Adolescent idiopathic scoliosis: a comprehensive approach to aetiology, diagnostic assessment and treatment

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Abstract

Adolescent idiopathic scoliosis is the most common type of spinal deformity seen in the paediatric population. The deformity affects otherwise normal functioning teenagers with no underlying pathological condition, no neurological abnormalities and normal imaging of the neuraxis. It produces a cosmetic deformity occasionally associated with muscular back pain and respiratory compromise in severe degrees of deformity. The aetiology is likely multifactorial and the risk of curve progression depends on the age of the patient at initial presentation, amount of remaining spinal growth and initial size of the curvature. Patients should be assessed with a thorough clinical evaluation including radiographic and often magnetic resonance imaging. Observation is recommended in growing patients with small curves up to 20–25°. Bracing can stop or slow down curve progression in growing patients with curves up to 40°. Surgical treatment is indicated in the presence of a severe deformity which is producing pain or pulmonary symptoms and is associated with cosmetic dissatisfaction. Most commonly this is performed through a posterior spinal fusion with the use of instrumentation and bone graft. The aim of the surgery is to stabilize the spine, correct the deformity and prevent deterioration. This has to be performed safely minimizing the risks of neurological/vascular/visceral injury, infection or non-union with instrumentation failure.

Keywords adolescent idiopathic scoliosis; aetiology; assessment; investigations; treatment

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Introduction

Scoliosis has been defined by the Scoliosis Research Society as a lateral curvature of the spine greater than 10° in the coronal plane. It is a three-dimensional deformity. In addition to the coronal lateral curvature, there is associated segmental vertebral rotation maximal at the apex of the curve. Vertebral rotation in the thoracic spine results in a thoracic rib hump and in the lumbar/thoracolumbar spine in a loin hump and waist asymmetry. Furthermore, adolescent idiopathic scoliosis (AIS) is usually associated with reduction in thoracic kyphosis and therefore is a lordo-scoliotic deformity rather than the commonly thought of kyphoscoliosis.

AIS is the most common type of scoliosis affecting around 0.5–3% of adolescents in varying degrees of severity. It presents at 10–18 years of age and is 8–10 times more common in females than males.[1] The most common type of curve is a right thoracic curve with associated right-sided rib hump. Left-sided thoracic curves in males require more focused investigations to exclude a non-idiopathic cause.

Aetiology

The term 'idiopathic' to some degree is a misnomer as it indicates that the aetiology and pathophysiology of AIS is unknown. However, although AIS does not seem to be associated with any particular condition and generally presents in otherwise healthy adolescents, over the years the literature has shown AIS to be a complex, multifactorial condition. Genetic, hormonal and mechanical factors are mostly implicated.[2,3]

Genetic factors in AIS have been strongly implicated since twin studies as early as 1875 have shown high concordance rates amongst monozygotic twins with AIS.[3,4] In 1997, Kesling et al.[5] published their meta-analysis, which showed concordance rates of 73% amongst monozygotic twins and 36% amongst dizygotic twins. Other studies have demonstrated high prevalence rates of AIS amongst families with AIS subjects with the highest frequency amongst first-degree relatives.[6,7] Most authors feel this is indicative of a dominant or polygenic type inheritance.

More recent studies looked into specific loci and genes that may be associated with AIS. Due to the high prevalence in females the X chromosome has been implicated.[8–10] Other chromosomes have also been linked to AIS particularly chromosomes 6, 10, and 18. In 2011, Takahashi et al.[11] looked at over 1000 Japanese AIS patients; they found an association between AIS and the LBX1 gene on Ch10. This gene is expressed in the dorsal spinal cord and skeletal muscle and is thought to play a role in regulation of neural activity. Studies in Caucasian population have shown similar results.

More recently mutations in the POC-5 gene have also been implicated with high prevalence amongst families with AIS.[12] This gene is strongly expressed in the brain and is important for centriole development (and in turn affects cell division).

Humans are the only mammals affected by scoliosis; hence, mechanical factors have also been implicated. It is thought that bipedalism with the associated upright posture and higher centre of gravity (above the pelvis) may result in rotationally more unstable spinal segments, which in addition to other implicated factors such as osteopenia, growth imbalance and subclinical neuromuscular abnormality may result in scoliosis.[13–16]

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Biochemicals such melatonin and growth hormone have also been studied. Even though results in animal studies have shown association between hormones and spine deformity there is no strong evidence in humans.[17–19]

Such research into the cause of the condition will be hugely important in the future. It will help us as clinicians assess which patients are likely to get the condition and how severe the condition is likely to be. It will also help us determine those patients in whom the deformity is predicted to progress. This will have a huge impact on how we follow up patients and advise them with regards to treatment options.

Assessment

Patients usually present having noticed a change in the shape of their back. It is important to take a history that helps ascertain the main symptoms and impact of deformity, the likelihood of scoliosis progression and any potential non-idiopathic causes that can led to the development of the curvature.

History

- Presenting complaint: usually change in the appearance of the back. Ascertain when first noticed and degree of progression since.
- History of presenting complaint and associated symptoms:
  - Pain is not usually a prominent complaint. However, patients may report intermittent mechanical/muscular back pain which overall does not seem to affect significantly their level of activities. Presence of severe pain should direct investigations towards a non-idiopathic aetiology. Other causes of pain in the context of AIS can be due to late presenting severe deformity causing muscle imbalance and costo-pelvic impingement. Lower back pain can be due to spondylolysis or degenerative disc disease.
  - Neurological deficit: pins and needles, weakness, tingling, headaches (think of intraspinal anomalies such as Arnold–Chiari malformation, syringomyelia, diastematomyelia, tethered cord).
  - Other symptoms: poor exercise tolerance in severe scoliosis due to restrictive lung disease.
- Obtain past medical history predominantly to exclude a non-idiopathic cause (including birth history): spina bifida, Arnold–Chiari malformation (or other spinal dysraphism), neuromuscular disorders and leg length discrepancy. Exclude other co-morbidities which will be essential if surgical treatment is anticipated.
- Assess skeletal maturity: chronological age, patient height, maternal and paternal height, time of onset of menarche and thelarche.
- Family history of AIS and social history to assess child future aspirations, impact of scoliosis progression and treatment options.

Examination

- Gait: check for short leg gait. Leg length discrepancy can cause a curvature of the spine in an attempt to compensate. The curvature of the spine can be corrected by sitting the patient on the couch or by using a block to raise the shorter leg. This will level the pelvis and allow the compensatory spinal deformity to correct.
- Standing position (appropriate exposure of the back):
  - Coronal balance to assess truncal shift (Figure 1).
  - Sagittal balance (usually a hypokyphosis of thoracic spine and hyperlordosis of lumbar spine).
  - Rib hump prominence (usually a right-sided scoliosis with right-sided rib hump) (Figure 2). Left-sided scoliosis is less likely and requires investigations to exclude a non-idiopathic cause.
  - Shoulder height difference. With a right-sided curve the right shoulder is usually raised. This indicates that any proximal left upper thoracic curve is non-structural. If the left shoulder is raised that usually indicates a structural proximal thoracic curve. This has impact on the proximal level of instrumentation required during correction surgery.
  - Waist asymmetry, loin hump and pelvic tilt (Figure 3).
  - Features suggestive of spinal dysraphism (hairy patches, dimples, nevi, haemangiomas overlying the spine, cavovarus feet)
  - Exclude marfanoid features (slim/slender extremities, joint hyperlaxity) and features of neurofibromatosis (café au lait spots, axillary freckling).
  - Adams forward bending test to evaluate convex rib prominence and assessment of scoliosis flexibility. By asking the patient to keep their knees straight and bend forwards to touch their toes, the rib prominence becomes more visible. You can use a scoliometer to quantify the amount of rotational deformity. You can then stand on the side of the curvature (usually right) and use your body as a fulcrum to correct the deformity. This usually results in some correction of the scoliosis and flattening of the rib hump giving an indication of scoliosis flexibility.
  - Complete neurological examination including abdominal reflexes to exclude a central cause.

Investigations

- Initial imaging to assess degree of scoliosis: standing coronal and lateral whole spine radiographs.
- Exclusion of non-idiopathic cause:
  - MRI scan of the whole spine including the craniocervical junction to exclude spinal dysraphism (tethered cord, diastematomyelia, spina bifida, syringomyelia, Arnold–Chiari malformation).
  - Brain MRI if high suspension of a neuromuscular cause.
  - CT scan: on occasions to exclude lumbosacral abnormalities (spondylolysis, Bertolotti syndrome) and congenital scoliosis.
  - Genetic testing: if indicated to exclude genetic conditions such as neurofibromatosis, Ehlers–Danlos and Marfan's syndrome.
- Radiographic assessment of skeletal maturity (help assess risk of curve progression):
  - Risser sign and tri-radiate cartilage on postero-anterior (PA) whole spine radiograph.
  - Anteroposterior (AP) pelvis: greater trochanter physeal closure.

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