7 Congenital Scoliosis

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Introduction

Congenital spinal deformity occurs in approximately 1 in 1,000 live births, with scoliosis being the most commonly identified deformity (80%). The deformities usually arise sporadically and the rate of transmission is quoted as 1%. There is, however, an association with the Homeobox genes of the Hox class.[1,2]

Congenital scoliosis is defined as a lateral curvature with associated rotation of the spine caused by vertebral anomalies that produce a coronal plane deformity. The anomalies are present at birth but it may take several years for the curvature to develop.

The anomalies result in a spectrum of deformity from a very benign curve with low potential for progression to a much more severe curve that can rapidly progress and cause significant functional and cosmetic complications.

Embryology and Etiology

Vertebral anomalies form around 4 to 6 weeks gestation. Embryologic development of the spine occurs around the same time as many organs and therefore it is not surprising that vertebral anomalies are often associated with abnormalities in other organ system.[3,4] Sixty percent of patients with a vertebral anomaly will have another issue.[5] Most commonly, the genitourinary tract is affected with issues such as renal agenesis, ectopic or duplex kidney, and reflux. Other spinal cord problems can also coexist; the most common being a tethered cord, diastematomyelia (present in 5–20%), and syringomyelia. Around 50% patients with a unilateral unsegmented bar and same level contralateral vertebra have an associated neural axial abnormality.

The acronym VACTERL (V—vertebral anomalies, A—anal atresia, C—cardiac defect, TE—tracheoesophageal fistula, R—renal defects, L—limb defect) is a group of anomalies which are all linked. These systems should all be investigated as part of the routine work-up of a new patient presenting with a congenital scoliosis.

Environmental factors have been linked with the development of congenital scoliosis in animal studies; hypoxia and vitamin A deficiency have predominantly been seen.[6] Further investigation is being undertaken in this field.

Pearls

- Scoliosis genetics are not fully understood.
- Both genetic and environmental factors are considered to be involved.

Classification (Fig. 7.1)

Two main types of vertebral anomaly occur: failure of formation (type I) and failure of segmentation (type II) (Fig. 7.1). A third group is often included in this classification—a mixed pattern. We will discuss these further.

This, commonly used, classification system[7] focusses on the anterior anatomy only, and therefore when deciding to operate, an understanding of the posterior anatomy is essential, which can be achieved with three-dimensional imaging (discussed later).

Failure of formation can be partial or complete causing either a wedged vertebra

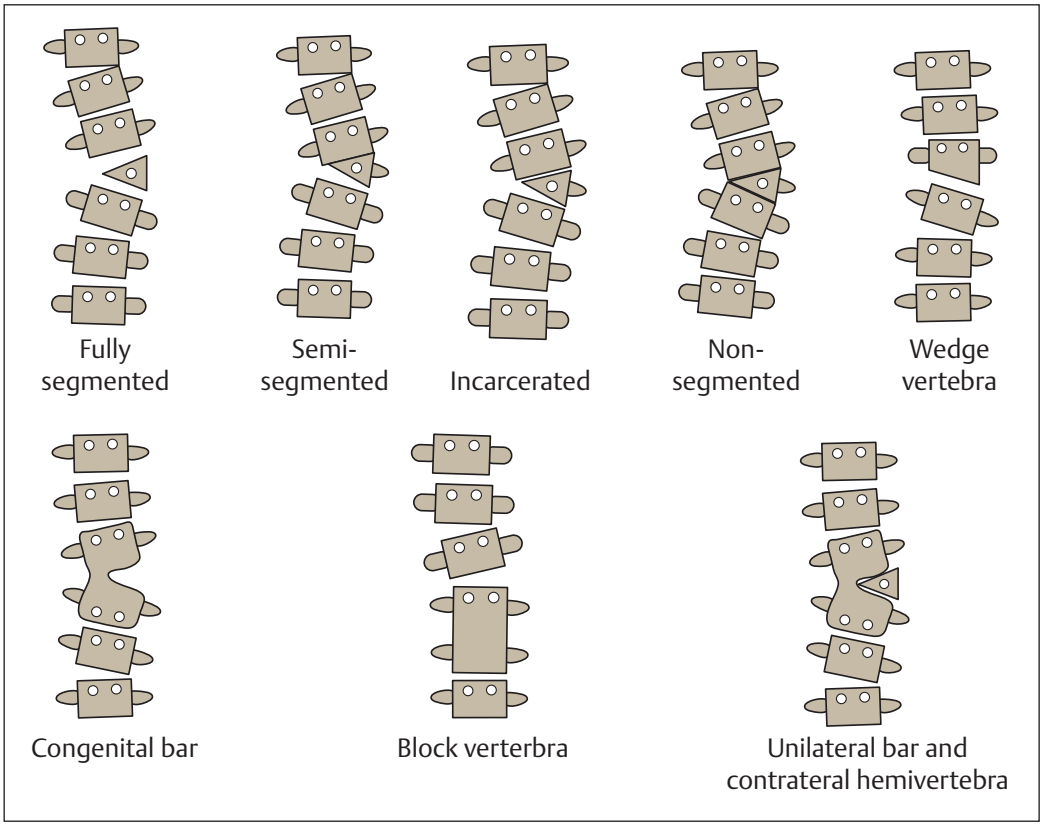

Fig. 7.1 Pictorial representation of the types of formation/ segmentation deformities.

with intact pedicles or a hemivertebra with a single pedicle.

Wedge vertebra—This can produce asymmetrical growth.

Fully segmented hemivertebra—The adjacent vertebrae shape is normal and both the superior and inferior end plates of the hemivertebra have growth potential. Rate of progression is approximately 1 to 2 degrees per year.

Semisegmented hemivertebra—The hemivertebra is fused at one end with the adjacent vertebra and so there is only growth potential at the opposite end. This produces a tilting of the spine and a more slowly progressing curve.

Incarcerated hemivertebra—The hemivertebra has growth potential both superiorly and inferiorly but this is compensated for by the adjacent levels. There are abnormal looking vertebrae on either side of the hemivertebra. There is very little deformity associated with this type of anomaly.

Nonsegmented hemivertebra—There is no growth potential in these cases as the hemivertebra is fused with the adjacent levels on either side; therefore, there is no progression in deformity.

Failure of segmentation can again be either partial or complete. Partial cases have a congenital bar (anterior, posterior, lateral, or mixed) whereas completed cases have a block vertebra. The location of the bar will determine the type of deformity that develops; kyphosis, lordosis, scoliosis, or kyphoscoliosis. Block vertebra tend to cause the spine to shorten but have limited progression in the coronal plane.

Many cases have complex vertebral anomalies with both failures of segmentation and formation across several levels giving a mixed deformity (Type III)

A unilateral unsegmented bar with a contralateral hemivertebrae is the most progressive with around 6-degree progression per year.

These anomalies cause an unbalanced spine. The number of unbalanced healthy growth plates determines the degree of deformity and rate at which the deformity will progress (**Table 7.1**). Other risk factors for progression of the deformity include the age of the patient (they progress more through periods of rapid growth, i.e., first 2 years of life and adolescent growth spurt) and the location of the anomalies—lumbosacral hemivertebra causes a far greater imbalance than that of a mid-thoracic.

Assessment

History and Physical Examination

Patients with suspected congenital scoliosis should be referred early to a specialist in pediatric deformity for assessment and management of this condition. These patients are often referred to usAQ1 by a pediatrician who may be responsible for their care due to an associated issue.

A detailed history including birth history and achievement of developmental milestones should be noted during the standard history taking.

Clinical examination should be performed at each visit beginning with an assessment of the patient's height (sitting or standing dependent on ability) and weight. This should then progress to assessing sagittal and coronal balance clinically. Shoulder and waist asymmetry should be documented as well as any listing of the trunk from the center of pelvis. Gait should be checked if possible and any leg length discrepancy quantified.

Neurological status should be documented but may have to be adjusted depending on patient age and ability to cooperate with examination. Reflexes and any evidence of muscle atrophy should be noted.

Note should also be made at each assessment of the flexibility of the deformity—this can be done by a simple stretch (lifting the child under their arms and viewing how the deformity corrects).

Other anomalies should be examined for such as neck motion, short neck, and low hairline (congenital anomalies are also found with Klippel-Feil syndrome) as well as other deformities in the extremities (in particular, radial malformation and foot deformities such as pes cavus). Cutaneous markings or dimpling of the back should also be noted (signs of diastematomyelia).

Imaging

Imaging should make up part of the initial assessment of a congenital scoliosis and a plain radiograph is the first step. Ideally initial radiographs should be taken before

Table 7.1 Risk of progression by deformity

Risk of progression (highest to lowest)	Curve progression
Unilateral unsegmented bar with contralateral hemivertebra	Rapid and constant
Unilateral unsegmented bar	Rapid
Fully segmented hemivertebra	Steady
Partially segmented vertebra	Slow
Incarcerated vertebra	May progress slowly
Unsegmented vertebra/block vertebra	Little progression

the age of 4 years as after this stage they are too ossified and classification is more challenging. This allows the type of anomaly to be categorized. Whole spine posteroanterior (PA) and lateral films should be taken to check the type and location of the abnormality as well as Cobb angles should be measured to determine the size of the curve.[8] These films also allow the surgeon to assess pedicle size and presence as well as any rib abnormalities such as fusion or absence. Stretch films or suspended should be performed to assess for flexibility of the curve.

Subsequent radiographs should be compared with the original and one should not rely solely on Cobb angle measurements as these are known to have a higher rate of measurement error due to the irregular spinal landmarks.[9] Studies have shown inter- and intraobserver variance of anywhere between 2 and 10 degrees.[10] Awareness of the development of a compensatory curve as well as the assessment of the original curve can confirm the progression. Compensatory curves are made up of normal vertebrae and can be more reproducibly measured. If the compensatory curve does not progress, it is unlikely that the congenital curve will progress significantly.

Previously magnetic resonance imaging (MRI) was only considered in those patients who had neurological symptoms or those likely to proceed to surgery. Now, it is highly unusual to not perform MRI scanning of the whole spine in these children. A 30% associated with intraspinal anomaly is too high to ignore and so this should also be part of the initial investigation of these patients.[11] It is worth noting that in young children this is likely to require a general anesthetic and if possible could be combined with another procedure to avoid multiple anesthetics.

Computerized tomography (CT) with three-dimensional reconstructions allows the surgeon a better understanding of the structural deformity. These deformities can be difficult to interpret on plain radiographs due to overlying structures or the severity of the deformity. These reconstructions can now also allow three-dimensional printing of models of the spine which can be very useful in preoperative planning stage of treatment or, in certain centers, can even allow for patient-specific implants/guides to be created for theater.

Other investigations should include a renal ultrasound and urinalysis to look for renal anomalies—although these are often performed prior to visiting a spinal specialist. A cardiac assessment should also be requested in the form of an echocardiogram. Onward referral to the appropriate specialists should occur based on findings of these investigations.

Pulmonary function tests should be performed in patients who are being considered for surgical intervention as a thoracic height less than 22 cm will have a percent predicted FEV1 (forced expiratory volume in one second) and FVC (forced vital capacity) of <50%.[12]

Pearls

- Investigations should be completed before embarking on a treatment path.
- A multidisciplinary approach is required.
- MRI scan MUST be performed in all cases of congenital deformity.

Treatment

The treatment of congenital scoliosis is different from idiopathic scoliosis and should be adapted on a patient-by-patient basis. Treatment should be based on a thorough understanding of the deformity and the risk of progression. Surgery is often indicated early to prevent significant progression and the development of permanent large deformities at skeletal maturity.

Nonoperative

This can be in the form of observation alone or bracing.

All scoliosis patients require close clinical observation during periods of rapid growth. These regular assessments allow for the evaluation of curve progression.

Bracing has a limited role in the congenitally abnormal spine as it does not prevent progression of the structural curve but may be useful in controlling the compensatory flexible curve. In specific cases of a long flexible curve, progression can be slowed by bracing.[13] The other problem with bracing is the considerable period of time between onset of deformity and skeletal maturity and therefore bracing is merely a temporary measure. Compliance remains an issue in this group as it does in the idiopathic scoliosis group.

The use of rigid bracing to correct may actually be detrimental in some cases as it can exacerbate chest wall deformities.

That being said, the main stay of nonoperative treatment is clinical monitoring of a static curve.

Operative

The decision to operate or not is usually not debated. The type of surgery and timing is more difficult to determine. In adolescent idiopathic scoliosis it is usual to aim to delay surgery to close to skeletal maturity, allowing improved patient height. This is not the case in congenital scoliosis patients; surgery is often performed early to prevent the decompensation associated with structural curves and to allow fusion of the fewest levels.

The loss of height by early operative intervention is irrelevant in these cases as the growth that occurs is abnormal—increased rotation and development of compensatory curves.

That being said, the aim of achieving as much thoracic height as is safely possible prior to surgery is important because when scoliosis causes chest wall deformity it can cause cardiopulmonary failure. Decrease in thoracic height and width is associated with a reduction in exercise tolerance and poor or deteriorating pulmonary function.

As with all surgeries, complications and risks should be fully discussed with the patient (if applicable) and parents/guardian.[4]

Surgery for congenital scoliosis carries all the usual risks but neurological injury is known to be higher than for other scoliosis types and as such intraoperative neurophysiology (both somatosensory- and motor-evoked potentials) should be used.

Nutritional status should be fully optimized prior to embarking on any surgical intervention as soft tissue healing is paramount and this can be compromised by undernutrition. Superior mesenteric artery syndrome is associated with any spinal deformity surgery.[14] A multidisciplinary approach involving gastroenterology and pediatric surgeons is often required in its management, so it is useful to have these teams involved prior to embarking on any surgery.

The aim of operative treatment in all scoliosis surgery is to achieve a balanced spine.[15] In congenital scoliosis one side of the spine is growing more quickly that the other and balance can be achieved either with or without reduction of the deformity by altering growth.[16]

Multiple ways to achieve this have been described but we will focus on the most common ones:

- Posterior spinal fusion (with or without instrumentation).
- Combined anterior and posterior spinal fusion.
- Hemiepiphysiodesis.
- Hemivertebra excision.
- Guided growth systems.

Posterior Spinal Fusion

The decision of whether to fuse the spine is based on curve size and risk of progression. In the small curve that is anticipated to worsen (i.e., a unilateral unsegmented bar) the decision to proceed with an uninstrumented

in situ fusion may be appropriate whereas larger curves which require some correction will need instrumentation. It is worth noting that the rigid congenital deformity will not correct with a posterior-only fusion but the surrounding flexible levels will allow some improvement in the overall curve.

Posterior spinal fusion is the simplest and safest way to halt growth. The approach is a standard posterior midline approach although care must be taken to avoid neurological injury as there may be unrecognized laminar defects. The introduction of CT scanning preoperatively has reduced this risk. Intraoperative imaging should be available from the start of the case to ensure the anomaly level can de targeted. Levels should be confirmed both prior to incision and prior to fusion to ensure all levels of the involved AQ2 in the congenital curve can be included in the fusion. The fusion should extend laterally as far as the transverse processes.

Postoperative bracing is required to achieve fusion and should be in place for 4 to 6 months.

As the anterior spine is untouched, anterior spinal growth remains active and therefore there is an increased risk of rotational deformity with bending of the fusion mass, known as crankshaft phenomenon. The risk is increased in the younger patient and in larger curves at the time of correction. Studies quote 15% crankshaft incidence in patients undergoing posterior spinal fusion before the age of 10 years with curves greater than 50 degrees.[17]

The use of instrumentation is now widely accepted and the development of smaller implants aimed at the pediatric population has reduced the risk of implant prominence and subsequently reduced reoperation rates.[18] As with all instrumentation the risk of neurological injury remains, although current intraoperative spinal cord monitoring and careful preoperative planning (anticipating difficult anatomy with 3D imaging) has again reduced this risk and makes instrumentation more feasible in the young population.

Combined Anterior and Posterior Spinal Fusion

The advantage of performing an anterior surgical procedure is that it allows a full discectomy and removal of the end plates resulting in a better deformity correction.

Bone graft is used to aid the fusion and this can be allograft, autograft, or synthetic.

The combined anterior and posterior spinal fusion versus posterior-only fusion reduces the risk of pseudoarthrosis.[19] Anterior approach obviously carries its own risks and in patients with multiple anomalies it is important to be aware of the potential vascular anomalies to the spinal cord which could result in cord ischemia after vessel ligation during the approach.

In areas such as the thoracolumbar junction the anterior spine may be approached laterally or from the back as retroperitoneal dissection can allow enough visualization of the spine.

More modern techniques such as endoscopy allow the anterior spine to be prepared through a minimally invasive approach.

Hemiepiphysiodesis

This is a concept taken from other forms of pediatric orthopaedic surgery, i.e., long bones. This involves slowing the growth on the convex side of the curve to allow the deformity to correct as the concave side grows.[20] This results in gradual correction of the deformity.

The same prerequisites as that of long bone surgery apply here—there must be enough remaining growth to allow for correction (i.e., a young patient—usually under 6 y of age), specific to the spine is that it should be a short curve (less than 7 levels), no pathologic kyphosis or lordosis, curve < 70 degrees and there must be enough growth potential on the concave side (i.e., functioning growth plates).

This technique obviously requires a combined anterior and posterior approach

and again requires a significant period of immobilization with bracing postoperatively.

Anteriorly the convex discectomy, end plate preparation, and bone grafting are performed while posteriorly a unilateral facetectomy and fusion with bone graft are performed at each level. This technique can provide up to 20 degrees of correction by skeletal maturity. Convex compression with instrumentation posteriorly can improve immediate correction.

Hemivertebra Excision

This procedure traditionally involved a combined anterior and posterior surgical exposure, followed by excision of the hemivertebra and anterior and posterior fusion. Anterior structural bone graft is utilized on the concave side to maintain correction. If the patient is old enough (>5 y) and has a large curve, instrumentation should be used to improve the rate of fusion. It has been shown that patients undergoing posterior hemivertebrae excision before the age of 6 years (with pedicle screw fixation) have better deformity correction without compromising growth when compared with their older peers (6–10 y).[21]

The use of instrumentation allows a brace to be used in the postoperative period rather than a nonremovable cast.

More commonly now this is being performed through a single posterior approach.[22]

This procedure is usually reserved for patients with an unacceptable deformity such as a fixed lateral translation of the trunk. This procedure is safest in the lumbar and lumbosacral regions.

Guided Growth Procedures

Concern regarding thoracic insufficiency syndrome was the driving factor in the development of growth-preserving surgical techniques. The use of guided growth rods and VEPTR (vertical expandable prosthetic titanium rib) devices aims to allow continued spinal growth and relive constriction on the concave hemithorax in patients with congenital scoliosis.

Allowing patients to achieve maximum spinal height before definitive fusion optimizes thoracic volume and therefore pulmonary function.[23–25]

VEPTR has a role in patients who not only require guided growth but also have associated rib fusions. The main aim of VEPTR is to improve thoracic volume and maintain correction until skeletal maturity. It can be used to expand the concave hemithorax at 6 monthly intervals. VEPTR can use hooks as either a rib-to-rib or a rib-to-spine construct. When VEPTR is used before the age of 2 years it has been shown that pulmonary function at 5-year follow-up is significantly better than compared with children who undergo surgery later.[26]

These procedures are not the holy grail of scoliosis surgery and are associated with multiple complications; therefore, patients and their families need to be appropriately counselled before embarking on this route of treatment.

Traditional growing rods are inserted through a posterior approach and require lengthening at 6 monthly intervals under general anesthetic.[27] There has been debate on the use of single- and dual-rod constructs; studies have shown that dual-rod constructs achieve greater overall growth and curve correction.

The most recent addition to the guided growth armamentarium is the magnetic growing rods. These are based on the same principle of the traditional growth control rods but allow the rods to be lengthened in the outpatient setting. This obviously has significant impact on the children with less hospital stays, reduced anesthetic time, less time off school, etc.

The MAGEC system has been shown to be more cost effective than conventional growth rods. Estimated cost saving after 6 years is approx. £12,000. The MAGEC system (NuVasive, introduced in 2009) is recognized in the NICE (National Institute for Clinical Excellence) guidelines for scoliosis as an

effective surgical option in children who require surgery for scoliosis.[28]

MAGEC rods have been controversial in recent years with reports of metallosis and concerns regarding metal ions, but the British Spine Society, the British Orthopaedic Association, and the UK Spine Societies Board are of the opinion that although there are problems with the MAGEC rods they remain the optimal choice in many children with severe, progressive spinal deformity.

Guided growth procedures require conversion to definitive fusion either when adequate correction has been achieved or the patient is old enough for this—usually when they have reached double-digit age.

Pearls

- These are difficult cases to treat with many options available to the surgeon; this is a condition that is very different to adolescent idiopathic scoliosis.
- Cases need to be taken on an individual basis.
- Aim to prevent deformity and preserve respiratory function.
- Nonoperative treatment is of limited value in a progressive curve.

Conclusion

Patients with congenital scoliosis are complex who require a multidisciplinary management approach with significant surgical planning.

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Author queries

AQ1. Please check whether this can be changed to “deformity specialists”

AQ2. there should be some term after “involved” Please check.