Congenital scoliosis in non-identical twins: case reports and literature review

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Congenital scoliosis due to vertebral anomalies may occur in less than 0.1% of the population. Several different theories have been put forth in the literature to account for the etiology of congenital scoliosis and the vertebral anomalies which contribute to its development. The study of scoliosis in twins has contributed to the understanding of causative factors including genetics, environment and in utero events during embryologic development. Case reports of fraternal (non-identical) juvenile male twins with congenital scoliosis associated with differing congenital vertebral anomalies are presented. Both children were asymptomatic at the time of the initial consultation and showed no signs of neurologic compromise. Rapidly progressive, severe genetic scoliosis requires prudent observation and referral to a pediatric orthopedic surgeon to determine appropriate options for care and to screen for potentially life threatening disorders. Chiropractors may be seen as

La scoliose congénitale due à des anomalies vertébrales peut se produire chez moins de 0,1 % de la population. Plusieurs théories différentes ont été avancées dans la recherche scientifique pour expliquer l'étiologie de la scoliose congénitale et les anomalies vertébrales qui contribuent à son développement. L'étude de la scoliose chez les jumeaux a contribué à la compréhension des facteurs étiologiques, dont la génétique, l'environnement, et les événements in utero au cours du développement embryonnaire. On présente des rapports de cas de frères jumeaux (non identiques) mineurs atteints de scoliose congénitale associée à différentes anomalies vertébrales congénitales. Les deux enfants étaient asymptomatiques au moment de la consultation initiale et n'ont montré aucun signe d'atteinte neurologique. Susceptible de progresser rapidement, la scoliose génétique grave nécessite une observation attentive, et le renvoi à un chirurgien orthopédiste pédiatrique pour déterminer les options appropriées des soins et pour le dépistage de maladies potentiellement mortelles. Les chiropraticiens peuvent être considérés comme des remparts contre la scoliose. Ceci étant dit,

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©JCCA 2014 gatekeepers for scoliosis and a thorough understanding of appropriate standards of care is required.

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KEY WORDS: scoliosis, twins, congenital, hemivertebra, chiropractic

Introduction

Scoliosis is a lateral curvature and twisting of the spine measuring 10 degrees or more. The Cobb method of mensuration determines the degree of scoliotic curvature by the angle created between lines drawn on endplates of the end vertebrae (superior endplate of upper end vertebra; inferior end plate of lower end vertebra). (Figure 1) This method has been adopted and standardized by the Scoliosis Research Society, which also classifies the severity of scoliosis. (Table 1) The Nash and Moe method measures vertebral rotation on a frontal radiograph using the dis-

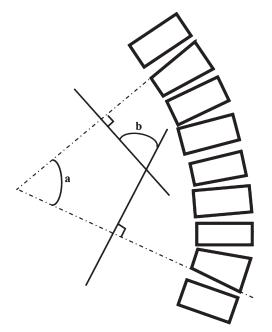


Figure 1. Cobb method of scoliosis mensuration (Reproduced with permission of the Radiological Society of North America (RSNA). Kim H, Kim HS, Moon ES et al. Scoliosis imaging: What radiologists should know. RadioGraphics. 2010;30:1823-1842)

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MOTS CLÉS : scoliose, jumeaux, congénital, hémivertèbre, chiropratique

placement of the pedicle on the vertebral body. (Figure 2) Juvenile idiopathic scoliosis is defined as a spinal curvature diagnosed between 3 years and 9 years 11 months of age, whereas congenital scoliosis is associated with bony abnormalities of the spine present at birth.¹ Juvenile onset scoliosis has been reported to account for 8% to 21% of patients with scoliosis, although these numbers are based on studies with small numbers of participants and may not be statistically accurate.^{2.3} The incidence of congenital scoliosis in the juvenile population is unknown since many spinal anomalies go undetected due to the presence

Table 1.Lippman-Cobb Classification of Scoliotic Curvature

Group	Angle of Curvature in Degrees
I	<20
II	21-30
III	31-50
IV	51-75
V	76-100
VI	101-125
VII	>125

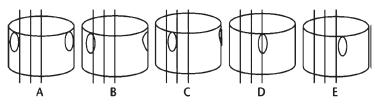


Figure 2. Nash and Moe pedicle method for determining vertebral rotation

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Figure 3A. Photograph of Twin A at 5 years 9 months





Figure 3C. Twin A Lateral standing lumbar spine radiograph showing L4 facet hypoplasia and L4 anterolisthesis

Figure 3B. Twin A-AP standing radiograph at 4 years 11 months. 10 degree right lumbar scoliosis with mild compensatory left thoracic curve associated with decreased vertical interpediculate distance between the left L4 and left L5 pedicles as compared to the corresponding pedicles on the right, and low right hemipelvis

of minimal spinal deformity. The incidence of vertebral anomalies has been estimated to be 0.05-0.1% of live births.⁴ Several theories exist as to the etiology of congenital scoliosis. The overall impression among researchers is that the cause is multifactoral. ⁵⁻⁷

These case reports detail the presentation of a pair of fraternal juvenile twins with dissimilar scoliotic curve characteristics in a private chiropractic practice. A literature review was conducted to appreciate the etiology of congenital and juvenile onset scoliosis particularly in twins, as well as to briefly outline current standards of care.

Case Report

Twin males aged 5 years 9 months presented to a private chiropractic office on referral from the family medical physician for evaluation of juvenile scoliosis. They were escorted by their birth mother who was interviewed with respect to family history, birth history and the twin's history to date. At the time of this presentation the twins were asymptomatic, apart from visible signs of truncal asymmetry and postural imbalance. Twin A was 119.5 cm tall and weighed 24.5 kg, while twin B measured 115.5 cm in height and weighed 21.8 kg. Both boys were of nor-





Figure 4A. Photograph of Twin B at 5 years 9 months

Figure 4B. Twin B AP standing radiograph at 22 months Hemivertebra at T10 with a 30 degree right thoraco-lumbar scoliosis



Figure 4C. Twin B Twin B AP standing radiograph 4 years 11 months Hemivertebra at T10 with a 35 degree right thoraco –lumbar scoliosis

mal intellectual development for age and were able to follow directions when asked. They were active throughout the interview and examination and appeared to possess equal strength, co-ordination and physical ability. During standing postural assessment, twin A demonstrated pelvic and shoulder unlevelling, lower on the right side, with left head tilt. (Figure 3A) No rib humping was observed during forward trunk flexion (Adam's test). Twin B demonstrated trunk rotation left posterior and right head tilt (Figure 4A). Slight left lower rib humping was evident on forward trunk flexion. Lateral bending and extension ranges were tested in twin A and were found to be within normal limits, while twin B exhibited mild segmental restriction in left lateral bending at the thoraco-lumbar junction. No pain was elicited during examination in either twin. Babinski sign was absent when tested, and gait and balance was normal in both twins.

The mother's family history is unremarkable for scoliosis, congenital spinal anomaly or other serious disease. She has two other older female children, neither having any evidence of scoliosis. She denies smoking, drug use, or illness prior to conception or during pregnancy. The father's family history is also negative for scoliosis or spinal anomaly, and there is no consanguinity with the twin's mother. The boys were full term at delivery and were determined to be fraternal, not identical twins. At birth, twin A was 4.2 kg and 52.5 cm in length while twin B was 3.7 kg and 54 cm in length. They were considered to be very large babies at birth for twins, and gestational diabetes was ruled out as a contributing factor to their size. The twin's siblings were also born with similarly high birth weights.

The mother was questioned about her health and lifestyle during the early stages of pregnancy with the twins. She recalled that she had been breast feeding her second child and had been menstruating for one year following a one year absence of her monthly cycle following the delivery of this child. She normally has a 6-8 week menstrual cycle and was unaware that she was pregnant until approximately 6 weeks of gestation. She had been exercising regularly and enjoyed very good health during this time. She was not aware that she was carrying twins until 5 months of gestation.

Radiographs of the twins were provided and reviewed during consultation. Twin A had a single AP standing radiograph taken at 4 years 11 months of age which demonstrated a 10 degree right lumbar scoliosis measured using the Cobb method between L2 and L5. There was also a mild compensatory left thoracic curve present. (Figure 3B) This view also demonstrated pelvic unlevelling, lower on the right which was possibly associated with a lower limb discrepancy due to a shorter right leg. A decreased vertical interpedicular distance was noted at L4-5 suggestive of a failure of segmentation. A lateral lumbar film was recommended and taken at a later time to confirm this anomaly. (Figure 3C) An independent chiropractic radiologist was consulted to review these films. The anomaly was reported as "L4 hypoplastic facet development with anterolisthesis of L4 on L5". "The findings suggest congenital etiology scoliosis". (Addendum)

Twin B had serial AP standing radiographs taken at 22 months and again at 4 years 11 months. The initial view demonstrated a hemivertebra at T-10 with incomplete development of the right side of this anomalous segment. This was associated with a 30 degree right thoraco-lumbar scoliosis measured using the Cobb method between T12 and L4 which had developed below the anomaly. (Figure 4B) The second radiograph demonstrated progression

of the thoraco-lumbar scoliosis to 35 degrees associated with the hemivertebra. (Figure 4C)

Discussion

The study of scoliosis in twins, particularly adolescent idiopathic scoliosis is well documented. ⁸⁻²¹. Grauers et al published their findings on "Heritability of Scoliosis" following a survey of 64,578 twins in the Swedish twin registry and concluded that genetic factors were responsible for 38% of scoliosis cases as compared to 62% environmental association with the development of scoliosis. This study also concluded that in monozygotic twins, concordance for idiopathic scoliosis is much higher than in same sex dizygotic twins.²² There were three studies published regarding juvenile scoliosis in twins.²³⁻²⁵

Congenital scoliosis is a lateral curvature of the spine associated with vertebral anomalies such as block vertebra, wedge vertebra, single hemivertebra, two unilateral hemivertebrae, a unilateral unsegmented bar, or a unilateral unsegmented bar with contralateral hemivertebrae at the same level. These represent the five classifications of vertebral anomaly as described by McMaster and Ohtsuka.²⁶ A single hemivertebra, which is classified as a failure of formation is a common vertebral anomaly found in congenital scoliosis and depending on the location, will contribute to scoliotic progression in the growing child. A fully segmented hemivertebra may be associated with rapid scoliotic progression and may be resistant to conservative management.²⁷⁻³¹Genetic signaling in the embryological development stage of somitogenesis, as well as temporary vascular insufficiency of the growing fetus may contribute to failure of ossification of a vertebra or vertebrae, osseous metaplasia of the annulus fibrosus or persistent notochord. These proposed theories in the formation of vertebral anomalies, such as hemivertebrae and other structural malformations suggest a role for environmental and genetic contributors.32-37

Juvenile scoliosis is unique in that progression may occur during a period of time where growth is dormant.^{2,36,37} Progression of the spinal curvature may initially be so subtle that clinical observation without serial radiographs will not demonstrate the rate of progression. Curve patterns in juvenile scoliosis tend to be similar to those in adolescent idiopathic scoliosis. Studies which follow cases of juvenile scoliosis report that progression is more likely to be aggressive in younger patients.^{2,38,39} However some patients within these studies showed gradual regression of their curves with time as they approached puberty, further confounding the theories of the natural course of scoliotic progression and complicating clinical decision making with respect to treatment.^{40,41} Unrecognized physical activity, particularly in male patients may lead to curve regression. Symmetric loading of vertebral structures during weight bearing exercise may be a contributing factor in gradual regression. Prepubescent curve regression is not a guarantee that a scoliotic curve will not progress rapidly during adolescence.^{42,43}

Congenital scoliosis due to hemivertebra is more likely to show rapid progression at a younger age than juvenile idiopathic scoliosis and a referral for a surgical opinion at an early age is necessary.^{30,31,42} Anomalies of the neurologic or visceral structures, especially of the genitourinary system, may also occur when errors of formation or segmentation of the spine exist. During the fifth week of embryonic development, the vertebral column and the genitourinary system may be subject to embryonic insult which could lead to abnormalities.44,45 This may present challenges when considering surgical intervention to minimize the progression of scoliosis. A complete evaluation of the surgical candidate with vertebral anomalies, including spinal and abdominal MRI, as well as diagnostic ultrasound and occasionally voiding cystourethrograms may be necessary to minimize surgical risk as well as to diagnose and treat potentially life threatening disorders.⁴⁶⁻⁴⁹ Neither Twin A or Twin B has yet been assessed for other developmental anomalies.

Winter and Lonstein published a retrospective case series of 1250 patients with congenital spinal deformities and found that only seven patients with scoliosis secondary to a hemivertebra showed gradual improvement without treatment. This is not a favorable prognosis for children with congenital scoliosis due to a vertebral anomaly and points to the importance of identifying and determining the classification of the anomaly at the earliest possible age. One of the children in Winter and Lonstein's study was a twin with a hemivertebra at L1 while his twin brother had no vertebral anomaly. The child with hemivertebra was followed from age 15 months to age 16 years and showed a reduction of the spinal curvature from 42 degrees to 31 degrees without intervention, and was asymptomatic at all times.⁵⁰

Non-identical twins are dichorionic, diamniotic twins

and the differences in their genetic makeup would be similar to siblings born as a result of pregnancies separated by time. However, the twins in these case studies offer a unique opportunity to observe the progression of this challenging clinical condition for the practicing chiropractor. These case studies and review of the literature suggest that while there is an understanding of the etiology of scoliosis, accurate prognosis cannot be made on a case by case basis as to the likelihood of progression or regression of scoliosis. There are many and varied contributing factors during fetal development and during childhood which affect the progression of scoliosis. It is incumbent on the practitioner monitoring patients with scoliosis to diligently follow and to carefully observe subtle changes that may foretell progression and to make clinical decisions regarding appropriate standards of care. Examinations should be conducted at 2-3 to 36-60 months intervals according to the specific clinical situation, and standing frontal full spine radiographs including the occiput and pelvis should be obtained when progression is apparent.51 A scoliometer, which is a variant of a carpenter's level, can be incorporated in the examination process to measure the severity of the rib hump and lumbar bulge. Raster stereography may also be considered to document the shape of the spine using reflected light beams without the use of ionizing radiation.⁵²

Pediatric orthopedic referral would be the most prudent course of action for twin B with congenital scoliosis. Hemivertebra resection and transpedicular instrumentation or contralateral hemiepiphysiodesis are options for surgical intervention in young children and should be performed early to prevent severe local deformities and secondary structural changes. Adequate post surgical bracing is essential to prevent failure of instrumentation, which has been reported as a frequent occurrence. ^{28,29,31,40,43,47}

Twin A shows signs of a mild congenital scoliosis associated with a subtle vertebral anomaly. Some evidence in the literature supports the use of chiropractic spinal manipulation and rehabilitative exercises for the management of scoliosis, although long term trials have not yet been conducted.⁵³⁻⁵⁶ None of the studies cited were conducted on juvenile patients. Brace management has been shown to be a recognized and beneficial form of conservative care in some cases and may be indicated if idiopathic scoliosis progresses at any time prior to skeletal maturity.^{51,57-59}

Limitations

These case studies represent one pair of non-identical twins.

Conclusion

The twins in these case studies were both born with congenital vertebral anomalies which contribute to dissimilar scoliotic characteristics. Referral and screening for potentially life threatening disorders including other CNS or genitourinary anomalies as well as potential referral for surgical intervention is important. By gaining a better understanding of the etiology and time of onset of scoliosis, improved screening methods and standards of care may be developed in the management of this enigmatic childhood disorder, particularly in twins. These cases demonstrate the importance of the role of the chiropractor in examination, monitoring, discussing risks of progression, and in making an appropriate referral to a pediatric orthopedic surgeon for follow up care in severe cases of congenital scoliosis. Educational resources should be provided to parents of twins for the purpose of monitoring scoliosis progression, especially in families where scoliosis or congenital structural anomalies are present. Funding for long term studies of conservative management of juvenile idiopathic scoliosis and congenital scoliosis including spinal manipulation should be considered to determine effectiveness.

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Addendum

Independent Chiropractic Radiologist's report for Twin A radiographs

11-28-1013

This is review of a two view series of a five year old twin for scoliosis

Anteroposterior upright thoracic, lumbar spine and pelvic and lateral lumbar spine views reveal Lovett positive dextroscoliosis of the lumbar spine, apex at the L3-L4 level measuring less than ten degree Cobb angle. The right hemipelvis is inferior to the left as seen at the iliac crest and femoral head levels. No compensatory levoscoliosis of the thoracic spine is noted. On the anteroposterior view the left sided disc space is unilaterally less than the right and the pedicle length development at this site is noted on lateral projection to be mildly decreased compared to the superior adjacent L3-L4 level. The L5 pedicle formation is less in sagittal measurement than the L4 or L3 levels. The inferior facet development is hypoplastic at the L4 level and the McNabb line drawn under the L5 inferior vertebral body shows the first sacral facet to fall markedly superior to it. There is hyperextension of L5 on the sacrum. This creates a facet syndrome of imbrication of the first sacral facet into the upper intervertebral foramen of L4-L5. The L4-L5 osseoligamentous canal area is also diminished in comparison to the superior L3-L4 level. The L4 vertebral body is anterior on the fifth lumbar body as seen both at the anterior and posterior vertebral body alignment. No pars interarticulares deformity is detected on this two film study.

Impression:

- 1. Dextroscoliosis of the lumbar spine
- 2. L4 hypoplastic facet development with anterolisthesis of L4 on L5 and intervertebral foraminal narrowing at the L4-L5 level
- 3. L5 hyperextension and facet syndrome of L5-S1 resulting in imbrication of the first sacral facet into the L5-S1 intervertebral foramen
- 4. Low right hemipelvis as noted above

Comment:

The findings suggest congenital etiology scoliosis. Oblique views with CT scanning would render further detailed anatomical structural confirmation of the impressions given in this report.

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