pouter Pigeon Breast. Currarino-Silverman syndrome is a rare disorder characterized by premature fusion of manubrio-sternal joint and the sternal segments, resulting in a high carinate chest deformity; it is frequently associated with congenital heart disease [22]. However, chondromanubrial forms of PC are frequently isolated findings in patients (without cardiac association).

Reactive Pectus Carinatum This is observed in some patients who have undergone PE repairs. The occurrence of reactive PC which is evident after the first few months or years of minimal access as well as open repair of PE, and presents as a progressive ventral displacement of the sternum [23]. It is more frequent in patients with connective tissue disorders.

Type 3- Rib Deformities

Based on the radiological overview on pediatric rib deformities presented in 2002, the deformities in ribs are based on their origin- congenital or acquired [24]. Since the overview presents all radiological conditions that affect ribs, the following section refers to those rib deformities that are associated with CWD's. Table 2.2 summarizes the list of rib deformities that form the spectrum of CWDs (Table 2.2).

Congenital Deformities

1. ***Cervical ribs***: Cervical ribs arise from the seventh cervical vertebra. They are most commonly an incidental finding. Cervical ribs are rarely symptomatic in early childhood; but in older children and adults, compression of the brachial plexus or subclavian artery can give rise to the formation of a thoracic outlet syndrome [25].
2. **Altered rib count**: An alteration in the number of ribs has been found in trisomy 21 syndrome and in patients with the VATER association (Fig. 2.15) [26, 27]. It is not uncommon to see 11 pairs of ribs in the

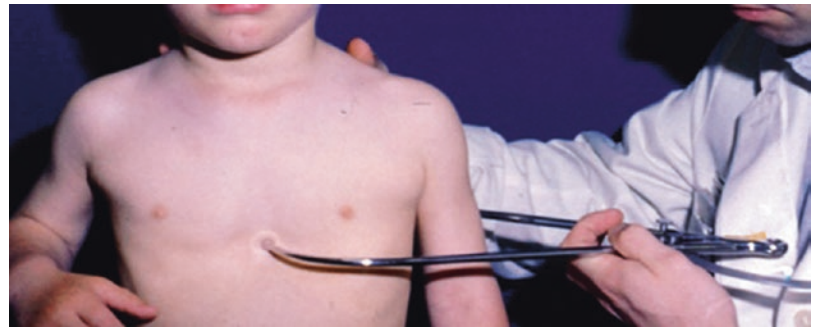


Fig. 2.6 Measurement of sterno-vertebral distances using a caliper

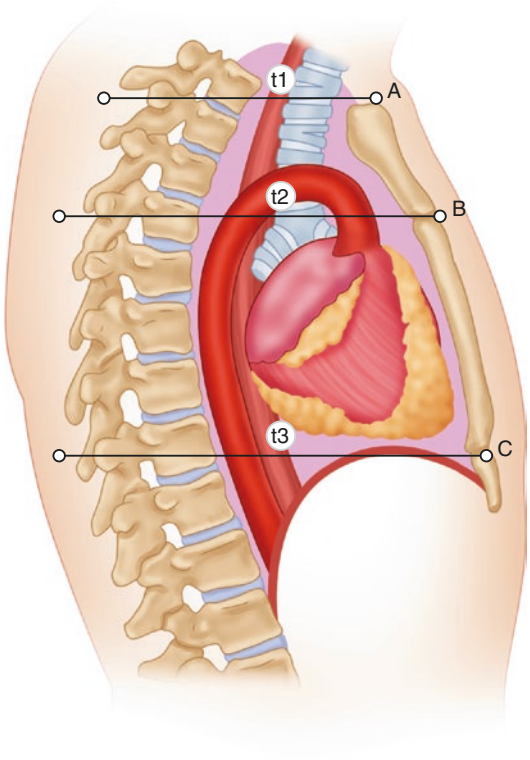


Fig. 2.7 Three areas of reference are measured between the sternum and the vertebral column using the caliper: A-Manibrium-Vertebral distance, B-Corpus-Vertebral distance and C-Xiphoid-Vertebral distance

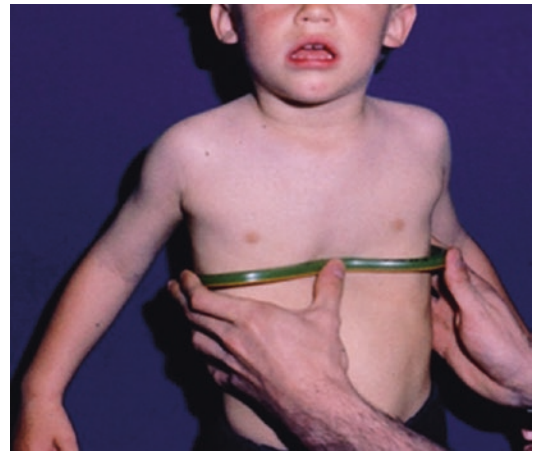


Fig. 2.8 The flexible ruler is placed anteriorly and then posteriorly at the point of maximum deformity to obtain the contours of the thorax

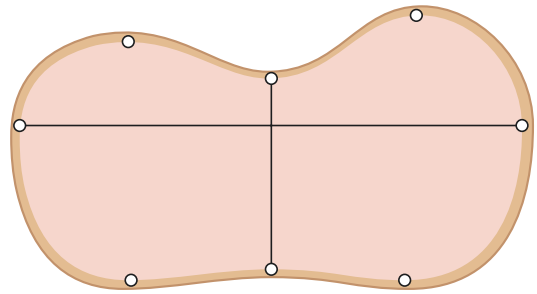


Fig. 2.9 The contours of the thorax obtained flexible ruler are plotted at the point of maximum deformity to determine the variations in depth of the chest wall

absence of associated anomalies; this situation occurs in 5–8 % of normal individuals. Eleven pairs of ribs occur in one-third of patients with trisomy 21 syndrome [27], as well as in association with cleidocranial dysplasia and campomelic dysplasia.

3. **Rib size variations**: Short ribs constitute an integral part of several syndromes. Due to their size, short ribs do not extend as far anteriorly to reach and connect with the sternum. This results in diminished volume of the chest

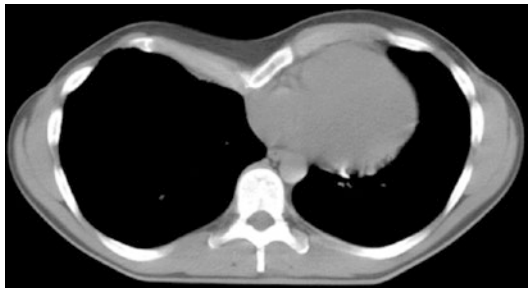


Fig. 2.10 Computed tomography scan of the thorax in a 14-year old patient showing the contours of a symmetric pectus excavtum with a rotation of the sternum

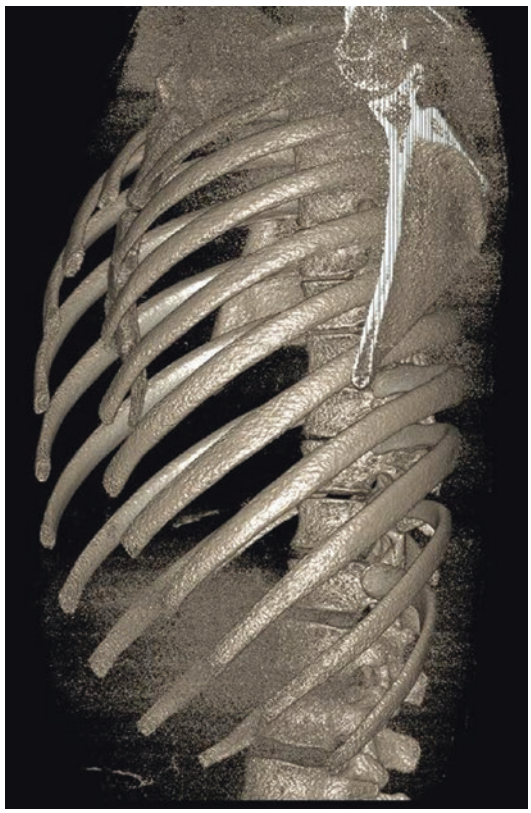


Fig. 2.11 Three-dimensional reconstructed CT images demonstrating sternal depression in relation to the rib cage in a 15-year old pectus excavtum patient

and restricts respiratory motion that further leads to respiratory insufficiency. Definitive diagnosis should be performed early in the neonatal period to exclude a lethal dysplasia so that a resuscitation policy can be planned.

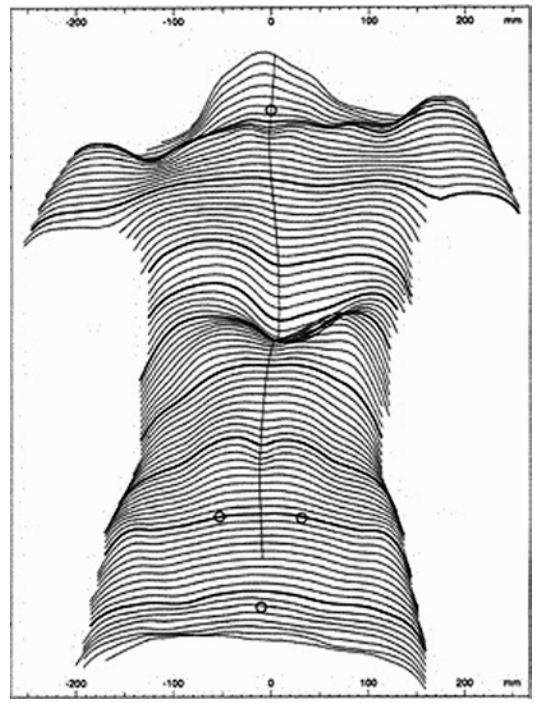


Fig. 2.12 Videoraster examination of a 16-year old male patient with slight asymmetric pectus excavtum deformity

Other conditions with rib size variations are the short rib-polydactyly syndromes (Saldino-Noonan, Majewski, and Verma-Naumoff); chest diameter is critically small in these syndromes.

(a) ***Thanatophoric Dysplasia*** —Thanatophoric dysplasia is a lethal dysplasia transmitted by a dominant gene mutation and is the most common lethal neonatal skeletal dysplasia after osteogenesis imperfecta type II. The ribs in these patients are very short and do not extend beyond the anterior axillary line. Associated findings are cloverleaf skull deformity, polydactyly, and hypoplastic iliac bones. Other conditions with these deformities include thanatophoric variants, homozygous achondroplasia, achondrogenesis and asphyxiating thoracic dysplasia.

(b) ***Jeune Asphyxiating Thoracic Dysplasia*** —Jeune asphyxiating thoracic dysplasia

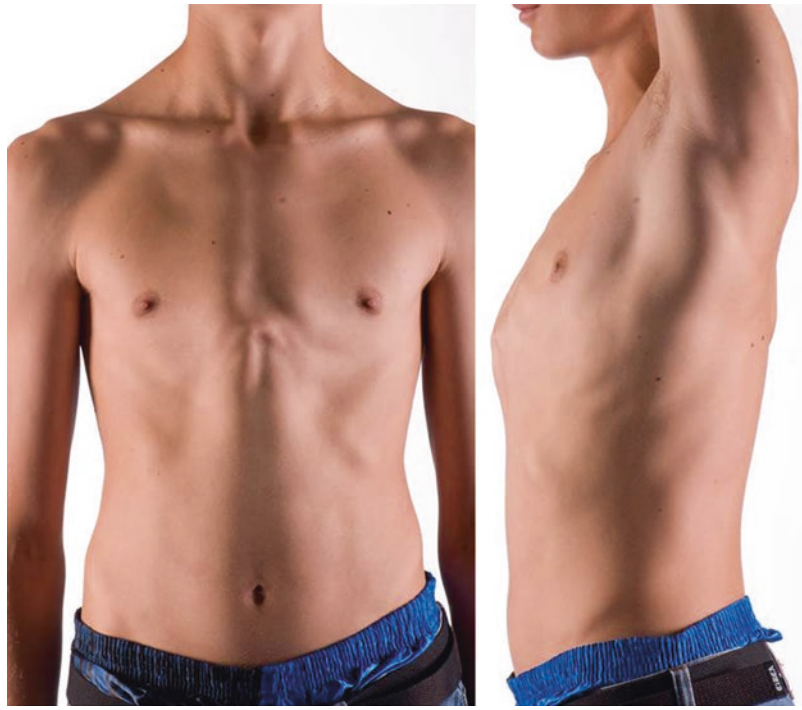


Fig. 2.13 Pectus carinatum deformity in a 16-year old male patient showing the protrusion of the sternum- frontal view (*left*) and lateral view (*right*)

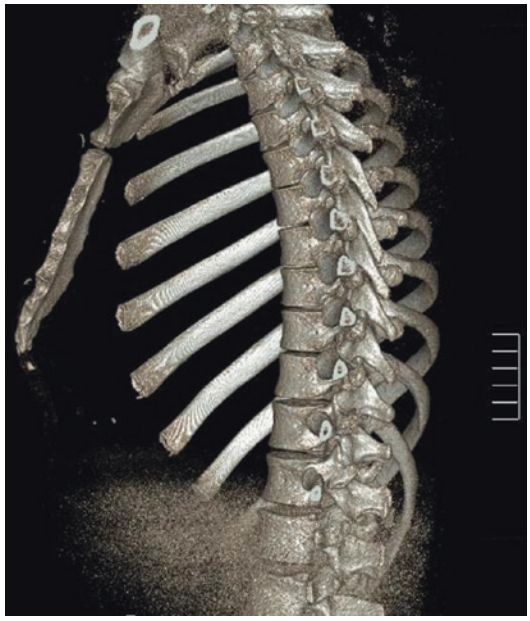


Fig. 2.14 Three-dimensional reconstructed CT images demonstrating sternal protrusion in relation to the rib cage in a pectus carinatum patient

is an autosomal recessive dysplasia [28]. The ribs are short, with a horizontal course, and the chest diameter is significantly decreased compared with that of the abdomen. Although the findings of the ribs on the chest radiograph may be suggestive of Jeune dysplasia, their appearance also resembles that of Ellis-van Creveld dysplasia. Children who do not have a severe affection and reach adulthood, are reported to have an increased incidence of medullary cystic renal disease [29].

(c) ***Ellis-van Creveld Chondroectodermal Dysplasia*** —Ellis-van Creveld chondroectodermal dysplasia is an autosomal recessive dysplasia in which the ribs are short, and the chest cavity is narrow. The narrow thorax amplifies the size of the heart; cardiomegaly is often present because of associated anomalies. Acromelic limb shortening, polydactyly,

Table 2.2 Rib deformities that cause chest wall deformitied are classified as congenital or acquired

Congenital deformities	Acquired deformities
Cervical ribs	**Metabolic disorders** Rickets Hyperparathyroidism
Altered rib count	**Iatrogenic causes**
Rib size variations Thanatophoric dysplasia Jeune Asphyxiating Thoracic Dysplasia Ellis-van Creveld Chondroectodermal dysplasia Achondroplasia	**Trauma** Child abuse Surgical trauma
Abnormal rib morphology Cerebrocostomandibular syndrome Rib notching Bifid ribs Slender ribs Trisomy 18 syndrome Neurofibromatosis	**Neoplastic disorders** Enchondromatosis Osteochondroma Langerhans cell histiocytosis Xanthogranuloma Ewing Sarcoma Primitive Neuroectodermal Tumor
	Infections

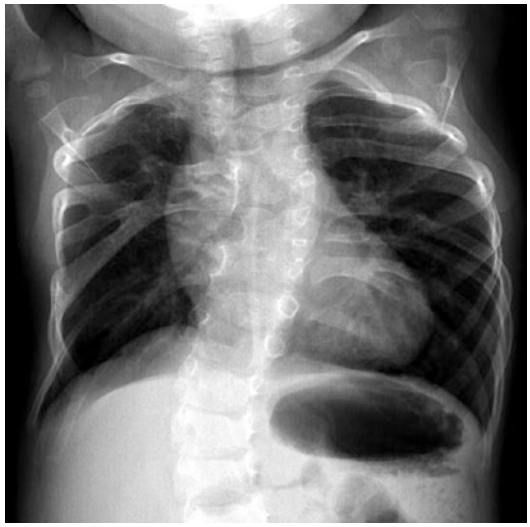


Fig. 2.15 Chest film showing alteration in the number of ribs and dyplasia of the right thoracic cage in a 2-year old with VATER association

and cone epiphyses are characteristic. The proximal humeral and femoral epiphyses may be prematurely ossified at birth. Diagnostic work up is used to rule out Jeune dysplasia and the short rib-polydactyly syndromes.

(d) ***Achondroplasia*** — In achondroplasia, the ribs are short and wide with concavity at the end of the ribs. The inheritance is autosomal dominant, with spontaneous mutations in 80 % of cases. Associated diagnostic features include macrocephaly, depression of the nasion, craniocaudal narrowing of the interpediculate spaces, square iliac bones, and a narrow pelvic inlet.

4. **Abnormal rib morphology**

(a) ***Cerebrocostomandibular Syndrome***.—Cerebrocostomandibular syndrome is very rare and the genetic transmission is not yet established [26]. These patients have eleven pairs of ribs which are characterized by abnormal costovertebral articulations and posterior ossification gaps that bear similarities to fractures. The posterior ossification gaps ossify in the adult age. Respiratory distress is common in neonates due to flail chest and airway abnormalities. Other features of this syndrome are microcephaly, micrognathia, and congenital heart disease.

(b) ***Rib notching***.—Concave notches are found on the inferior rib surface and are generally considered normal. Pathologic notching is vascular or neural in origin. Notching associated with aortic coarctation usually affects ribs 4–8 and is rare in affected children before the age of 8 years [30]. Rib notching is also encountered in neurofibromatosis and thalassemia.

Notching is unilateral, and on the right sided, with coarctation proximal to the origin of the left subclavian artery and a postoperative Blalock-Taussig shunt.

(c) **Bifid ribs**: Gorlin basal cell nevus syndrome is a rare, autosomal dominant syndrome characterized by multiple nevoid basal cell carcinomas that arise in childhood jaw cysts and bifid ribs. The fourth rib is the most commonly bifid. A single bifid rib is most commonly a normal incidental finding and may be detected as a palpable chest wall mass. Other rib anomalies include agenesis, supernumerary ribs, distorted shape, and fusion of adjacent ribs [31]. The lateral clavicle may be deficient. This condition is associated with features such as mandibular hypoplasia, macrocephaly, and mental retardation.

(d) **Slender Ribs**

- ***Trisomy 18 Syndrome*** — In this syndrome, eleven rib pairs are present, and they are classically hypoplastic and thin. The short sternum in these patients presents as a typical shield deformity of the chest. These children are spastic and neurologically delayed. The fingers are maintained in ulnar deviation with flexion deformities. Associated findings are dolichocephaly, micrognathia, and various multisystem anomalies. Other conditions that can cause such changes include Cockayne syndrome, trisomy 13 syndrome, Werdnig-Hoffmann disease and osteogenesis imperfecta.
- ***Neurofibromatosis***.—Peripheral neural tumors are the characteristics of neurofibromatosis, an autosomal dominant neurocutaneous syndrome. The ribs are slender; twisted, deformed, and separated by neurofibromas that arise from the intercostal nerves [32]. The mesenchymal abnormality in neurofibromatosis results in modeling deformity and even stranger appearance of the ribs. Intercostal neuromas can cause notching of the inferior surface of the ribs. Sphenoid dysplasia, modeling deformity of the long bones, and posterior vertebral body scalloping are associated findings. Cleidocranial dysplasia, myotonic dystrophy, Melnick-Needles osteodysplasty, and Werdnig-Hoffmann disease should also be considered when diagnosing these patienst.

Acquired Deformities

Acquired deformities can be grouped under those resulting from metabolic disorders, infections, neoplasms, trauma and iatrogenic causes.

1. **Metabolic Disorders**
   - ***Rickets***— Rickets in present times is more common with very low-birth-weight prematurity and secondary to renal or hepatic dysfunction and anticonvulsant administration. Delayed ossification of the osteoid matrix in immature bones is marked in the metaphyses around the knees, wrists and the rib ends. The hyperplastic rib osteoid may be palpable on the chest wall. Other differential diagnostic considerations for rib cupping and flaring include achondroplasia, hypophosphatasia, and metaphyseal chondrodysplasia.
   - ***Hyperparathyroidism***.—Hyperparathyroidism occurs most commonly secondary to chronic renal failure. Hyperparathyroidism results in subperiosteal and endosteal bone resorption. Brown tumors are a rare manifestation of hyperparathyroidism, in which well-defined lucent areas of lysis occur without adjacent reactive bone formation. The other differential diagnostic considerations are Langerhans cell histiocytosis, enchondromas and fibrous dysplasia.
2. **Iatrogenic Conditions**
   Generalized new bone formation occurs after prostaglandin administration in children with cyanotic heart disease. The formation has been reported to be as early as 6 days but is more evident after 30–40 days therapy. The changes are usually more symmetrical than those of infantile cortical hyperostosis (Caffey

disease or syndrome). After treatment, periosteal new bone becomes incorporated and remodeled [33]. Differential diagnostic possibilities include child abuse, extracorporeal membrane oxygenation, and infantile cortical hyperostosis.

3. **Trauma**
   - ***Child Abuse***.—Fractures of the ribs with child abuse are considered highly specific for child abuse [34]. Although abuse fractures can occur in any part of the ribs, they are more common in the posterior part because of the significant posterior levering force inflicted in the abuse situation. Healing fractures are more easily detectable than in the acute phase. The most important differential diagnoses to consider are osteogenesis imperfecta, long-term ventilator therapy in prematurity, birth trauma, and Menkes syndrome.
   - ***Surgical Changes***—Rib resections and deformities that result from thoracotomy are asymptomatic. However, fusion of the ribs that can lead to scoliosis and restrict the chest wall expansion may require surgical intervention. Fused ribs are also seen in patients with Gorlin syndrome.
4. **Infections**

   Rib osteomyelitis is most commonly acquired from an infectious source with the thoracic cavity, such as empyema or pneumonia. Although hematogenous spread is rare, but when present it manifests anteriorly near the costochondral junction or posteriorly near the rib angle [35]. The transformation resembles that of typical chronic long bone infection, in which there is formation of a sequestrum in the presence of periosteal fresh bone formation.
5. **Neoplasms**

   The incidence of primary neoplasms originating in the thoracic wall is 5–10 % of all bone tumors [36]. Large tumors extend both towards the inside and outside of the thorax; with the soft-tissue mass that extends externally being clinically obvious. As a result, primary rib tumors are significantly larger than bone tumors in other locations [37].
   - ***Enchondromatosis*** — These are characterized by the presence of medullary cartilaginous bone tumors, which are further subdivided by the presence of hemangiomas (Maffucci syndrome) or their absence (Ollier disease). Rib lesions are common in enchondromatosis. Enchondromas occur in association with a variety of systemic malignancies and are themselves associated with a 25 % prevalence of malignant degeneration [38, 39]. Differential diagnostic considerations are Langerhans cell histiocytosis and osteomyelitis.
   - ***Osteochondroma***.—Exostoses are benign bony masses with a cartilage cover. These can be solitary or associated with multiple hereditary exostoses. It has been estimated that rib osteochondromas arise in almost 50 % of patients with multiple hereditary exostoses [40]. Those rib exostoses that project externally are palpable on the chest wall. Internal exostoses can be asymptomatic [41]. Osteochondromas can mimic pulmonary nodules; calcified osteochondromas resemble granulomas.
   - ***Langerhans Cell Histiocytosis***.—Langerhans cell histiocytosis is presumed to be associated with immune dysfunction. Although most bone lesions are asymptomatic, some may present as a painful soft-tissue mass [42]. The ribs are commonly involved, and multiple bone involvement is not unusual. Differential diagnostic considerations include metastases, hyperparathyroidism (brown tumors), Ewing sarcoma, Askin tumor and lymphoma.
   - ***Xanthogranuloma***.—Xanthogranulomas are rare benign tumors that occur in the ribs and other flat bones. They are invariably solitary lesions and are almost twice as common in male patients [43]. The tumor has an excellent prognosis, and complete or partial removal is curative. The differential diagnosis includes aneurysmal bone cyst, hyperparathyroidism (brown tumor), and Langerhans cell histiocytosis.
   - ***Ewing Sarcoma***.—Ewing sarcoma is an extremely aggressive malignant tumor that occurs in adolescents and young adults. It is the most common malignant tumor that affects the ribs of children and adolescents [37]. Most

Ewing sarcomas have an associated soft-tissue mass that is significantly larger than the intraosseous tumor [44]. The differential diagnosis includes osteomyelitis; Langerhans cell histiocytosis, osteosarcoma, primitive neuroectodermal tumor, metastatic neuroblastoma, and lymphoma.

- ***Primitive Neuroectodermal Tumor***.— Primitive neuroectodermal tumor or Askin tumor is a rare malignant tumor that arises from the chest wall (Fig. 2.16). This tumor very closely resembles Ewing sarcoma, and differentiation depends on detection of neurosecretory granules at electron microscopy [45]. Rib destruction occurs in over 1/4th of the patients. Differential diagnostic considerations include osteomyelitis, Langerhans cell histocytosis, osteosarcoma, Ewing sarcoma, metastasis, and lymphoma.

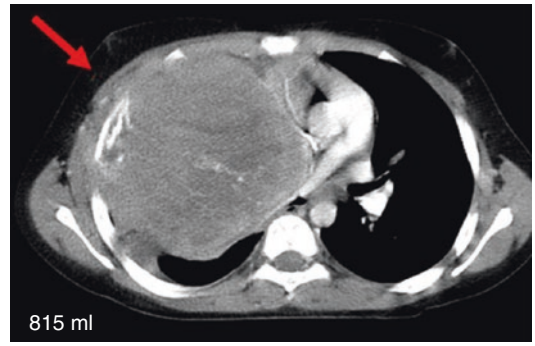


Fig. 2.16 Computed tomography image of the chest demonstrating the size and location (*red arrow*) of the Askin's tumor in a13-year old female patient

Type 4- Combined Costal Cartilage and Rib Deformities

Poland Syndrome (PS)

PS has an incidence of approximately 1:30,000 live births and is characterized by the absence or hypoplasia of the pectoralis major muscle, frequently combined with other ipsilateral abnormalities of the chest wall, breast and upper limb [46]. The defect is generally unilateral and in two thirds of cases right-sided. Very rare bilateral cases have been described [47, 48]. There is a male preponderance with a ratio of about 2:1 (Figs. 2.17 and 2.18). The etiology is unknown, but the most plausible hypothesis is the interruption of the vascular supply in subclavian and vertebral artery during embryonic life, leading to different malformations in the corresponding segments affected [49]. Beside this hypothesis, paradominant inheritance or the presence of a lethal gene survival by mosaicism have also been proposed to explain the genetic etiology of this anomaly [50, 51]. PS is usually sporadic, but the occurrence of familial cases has raised the theory of a possible transmission with an autosomal dominant pattern. Association of PS with other anomalies, as Moebius, Klippel Feil syndromes and Sprengel anomaly, has been reported [49, 52–54].

The thoracic defect is usually evident at birth in PS, but it can remain undiagnosed until the child gets older. The pectoral muscle deficiency causes an asymmetric aspect; but if there are costal anomalies associated the defect, these are more evident. In case of rib agenesis, particularly if multiple ribs are involved (generally the third and the forth rib), lung herniation and paradoxical respiratory movements are always present. Ribs can also be smaller or anomalous in patients with PS.

Anomalies like a PE or PC or both can occur concomitantly with PS, but in less than 10% of cases they require surgery. The breast region and nipple are frequently involved in these patients, which can range from a mild degree of breast hypoplasia to a complete absence of mammary gland. Associated cardiac and renal anomalies, as well as scoliosis, have been reported, but they are uncommon [53]. Dextroposition is reported frequently and is almost always associated with left PS. Patients with PS are asymptomatic, and there are have usually no limitations from the muscle defects.

Type 5- Costoverteberal Junction Deformities

- **Osteogenesis imperfecta**
  Osteogenesis imperfecta (OI) is a group of genetic disorders that mainly affect the bones [55]. There are at least eight recognized forms

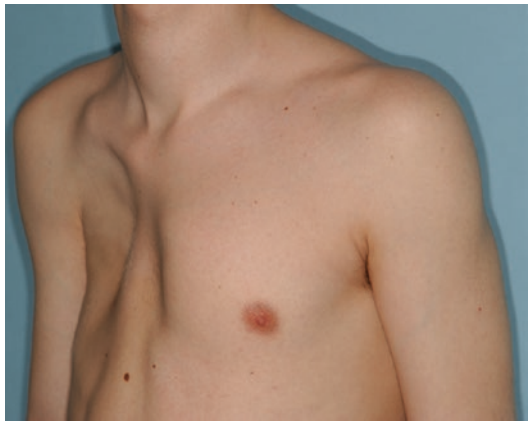


Fig. 2.17 Poland's syndrome with hypoplastic left chest wall in a 14-year old male patient

of osteogenesis imperfecta, designated type I through type VIII. The types can be distinguished by their signs and symptoms, although their characteristic features overlap. Type I is the mildest form of osteogenesis imperfecta and type II is the most severe; other types of this condition have signs and symptoms that fall somewhere between these two extremes. Increasingly, genetic factors are used to define the different forms of osteogenesis imperfecta. OI type II, can include an abnormally small, fragile rib cage and underdeveloped lungs. Infants with these abnormalities have life-threatening problems with breathing and often die shortly after birth.

- **Jarcho-Levin syndrome**
  Jarcho-Levin syndrome is a rare genetic disorder characterized by malformations of bones of the vertebrae and the ribs, respiratory insufficiency, and/or other abnormalities. Jarcho-Levin syndrome is a type of segmental costovertebral malformation [56]. Infants born with this syndrome have short necks, restricted neck motion due to abnormalities of the cervical vertebrae and short stature. In most cases, infants with Jarcho-Levin syndrome experience respiratory insufficiency and are prone to repeated respiratory infections that result in life-threatening complications. The vertebrae are fused and the ribs fail to develop properly, therefore, the chest cavity is too small to accommodate the growing lungs. There are two forms of Jarcho-Levin Syndrome that are inherited as autosomal recessive genetic traits and termed spondylocostal dysostosis type 1 (SCDO1) and spondylocostal dyostosis type 2 (SCDO2).

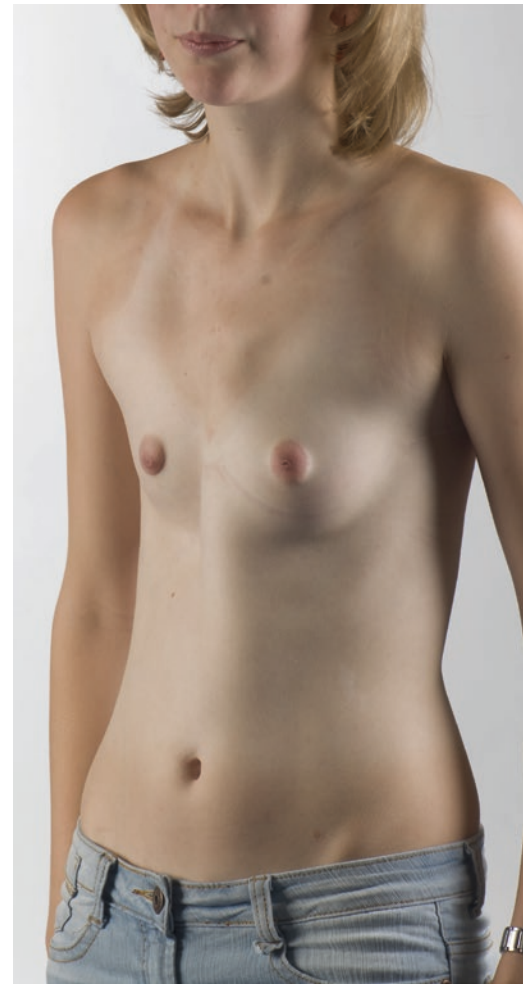


Fig. 2.18 Poland's syndrome with hypoplastic left chest wall with breast involvement in a 15-year old female patient

- **Aicardi syndrome**
  Aicardi syndrome is a rare genetic disorder first described by a French Neurologist Jean Aicardi in 1965. Aicardi syndrome is characterized by the following associations: (a) absence of the corpus callosum, either partial or complete, (b) infantile spasms, (c) lesions or "lacunae" of the retina of the eye

(lacunar chorioretinopathy), (d) other types of defects of the brain such as microcephaly, or porencephalic cysts and (e) costovertebral anomalies [57]. Aicardi syndrome only affects females, and in very rare cases, males with Klinefelter syndrome (XXY). Children are most commonly identified with Aicardi Syndrome around the age of 5 months. A significant number of these patients are girls who seem to be developing normally until around the age of 3 months, when infantile spasms commence. The known age range of affected children is from birth to the late forties.

- **Klippel-Feil Syndrome**
  Kippel-Feil Syndrome is a rare disease, initially reported in 1912 by Maurice Klippel and André Feil [58] characterized by the congenital fusion of any 2 of the 7 cervical vertebrae. The syndrome occurs in a heterogeneous group of patients unified only by the presence of a congenital defect in the formation or segmentation of the cervical spine. Klippel-Feil syndrome has following characteristics (a) scoliosis (b) spina bifida, (c) anomalies of the kidneys and ribs, (d) cleft palate (e) heart malformations (f) short stature. The disorder also may be associated with abnormalities of the head and face, skeleton, sex organs, muscles, brain and spinal cord, arms, legs, fingers and heart defects. These heart defects often lead to a shortened life expectancy, the average being 35–45 years of age among males and 40–50 among women. This condition is similar to the heart failure seen in gigantism.

Summary

CWDs encompass a wide range of anomalies involving the musculosketal system of the thorax. Enormous syndromes are associated with these deformities, detailed information on which is beyond the scope of this chapter. The classification of CWDs based on anatomic topography encompasses the spectrum of CWDs in a methodical way with attention given to each structure comprising the chest wall. Correct identification of the type of deformity according to its classification, an accurate diagnostic assessment and selection of the proper management option among those present, are the key features in the modern day approach to the treatment of CWDs.

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