Epidemiology, Classification, and Natural History

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Introduction

Early-onset scoliosis (EOS) includes a heterogeneous group of conditions with varied presentations, progression, and natural history.\(^1\) The etiology of EOS can be idiopathic, or due to a variety of scoliosis-associated syndromes such as congenital vertebral anomalies or neuromuscular diseases.\(^2\) The Global Spine Study Group, Children Spine Study Group, and Scoliosis Research Society have defined EOS as scoliosis with age of onset less than 10 years, irrespective of the etiology.\(^3\)

Management of EOS requires a thorough knowledge of the normal development of the spine and also the immature lung. The greatest potential for progression of spinal deformity is during periods of rapid growth, i.e., during the first few years of life, and again during the adolescent growth spurt. The critical time in the pulmonary development of a child is within the first few years of life, when the number of alveoli and the lung volume increase rapidly. Untreated EOS may show malignant curve progression resulting in a horrendous spinal deformity. The accompanying changes in the shape and volume of the thoracic cage can lead to restrictive lung disease characterized by small lung volumes, reduced chest wall compliance, and respiratory muscle dysfunction (Fig. 1.1). In patients with severe scoliosis, the thorax may be unable to support normal respiratory function and lung development. This condition is called thoracic insufficiency syndrome (TIS).\(^4\) This has been associated with poorer quality of life scores than those of childhood epilepsy, heart disease, and cancer.

Although the periods of rapid growth can result in worsening of deformity, they also offer a window of opportunity for correcting the spinal deformity. Various treatment modalities such as bracing, serial casting, growth rods, magnetic growth rods, epiphysiodesis, and vertical expandable prosthetic titanium rib (VEPTR)
attempt to guide the growth of the spine with the goal of correcting and/or limiting spinal deformity. However, there are no definitive guidelines or even consensus on the indications, timing, and techniques that yield the best outcomes in these children.

**Epidemiology and Natural History**

The true prevalence of EOS is not known and it varies depending on the etiology. The incidence of EOS is four times more in Europe when compared with the United States (1–2 per 10,000), and is believed to be much more in Asia, especially Southern India.

Infantile idiopathic scoliosis (IIS) constitutes only 1% of the idiopathic scoliosis population. IIS is more common in males, tends to be left sided, and 70 to 90% occur in the mid to lower thoracic spine. IIS curves can be subclassified as resolving if the curve reduces in magnitude or disappears with growth, or progressive if the curve tends to increase in size with growth. The spontaneous resolution rate reported in literature varies between 20% and 80%. James et al studied 212 patients with IIS and reported spontaneous resolution in 31% of children. 135 of the 212 children (69%) showed aggressive curve progression with 50 children developing curves > 70 degrees and 26 children developing deformities > 100 degrees. Mehta measured the difference in the angle between the rib and the vertebral body on the convexity and concavity at the apex of the scoliosis (RVAD). She found that when the RVAD was less than 20 degrees, the curve resolved in 90% of children, while the curve was less likely to resolve if the RVAD was > 20 degrees. While RVAD has not been validated in noninfantile EOS, it continues to be used as an indicator of curve severity and progression of deformity. Untreated progressive curves have been reported to reach a Cobb angle of > 120°, resulting in poor cardiopulmonary outcomes, and shorter life expectancy.

Juvenile idiopathic scoliosis accounts for 12 to 21% of idiopathic scoliosis. They tend to be more common in females, with predominance of right-sided thoracic curve and double major curves. Juvenile onset curves progress in 55 to 71% patients (Fig. 1.2). The progression is at a slow to moderate rate and is noted with earlier age of onset and bigger curves at presentation.

Congenital scoliosis seems to be the most prevalent among all the causes of EOS. The prevalence rate of congenital scoliosis is approximately 1 in 1,000 live births and is more common in girls. The progression of congenital scoliosis depends on the type and location of anomaly and the growth potential of the child (Figs. 1.3 and 1.4). The natural history of congenital scoliosis has been reported by Winter and McMaster (Fig. 1.5). The deterioration of the curve is less in the upper thoracic spine, moderate in mid-thoracic region, and worst in the thoracolumbar region. Block vertebra and bilateral failure of segmentation are the benign forms of anomaly. Wedge vertebrae, hemivertebrae (Fig. 1.6), and unilateral unsegmented bar cause more severe deformity, respectively.

The incidence of scoliosis is 15 to 28% amongst children with cerebral palsy, with institutionalized patients, spastic quadriplegic CP, and children with limited motor function, being most commonly affected.

The natural history of untreated EOS is associated with significant morbidity and often profound cardiopulmonary compromise, including...
Fig. 1.3 Classification of congenital scoliosis showing failures of formation and segmentation. Incarcerated hemivertebrae have compensatory deformities proximal and distal to it. The highest risk for progression lies with a hemivertebrae opposite a unilateral bar.

<table>
<thead>
<tr>
<th>Defects of vertebral-body segmentation</th>
<th>Defects of vertebral-body formation</th>
<th>Mixed anomalies</th>
</tr>
</thead>
<tbody>
<tr>
<td>Partial</td>
<td>Anterior and unilateral aplasia</td>
<td>Anterior and median aplasia</td>
</tr>
<tr>
<td>Anterior unsegmented bar</td>
<td>Posterolateral quadrant vertebra</td>
<td>Butterfly vertebra</td>
</tr>
<tr>
<td>Complete</td>
<td>Anterior aplasia</td>
<td>Anterior hypoplasia</td>
</tr>
<tr>
<td>Block vertebra</td>
<td>Posterior hemivertebra</td>
<td>Wedge vertebra</td>
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</tbody>
</table>

Fig. 1.4 Classification of congenital kyphosis. A high percentage of true congenital kyphosis deformities will progress and can lead to paraplegia especially if in the thoracic spine.
Early Onset Scoliosis

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<table>
<thead>
<tr>
<th>Site of curvature</th>
<th>Type of congenital anomaly</th>
<th>Unilateral unsegmented bar and contralateral hemivertebra</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>Block vertebra</td>
<td>Wedge vertebra</td>
</tr>
<tr>
<td>Upper thoracic</td>
<td>&lt; 1°–1°</td>
<td>1°–2°</td>
</tr>
<tr>
<td>Lower thoracic</td>
<td>&lt; 1°–1°</td>
<td>2°–3°</td>
</tr>
<tr>
<td>Thoracolumbar</td>
<td>&lt; 1°–1°</td>
<td>1.5°–2°</td>
</tr>
<tr>
<td>Lumbar</td>
<td>&lt; 1°–1°</td>
<td>&lt; 1°–1°</td>
</tr>
<tr>
<td>Lumbosacral</td>
<td>&lt; 1°–1.5°</td>
<td>&lt; 1°–1°</td>
</tr>
</tbody>
</table>

Fig. 1.5 Risk of progression of different types of congenital vertebral malformations as compiled by McMaster. Highest progression was seen in unilateral unsegmented bar and contralateral hemivertebrae.

Fig. 1.6 A 4-year-old male child with congenital left thoracolumbar scoliosis and hemivertebrae with a Cobb’s angle of 56.3° from T8 to L3.

respiratory failure and cor pulmonale (Fig. 1.7).

A Swedish study comparing expected population death rates demonstrated more than twice the mortality rate by age 40 in patients with EOS, compared with that of the general population. Pehrsson et al reported that in patients with untreated scoliosis, the mortality rate was maximum in infantile-onset scoliosis followed by juvenile-onset scoliosis. The main cause of death was respiratory failure (40%).19,20 The average age at death was 54 years but some patients died as young as 16 years. Postmortem findings of these children showed very small lungs with markedly decreased numbers of alveoli. Consequently, the fundamental principle of treating EOS is to foster normal respiratory development and maximize spinal growth, while preventing additional deformity that can lead to TIS.
Dickson classified scoliosis in children as early (≤ 5 years) and late (> 5 years) onset. The early onset group coincided with the first growth spurt and has a higher risk of developing significant cardiopulmonary complications in a progressive curve. The Scoliosis Research Society has modified the EOS criteria from the original description given by Dickson, and now EOS includes all scoliotic deformities that occur and present before 10 years of age. The reason being that up to the age of 10 years, the children receive growth-friendly interventions, while children above 10 years are more likely to receive definitive fusion as the primary intervention. Use of the universal terminology is essential for research, communication, and teaching.

Based on the etiology, EOS could be classified as:
1. Idiopathic scoliosis: without a known attributable cause.
   a. Infantile scoliosis: Birth till 3 years of age.
   b. Juvenile scoliosis: Four to 10 years of age.
2. Congenital: Structural abnormality of the spine or thorax present at birth due to:
   a. Failure of formation.
   b. Failure of segmentation.
   c. Mixed.
3. Neuromuscular: Abnormalities in muscular tone leading to scoliosis.
   a. Neuropathic causes:
      i. Upper motor neuron lesions such as CP, spinocerebellar degeneration (Fredrick ataxia, Charcot–Marie–Tooth disease), syringomyelia, spinal cord tumors, and spinal trauma.
      ii. Lower motor neuron lesions such as poliomyelitis, spinal muscular atrophy, and myelomeningocele.
   b. Myopathic causes: arthrogryposis, muscular dystrophies (Duchene, limb-girdle, facioscapulohumeral), congenital hypotonia, and myotonia dystrophica.
4. Syndromic: Includes any other syndrome-associated scoliosis. It can be subdivided as:
   a. Connective tissue disorders (Marfan’s syndrome, Ehlers–Danlos syndrome).
   b. Osteochondrodystrophies (dystrophic dysplasia, mucopolysaccharidosis, spndyloepiphyseal dysplasia, multiple epiphyseal dysplasia, achondroplasia).
   c. Metabolic causes (Rickets, osteogenesis imperfecta).

Fig. 1.7 A 30-year-old male with neglected early-onset scoliosis with poor pulmonary reserve and effort intolerance.
**Associated Conditions/Syndromes**

A significant percentage of patients (61%) with congenital scoliosis have an associated anomaly in other organ systems, which may appear independently or as part of a syndrome. VACTERL (vertebral, anorectal, cardiac, tracheoesophageal, renal, and limb) syndrome is often found to be associated with congenital scoliosis.22–24

Syndromic scoliosis describes several inherited genetic and nongenetic conditions with scoliosis as one of the characteristic features. In the syndromic child, the underlying cause of scoliosis may be multifactorial and includes skeletal abnormalities, poor muscle tone, and connective tissue abnormality with laxity of ligaments and joints. The progression of the spinal curvature is highly variable and is based on the syndrome.

**The Classification for Early-Onset Scoliosis (C-EOS)**

EOS is a complex disease and till recently did not have a classification system. Recently, the Growing Spine Study Group and the Children Spine Study Group have suggested a C-EOS.25 The C-EOS consists of continuous age prefix, etiology, major curve angle, kyphosis, and an optional progression modifier. The etiology subgroups are listed from highest to lowest priority from congenital/structural, neuromuscular, syndromic, and lastly, idiopathic. When there are more than one diagnoses, the highest priority subgroup determines the etiological assignment.

1. **Major curve angle** (measurement of major spinal curve in position of most gravity) is divided into four groups:
   - $< 20^\circ$
   - $20^\circ$ to $50^\circ$
   - $51^\circ$ to $90^\circ$
   - $> 90^\circ$

2. **Kyphosis** (maximum measurable kyphosis between any two levels) differentiated into:
   - Hypokyphotic ($< 20^\circ$)
   - Normokyphotic ($20^\circ$ to $50^\circ$)
   - Hyperkyphotic ($> 50^\circ$)

3. **Annual progression ratio modifier** (optional): progression per year with minimum of 6 months interval between observations (Table 1.1).

This classification has the highest inter- and intraobserver validity. A treatment protocol based on this classification is yet to be designed. The classification is more commonly used for discussion among medical fraternity and needs further refinement in future to give guidelines regarding the treatment strategies and prognosis. A three-dimensional (3D) classification is in the making, but with the limitation of lack of sophisticated instruments and software needed for it.26

**Key Points**

- Scoliosis with age of onset less than 10 years, irrespective of the etiology, is termed as EOS.
- Causes of EOS include congenital, syndromic, neuromuscular, and idiopathic.

**Table 1.1 C-EOS classification**

<table>
<thead>
<tr>
<th>Age</th>
<th>Etiology</th>
<th>Major curve</th>
<th>Kyphosis</th>
<th>APR modifier</th>
</tr>
</thead>
<tbody>
<tr>
<td>Continuous prefix</td>
<td>Congenital/Structural</td>
<td>1: $&lt; 20^\circ$</td>
<td>(−): $&lt; 20^\circ$</td>
<td>P0: $&lt; 10^\circ$/y</td>
</tr>
<tr>
<td></td>
<td>Neuromuscular</td>
<td>2: $20^\circ$–$50^\circ$</td>
<td>N: $20^\circ$–$50^\circ$</td>
<td>P1: $10^\circ$–$20^\circ$/y</td>
</tr>
<tr>
<td></td>
<td>Syndromic</td>
<td>3: $50^\circ$–$90^\circ$</td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td>Idiopathic</td>
<td>4: $&gt; 90^\circ$</td>
<td>(+): $&gt; 50^\circ$</td>
<td>P2: $&gt; 20^\circ$/y</td>
</tr>
</tbody>
</table>

Abbreviations: C-EOS, Classification for Early-Onset Scoliosis; APR, annual progression ratio.
• Congenital scoliosis occurs in 1 in 1,000 live births, and is associated with a defect in another organ system in 61% patients.

• Infantile and juvenile idiopathic scoliosis account for 1% and 12 to 21% of idiopathic scoliosis, respectively.

• The significance of EOS is that relentless progression of the curve with growth can result in significant deformity, a short stature, small and deformed thoracic cage with limited lung function, especially when the age of onset is below 5 years.

• Natural history of EOS is dictated by the etiology and age of onset.

References


