REVIEW

Congenital scoliosis in a neonate: can a neonatologist ignore it?

S Jog, S Patole, J Whitehall

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The frequency of diagnosis of congenital scoliosis in the neonatal period is expected to rise given the increasing survival of high risk neonates in the surfactant era and their frequent exposure to x rays. Considering its significant long term implications a neonatologist cannot afford to ignore the diagnosis of congenital scoliosis in a neonate as close surveillance, early detection, and treatment may prevent/minimise the wide spectrum of potentially serious deformities that can affect the developing spine. The review provides general guidelines to help the neonatologists in counselling the parents and in planning the multidisciplinary follow up for management of congenital scoliosis.

was also conducted. The following brief review based on this literature search is aimed towards providing general guidelines to help neonatologists in counselling the parents and in planning follow up, which is crucial for management of congenital scoliosis using a multidisciplinary approach. It emphasises the role of a neonatologist in close surveillance, early detection, and management of this congenital anomaly that has a significant impact on the quality of life of patients for years after their discharge from the nursery.

INCIDENCE

The true incidence of congenital scoliosis in the general population remains unknown as minor deformities often remain undetected. Prenatal detection of an abnormal vertebral body, however, can alert the paediatrician to watch for congenital scoliosis.⁴ About 10% of cases of structural scoliosis are due to congenital intervertebral or vertebral body abnormalities that cause imbalance in longitudinal growth of the trunk.³ Congenital scoliosis is more common in girls than in boys, occurring in the ratio of 2.5:1. Curves occur to left and the right with equal frequency. The incidence of the curve at different levels of

Congenital scoliosis is a lateral curve of the spine due to an imbalance in the longitudinal growth of the spine caused by vertebral anomalies. The deformities caused may be mild, causing subclinical curvatures, or severe, associated with neurological complications, pulmonary restriction, cor pulmonale, and premature death.¹ The age at diagnosis is reported to vary from the first few days of life to 25 years (average 6 years).² Given the increasing survival of high risk neonates in the surfactant era the frequency of diagnosis of congenital scoliosis in the neonatal period (fig 1) is expected to rise in view of their frequent exposure to radiological imaging—a modality that has influenced the care of scoliosis for over 100 years.³

As practising neonatologists we recently faced a neonate of 26 weeks' gestation with respiratory distress where congenital scoliosis and a hemivertebra was diagnosed soon after birth on a routine chest radiograph. Repeated queries by the father—a devoted rugby player—about the baby's future ability to play contact sports, need for surgery, and risk of cosmetic deformity ultimately led to an orthopaedic consultation where the need for further follow up was emphasised. Attempting to provide specific answers to the parent's queries we soon realised that the issue of congenital scoliosis in the neonatal period was in "no man's land". A literature search was therefore undertaken to improve the quality of parental counselling while planning the multidisciplinary follow up as a neonatologist. Literature relevant to congenital scoliosis in the neonatal period was searched in Medline, EMBASE, CINAHL, the Cochrane Library, reference lists of review articles, and abstracts published in *Pediatric Research*. A hand search of paediatric and perinatal journals

Figure 1 Chest radiograph of a male neonate at 26 weeks' gestation showing lower thoracic scoliosis with concavity to the right, a single hemivertebra at T9, and 11 ribs on the right side.

See end of article for authors' affiliations

Correspondence to:
Dr Sanjay Patole, C/O A S
Pabla, PO Box 333,
Joondalup, Western
Australia 6027;
skpatole@hotmail.com

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the spine is as follows—upper thoracic: 33%, lower thoracic: 31%, thoracolumbar: 20%, lumbar: 11%, and lumbosacral: 5%.⁵

FAMILY HISTORY

Parental consanguinity and a positive family history (including twin girls and two brothers) of congenital scoliosis have been reported in 40% and 8% of patients respectively.² Discordance for congenital scoliosis has also been reported in monozygotic twins, suggesting that it may be acquired in utero rather than genetically determined.⁶

AETIOLOGY

Failure of formation, segmentation, or a mixture of both, has been proposed as the developmental aetiology of congenital scoliosis. Vertebral anomalies develop during the first six weeks of intrauterine life, when the anatomical pattern of the spine is formed in the mesenchyme.⁷ Hemivertebra is the most common anomaly that causes congenital scoliosis by acting as an enlarging wedge on the affected side of the spine.^{7,8} The degree of scoliosis produced by hemivertebra depends on their type/site/number/relationship to each other and patient's age.⁹

DIAGNOSIS

The degree of spinal curvature is measured by traditional Cobb angle determination.¹⁰ Recently Cobb angle determination has been aided by either computer software programs or a mechanical differential goniometer.^{11,12}

HISTOPATHOLOGY

A major problem in congenital scoliosis has been reported to relate to the vertebral bodies, which are irregular in size, shape, and position, and to the intervertebral discs, which are always abnormal.¹³ An earlier study reported that the vertebral bone and cartilage as tissues were histologically unremarkable, and that it was their positioning which was irregular. Additionally no molecular abnormality of collagen was reported in the bone, cartilage, or skin.¹³ A subsequent study reported a "normal" percentage of type 1 fibres in the thoracic erector spinae muscles on the concave side but a significantly greater percentage of such fibres on the convex side in patients with congenital scoliosis or scoliosis with early onset.¹⁴ A relatively recent study has reported reduced proteoglycan and water contents in the cartilage end plate and intervertebral disc, particularly towards the concavity of the curve in congenital scoliosis.¹⁵ It was suggested that though secondary to the altered loading in the scoliosis, calcification of the cartilage and end plate may be highly significant in the progression of the scoliotic curve.¹⁵

ASSOCIATED ANOMALIES

The incidence of spinal cord abnormalities associated with congenital scoliosis is reported to range from 20% to over 50% in most of the series.^{2,3,16} Three dimensional computed tomography and magnetic resonance imaging are useful in detecting these abnormalities including tethered cord, syringohydromyelia, Chiari malformation, spinal cord tumour, and the most common intraspinal anomaly, diastematomyelia.^{16,17} Among children with congenital spinal anomalies 30%–60% are reported to have anomalies at sites other than the spine. The most common of such sites are genitourinary tract, cardiac system, the spinal cord, and the cervical spine. Many are part of VATER syndrome and also incidence of Klippel-Feil syndrome is high. A review of 28 patients with VATER association reported death of three children during the first two years of life. Of the remaining 25 patients, 12 had required orthopaedic procedures for the treatment of congenital scoliosis or deformity of the upper or lower extremities.¹⁸ Congenital scoliosis has been reported to be associated in 19%–70% of

patients with Klippel-Feil syndrome.^{19–25} Duchenne muscular dystrophy best characterises myopathic type of neuromuscular scoliosis whereas cerebral palsy represents the most common form of neuropathic neuromuscular scoliosis. Approximately 90% of boys with Duchenne muscular dystrophy are reported to develop severe scoliosis by adolescence, not amenable to control by non-surgical means.²⁶ In a retrospective review of 45 patients of Duchenne muscular dystrophy no scoliosis was observed in ambulatory patients. However 96% of the wheelchair bound patients suffered from scoliosis with 96% having hip flexion or abduction contractures.²⁷ In a prospective review of 92 individuals with myotonic dystrophy 47% of the 17 patients with congenital myotonic dystrophy were noted to have relatively mild, non-progressive scoliosis. Seventy five per cent of these patients also had impaired intellectual and cognitive function, frequently severe.²⁸ Severe and rapidly progressive congenital scoliosis with multiple joint contractures has also been reported in myopathies like non-progressive central core disease and lipid storage myopathy.^{29,30} Recently it has also been reported in association with dysmorphic features and congenital anomalies as in Genoa's syndrome, Mayer-Rokitansky-Küster-Hauser syndrome, and in multiple enchondromatosis with features of dyssspondyloenchondromatosis and Maffucci's syndrome.^{31–33}

NATURAL HISTORY AND PROGRESSION

Until 1960 it was thought that congenital scoliosis was non-progressive and thus did not require treatment. McMaster and Ohtsuka and Winter *et al* refused the theory about the benign nature of congenital scoliosis.^{7,34} They reviewed a combined total of 485 patients, most until skeletal maturity and found out that almost 75% of patients required treatment and 84% of patients who were untreated developed curves greater than 40 degree by maturity. Progression and prognosis were related to type of anomaly and anatomical site of the curve. Children with clinical deformities in the first year of life had the worst prognosis and had early progression. If the curve was present before the patient was 10 years old, it usually increased, in particular during the adolescent growth spurt.⁷ All curves present during infancy and associated with an unsegmented bar deteriorated very rapidly with advancing age as the bar became more ossified.⁷ Similarly other researchers have reported hemivertebra as the most common type and unilateral unsegmented bar with contralateral hemivertebra as the most severe and most progressive pattern of deformity.² Increasingly severe scoliosis may also develop when an asymmetric block vertebra, a wedge vertebra or one or more lateral hemivertebra is present.³ Thoracolumbar curves tend to have worst prognosis and the greatest progression followed by lower thoracic curves and upper thoracic curves.

PULMONARY FUNCTION

Development of restricted pulmonary function is a concern. Curve magnitude directly affects pulmonary function, but the development of restrictive lung function occurs only as the curve approaches 90 degrees. The vital capacity of patients with congenital scoliosis has been compared with those with idiopathic scoliosis.³⁵ For any given Cobb angle the loss in vital capacity was approximately 15% greater in congenital compared with idiopathic scoliosis. This greater impairment in lung function in congenital scoliosis has been proposed to be due to the associated rib deformity or to an underlying lung anomaly.³⁶ Vital capacity screening has been recommended for patients with severe curves. A full spirometry work-up is recommended if surgery is planned for those with a vital capacity <60% of normal.³⁶

COSMETIC DEFORMITY

Cosmetic deformity occurs with most curves.⁷ With upper thoracic curves, elevation of the shoulder on the convexity of the curve, with tilting of the head into the concavity, may be seen. Curves >30 degrees may produce an unacceptable deformity, especially in girls. Structural congenital curves tend to have only a mild rotational component, producing only a mild hump. Unbalanced curves in the lower thoracic and lower lumbar region produce a pelvic obliquity with apparent shortening of the leg on the concave side of the curve. Difficulties in ambulation and balance may occur due to the tilting of the trunk away from the apex of the curve.⁷

NON-SURGICAL TREATMENT

Bracing can treat <10% of cases of congenital scoliosis.³⁷ However the recommended duration and effects of wearing braces remain controversial. A brace may not arrest or correct the development of curve, but may slow progression and maintain flexibility, allowing surgery at a later stage.³⁷

SURGICAL TREATMENT

Congenital scoliosis is often rigid and correction can be difficult. The goal of surgery is to arrest progression of the curve and correct the deformity to the extent possible. Ideally, surgery is indicated early in curves with rapid progression before they become severe and fixed.³⁷ Of the various options available (for example, combined anterior and posterior fusion, resection of hemivertebra) posterior spinal fusion is the oldest and safest spinal procedure and continues to be the gold standard.³⁷ Anterior and posterior hemiarthrodesis and hemiepiphysodesis are reported to be most effective in cases of hemivertebra.³⁷ Anterior and posterior arthrodesis (preferably in the first year of life) is also recommended by some authors as a prophylaxis in all midthoracic, thoracolumbar, and lumbar curves as surgical correction after 5 years of age was not successful in their patients.³⁸ Semisegmented and incarcerated hemivertebra usually do not require treatment. Fully segmented non-incarcerated hemivertebra may require prophylactic treatment to prevent significant deformity.⁹

It is important to note that the current recommendations for timing and types of options (surgical or non-surgical) for management of congenital scoliosis are not based on randomised controlled trials.

ROLE OF A NEONATOLOGIST

(1) Apart from recording the demographics a detailed birth, family, and social history should be obtained followed by a thorough clinical, especially neurological, examination.

(2) The nature of the problem and the need for investigations like echocardiography, abdominal/renal/cranial ultrasonographic studies, computed tomography, and magnetic resonance imaging at appropriate times needs to be discussed with the parents as associated malformations can have a serious impact on the outcome (for example, tethered spinal cord).

(3) It is important to emphasise technical difficulties. For example vertebral anomalies may not be recognised radiologically until the neonate is 3–4 years old due to lack of ossification. Similarly sharing the information that hemivertebrae (if diagnosed) could be seen radiologically before the age of 2 years, but as the deformity progresses they could get obscured due to progression of the curve will help the parents to appreciate the need for follow up.

(4) Implications of the site and severity of the spinal curve and the inconstant rate of deterioration of the curve with tendency to deteriorate during time of growth spurts need to be emphasised. This will also be a pointer to the fact that a prolonged follow up is needed. The possibility of need for surgical correction at an early age has to be mentioned in the presence

of vertebral anomalies like unilateral unsegmented bar with contralateral hemivertebra.

(5) Given the complexity and diversity of the information to be conveyed it is necessary that multiple meetings with parents and supporting family members are required. Being involved in the day-to-day care of their baby a neonatologist is in a unique position to convey such complex information to parents.

(6) Having counselled the parents about the nature, consequences of the problem, and the available therapeutic options it is then important to arrange a meeting between the parents and the orthopaedic surgeon before discharge of the neonate from the nursery if not earlier. The aim of this meeting is to establish contact between the family and the orthopaedic surgeon, the key person in the multidisciplinary management of the problem.

(7) Available evidence indicates that follow up examinations (measurement of spinal curve, pulmonary function, neurological status) are necessary every 12 months until the age of 10 years and then every 6–12 months until the age of 15 years as rapid deterioration of the spinal curve may occur with growth spurts.

CONCLUSION

Given its significant long term implications a neonatologist cannot afford to ignore the diagnosis of congenital scoliosis in a neonate as close surveillance, early detection, and treatment may prevent/minimise the wide spectrum of potentially serious deformities that can affect the balance of the developing spine.³⁷ In depth parental counselling by the neonatologist is crucial for planning multidisciplinary approach in the management of this problem, which has been known since antiquity.^{3,39} The task, however, is not easy, as the parents often perceive the neonatologist as doing nothing to “fix the problem”. Additionally the specific follow up starts long after discharge of the neonate from the nursery and lasts for years when the neonatologist is not on the scene.

Authors' affiliations

S Jog, S Patole, J Whitehall, Department of Neonatology, Kirwan Hospital for Women, Townsville, Queensland, Australia

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IMAGES IN MEDICINE.....

Superior lumbar hernia of Grynfeltt

Figure 1 Abdominal computed tomogram showing superior lumbar hernia (SLH). LK, left kidney; PR, perirenal fat passing through transversalis aponeurosis to form SLH (arrowed); ES, erector spinae; LD, latissimus dorsi. (1) External oblique, (2) internal oblique, (3) transversus abdominis.

A 44 year old woman was investigated for a soft, non-tender, reducible swelling, measuring 5 cm in diameter in her left lumbar region. There was no cough impulse. An abdominal computed tomogram revealed a superior lumbar hernia, first described by Grynfeltt. The hernial content was retroperitoneal fat from around left kidney, which had prolapsed through the transversalis aponeurosis. The external oblique (1), internal oblique (2), and the transversus abdominis (3) lie anteriorly (see fig 1). The erector spinae (ES) forms the posterior boundary. The superior border is formed by the 12th rib and the iliac crest forms the inferior border. The defect was repaired using interrupted non-absorbable sutures after retracting the latissimus dorsi. The patient has been followed up for a period of 16 weeks and there has been no recurrence.

Superior lumbar hernia is rare and may easily be misdiagnosed as a lipoma if a strong index of suspicion is not kept.

**A Pitale
V Laughlin**

Lagan Valley Hospital, Hillsborough Road, Lisburn,
Co Antrim BT28 1JP, UK; ashishpitale@lycos.com