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Mini-symposium: Chest Wall Disease

Primary Thoraco-spinal Disorders of Childhood

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EDUCATIONAL AIMS

- Understand the spectrum of disorders that produce early onset thoraco-spinal deformities
- Understand the pathophysiologic consequences of severe and progressive deformities
- Understand the impact of developing surgical devices and procedures on respiratory function in children with these disorders
- Understand the evolving role of pediatric pulmonary specialty centers in the management of children with these rare disorders.

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SUMMARY

Primary structural deformities of the spine and thorax were at one time rare and reportable in case series. With the development of new "growth friendly" implantable devices, children with these disorders are living longer and receiving both surgical and pulmonary care. As a result, there has been growing interest in the functional cardiopulmonary consequences of these deformities, the current surgical and non-surgical treatments, and the role of long-term supportive care. This article reviews current literature in this rapidly changing field, where new devices are developed and outcomes are changing. The respiratory consequences of early-onset thoraco-spinal disorders are emphasized and the roles of the pulmonologist and surgeons are discussed. There are more questions than answers as no long-term outcome data yet exists.

INTRODUCTION

Spine and thoracic cage disorders have often been considered separately in children. However, these structures interact with each other, modifying both structure and function of one another. The spine influences rib orientation and motion. The ribs, when fused, promote scoliosis. The thorax determines the insertion points of the diaphragm and hence its configuration and function. Unilateral pulmonary hypoplasia, as occurs with diaphragmatic hernias, increases the likelihood of scoliosis. Not all of these interactions result from congenital deformities. Thoracotomies early in childhood increase the risk for scoliosis later in childhood as do rib resections due to tumor resections. Not all deformities lead to impaired lung function, e.g. pectus carinatum. However, many do and can be lethal in some cases.

Another way to think of these disorders is to consider how the thoraco-abdominal cylinder affects breathing. The torso provides a stable and protected environment for the lung as well as the inspiratory and expiratory pumps that power breathing, vocalization, and host defenses such as sighing, sneezing, and coughing. Disorders and deformities of the bones and muscles of the thorax lead to restrictive respiratory disease [1]. Scoliosis has more respiratory consequences the higher the apex of the deformity resides among thoracic vertebrae [2]. However, even scoliosis in the thoracolumbar region can produce pelvic obliquity, raising one side of the pelvis up into the hemi-thorax in severe cases, producing restrictive changes from below the diaphragm. This chapter addresses the consequences of structural deformities of the spine and thorax rather than the secondary changes due to underlying primary neuromuscular weakness or spasticity. Infants and children with *primary* thoracic and/or spinal deformities may also have pulmonary co-morbidities, such as upper

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Abbreviations: EOS, early onset scoliosis; AIS, adolescent idiopathic scoliosis; AP, anterior-posterior; FRC, functional residual capacity; TLC, total lung capacity; RV, residual volume; MIP, maximum inspiratory pressure; FVC, forced vital capacity; BIPAP, bilevel positive airway pressure; CPAP, continuous positive airway pressure.

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airway obstruction or recurrent pneumonia, which further complicate the pathophysiology produced by these deformities. This review assumes that those co-morbidities have been diagnosed and minimized. Finally, some deformities are part of underlying syndromes that involve multiple organs, such as VACTERL syndrome, or multiple skeletal structures, such as arthrogryposis. The additional impacts of these extra-pulmonary anomalies are not discussed here.

Spine deformities can begin at all ages. Early onset scoliosis (EOS) is classified as infantile (0-3 years) or juvenile (3-10 years) idiopathic scoliosis as opposed to the more common adolescent (>10 years) idiopathic scoliosis (AIS). Congenital scoliosis, unlike idiopathic scoliosis, includes vertebral structural deformities such as hemivertebrae and unsegmented vertebrae and is part of the EOS spectrum. EOS is more likely to progress than AIS over time and creates more severe deformities [3]. Untreated EOS has been linked to shortened survival in adults older than 40 years of age based on a national health database in Sweden [4]. That report used onset before age 8 years as the definition of EOS but it also included people with underlying neuromuscular disease.

Some thoraco-spinal deformities are categorized as intrathoracic "volume depletion" deformities, such as Jarcho-Levin syndrome (i.e. spondylocostal dysostosis and spondylothoracic dyplasia) and Jeune's syndrome. Syndromes that can produce hypoplastic thoraces, including both spine and rib deformities listed in Table 1 were extracted from an excellent review about chest wall abnormalities [5]. These deformities lead to reduced chest wall circumference or shortened vertebral height. They produce prenatal pulmonary hypoplasia and often present with neonatal respiratory distress. They are associated with increased mortality in infancy and childhood due to respiratory failure if untreated [6]. However, there is a continuum of severity of thoracic cage hypoplasia among these conditions and mortality due to respiratory dysfunction is not invariable [7]. Up to 40% of children with Jeune's syndrome surviving the first 2 years of life will die of renal disease later in life [6]. People with spondylocostal dysostosis have survived well into adulthood [8].

Children with hypoplastic thoraces have reductions in intrathoracic volume and hence lung volume. Clinically they present with reduced chest wall excursion and tachypnea. The small lung volume, e.g. functional residual capacity, predisposes them to rapid and significant episodes of hypoxemia whenever superimposed events impose on breathing [9]. This occurs with acute respiratory infections and with upper airway pauses or obstruction during sleep. Children with these disorders eat in small proportions and often present with failure to thrive in infancy [10]. Processes that distend the abdomen, such as increased abdominal air or stool, produce an added restrictive process and further tachypnea or distress. Attention to constipation is a part of clinical management in severe cases.

Table 1
Conditions associated with hypoplastic thorax.

Achondrogenesis
Achondroplasia
Camptomelic Dysplasia
Cerebro-Costo-Mandibular syndrome
Chondroectodermal dysplasia
Cleidocranial dysostosis
Diastrophic dysplasia
Fibrochondrogenesis
Hypophosphatasia
Jarcho-Levin syndrome
Jeune syndrome
Lethal multiple pterygium syndrome
Melnick-Needles syndrome
Metaphyseal chondrodysplasias
Metatropic Dysplasia
Osteogenesis imperfect
Oto-Palato-Digital syndrome
Progeria syndrome
Pseudoachondroplasia Sponyloepiphyseal dysplasia
Short rib syndrome

Scoliosis also reduces intrathoracic volume as well as intrathoracic distortion and disorientation [11,12]. Although the severity of scoliosis is often depicted by the Cobb angle, which quantifies lateral displacement of the vertebrae from a single anterior-posterior (AP) radiograph of the spine, this single measurement oversimplifies the complexity of the deformed spine and ribs. Pertinent skeletal features of an individual with scoliosis include scoliosis (lateral displacement in the coronal plane), kyphosis and lordosis (in the sagittal plane), rotation of the spine, vertebral level where the apex of the scoliotic curve occurs, length of the spine deformity (number of vertebrae involved), and associated rib anomalies, such as fused ribs or absent ribs. Scoliosis is therefore a complex three-dimensional structural deformity of the vertebrae, ribs, and sternum that varies tremendously from patient to patient. It is not surprising that correlations between different indices of respiratory function do not correlated with the Cobb angle alone and that the Cobb angle cannot be used as a surrogate to depict respiratory function in an individual patient [10].

In addition to the structural complexity at initial presentation, scoliosis can progress over time. This is particularly true during periods of rapid growth, such as adolescence. This also occurs with growth hormone therapy [13]. In addition, certain combinations of skeletal deformities, such as block vertebrae and unilateral fused ribs, are more likely to progress substantially during childhood [14]. The nature of the progression also differs between individuals with the same deformity. Some patients will have more significant rotation than lateral displacement over time. Children with AIS who have scoliosis with Cobb angles of >40-50 degrees are at significant risk to progress without brace therapy and much less often despite brace therapy [15]. These prognostications are dependent on degree of skeletal maturity, as measured by the Risser sign radiographically [16]. Because the rate of progression is variable, serial monitoring of both structure and respiratory function is necessary in order to provide timely intervention. A major challenge for pulmonologists is how best to monitor respiratory functions across all age groups serially, including in very young children.

RESPIRATORY FUNCTION ABNORMALITIES

The respiratory abnormalities that accompany these deformities include reduced lung volumes, reduced chest wall compliance and excursion, and inefficient respiratory muscle function. In half of children with EOS, there is also progressive asymmetry in lung function due to intrathoracic deformity of the lungs on the concave and convex sides of the scoliotic curve [17]. This asymmetry has been demonstrated using ventilation and lung perfusion scans. These events lead to a clinical picture of progressive restrictive pulmonary disease, increased respiratory work, and eventual hypercapnic respiratory failure. Additional obstructive changes in lung function do occur in a 10-30% of children, usually produced by mainstem bronchial tethering and compression between the vertebrae and solid mediastinal organs [18]. Reversible airway obstruction, e.g. asthma, should always be ruled out but compression of the major bronchi will not produce reversible airway disease.

The reduction in lung volumes associated with chest wall restriction differs from the pattern seen with restrictive pulmonary parenchymal disorders. This is illustrated in Figure 1. Primary thoraco-spinal deformities reduce vital capacity more than functional residual capacity (FRC), residual volume, or total lung