THE PERIOD PREVALENCE OF CONGENITAL THORACIC AND LUMBAR SPINE ANOMALIES AND THE ASSOCIATION BETWEEN THE LITERATURE REPORTED CLINICAL FEATURES OF THESE ANOMALIES WITH THE SUBJECT’S PRESENTING CLINICAL FEATURES

By

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Dissertation submitted in partial compliance with the requirements for the Master’s Degree in Technology: Chiropractic at Durban University of Technology.

I, Amashnee Pillay, do declare that this dissertation is representative of my own work in both conception and execution.

A. Pillay Date

APPROVED FOR FINAL SUBMISSION

_________________________ _______________________
Supervisor Date
Dr. J. Shaik
DEDICATION

I would like to dedicate this dissertation:

To God, for the courage, wisdom and strength he has bestowed upon me.

To my mother and father, I would like to thank you for your love, protection and support throughout this course and in every aspect of my life. I am eternally grateful and appreciative for all the sacrifices you have made to provide me with the best of everything.

To my brothers Deshindren, Trevern and Ravishen, I would like to thank you for all your love, encouragement and for tolerating me during the worst of times. I don’t think you’ll realise how much you guys helped me out, so thank you.
ACKNOWLEDGEMENTS

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I would, further, like to extend my thanks to the following people who assisted me in this dissertation:

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- Mrs. L. Twiggs and Mrs. P. Van den Berg for all their support during my clinic years and for accommodating my requirements during my research process.

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- My dear friends Prenasen, Avita, Vinotham, Farouk and Shaun for everything especially for believing in me and for all your prayers. I am eternally grateful for all your words of encouragement and support, through this dissertation.

- My new found friends Mageshri and Melanee. Knowing you'll has made this course so much easier. Thank you for the memories and especially Mageshri for all your tremendous help in the past two years.

- My extended family, my aunts, uncles and cousins for all their support and understanding during all my academic years and through this dissertation. A special thanks to my cousin Priandri and my sister-in-law Salosh for all your help and faith in me.
ABSTRACT

Background: Various congenital spinal anomalies are common findings in the general population. Their clinical significance is controversial with no definitive association been made between any specific congenital spinal anomaly to any clinical features.

Project Design: This research study was designed in the form of a quantitative, non-experimental, empirical clinical survey.

Method: Data was obtained from thoracic and lumbar spine radiographs contained in the Chiropractic Day Clinic at the Durban University of Technology from 1 January 1997 to 31 December 2005 and from the corresponding patient files. Through the research procedure, 519 thoracic and lumbar spine radiographs were located in the confines of the Chiropractic Day Clinic. Due to the exclusion criteria of a past or present history of trauma to the thoracic or lumbar spine areas, 147 radiographs were excluded.

Objectives
1. To determine the period prevalence (1 January 1997 – 31 December 2005) of congenital thoracic and lumbar spine anomalies.
2. To determine if there is any association between the presenting clinical features and the congenital thoracic and lumbar spine anomalies in general.
3. To determine if there is any association between the presenting clinical features and individual congenital thoracic and lumbar spine anomalies.
4. To compare subjects presenting clinical features with reported clinical features from literature.

Results:
The total period prevalence of congenital thoracic and lumbar spine anomalies for the period 1 January 1997 to 31 December 2005 was 26.3%. The prevalence of lumbosacral transitional vertebra, spinal bifida occulta and thoracolumbar transitional vertebra and “other“ (pig snout vertebra, Cupid’s bow sign, clasp knife syndrome, hypoplastic TVP, congenital Schmorl’s node) was 11.9%, 7.8%, 5.4% and 3.8% respectively. The clinical features that were significantly associated with congenital thoracolumbar, lumbar and lumbosacral spine anomalies in general were low back pain ($p = 0.003$), decreased lordosis ($p = 0.005$) and scoliosis ($p = 0.006$). A decreased lordosis was significantly associated ($p = 0.047$) with complete lumbarisation. Scoliosis and radicular/leg pain were significantly associated with spina bifida occulta ($p = 0.017$; $p = 0.028$, respectively) while decreased range of motion, scoliosis and decreased lordosis were significantly associated
with thoracolumbar transitional vertebra ($p = 0.020$; $p = 0.012$; $p = 0.014$, respectively). Radicular/leg pain and scoliosis associated with spina bifida occulta were the only clinical features associated with a spinal anomaly that correlated with those reported in the literature.

**Conclusions:**
The prevalence of thoracic and lumbar spine anomalies varies with the population sample. Some of these anomalies may be the cause of the clinical features related to the spine while others maybe of no clinical significance. More well-controlled studies are required to confirm or refute the findings of this study.
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<table>
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<th>Description</th>
</tr>
</thead>
<tbody>
<tr>
<td>ABCS</td>
<td>Alignment, Bone, Cartilage and Soft tissue</td>
</tr>
<tr>
<td>A-P</td>
<td>Anteroposterior</td>
</tr>
<tr>
<td>ATR</td>
<td>Achilles tendon reflex</td>
</tr>
<tr>
<td>C7</td>
<td>Cervical vertebra seven</td>
</tr>
<tr>
<td>CT</td>
<td>Computed tomography</td>
</tr>
<tr>
<td>DH</td>
<td>Disc herniation</td>
</tr>
<tr>
<td>DUT</td>
<td>Durban University of Technology</td>
</tr>
<tr>
<td>HT</td>
<td>Hair tufts</td>
</tr>
<tr>
<td>IVD</td>
<td>Intervertebral disc</td>
</tr>
<tr>
<td>LBP</td>
<td>Low back pain</td>
</tr>
<tr>
<td>L1-L5</td>
<td>Lumbar vertebra to the corresponding number</td>
</tr>
<tr>
<td>LSTV</td>
<td>Lumbosacral transitional vertebra</td>
</tr>
<tr>
<td>MRI</td>
<td>Magnetic resonance imaging</td>
</tr>
<tr>
<td>MW</td>
<td>Muscle wasting</td>
</tr>
<tr>
<td>N/A</td>
<td>Not available</td>
</tr>
<tr>
<td>ROM</td>
<td>Range of motion</td>
</tr>
<tr>
<td>S1-S5</td>
<td>Sacral segment to the corresponding number</td>
</tr>
<tr>
<td>SBO</td>
<td>Spina bifida occulta</td>
</tr>
<tr>
<td>S-lithesis</td>
<td>Spondylolisthesis</td>
</tr>
<tr>
<td>S-lysis</td>
<td>Spondylolysis</td>
</tr>
<tr>
<td>T1-T12</td>
<td>Thoracic vertebra to the corresponding number</td>
</tr>
<tr>
<td>TVP</td>
<td>Transverse process</td>
</tr>
<tr>
<td>TVPs</td>
<td>Transverse processes</td>
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INTRODUCTION

1.1 INTRODUCTION
Congenital spinal anomalies are structural defects present at birth as a result of anomalous vertebral development in the embryo (Letts and Jawadi, 2004). With respect to the thoracic and lumbar spine, anomalies of these regions can be classified into two subdivisions involving the vertebral body and posterior (neural) arch anomalies (Yochum and Rowe, 2005).

It is difficult to establish the prevalence and incidence of most congenital spinal anomalies as they are considered to be rare (Garcia et al., 1993). Esses (1995), Taylor and Resnick (2000) and Yochum and Rowe (2005) have, however, provided some epidemiological data for a few of these anomalies, but some of these reported data are inconsistent with each other.

Congenital anomalies of the spine may be simple and benign, causing no spinal deformity, or they may be complex, resulting in severe spinal deformity or even paraplegia (Letts and Jawadi, 2004). Drolet (2004) stated that most large neural tube defects are usually diagnosed at birth but there are many smaller or occult malformations that are only discovered when symptoms arise during childhood or even in adult life. While Hollingworth (1996) states that the most common clinical presentation of congenital defects of the spine are painless deformities, Drolet (2004) is of the view that it is imperative that congenital anomalies be detected and corrected before symptoms arise, as neurological impairment associated with some of these defects may be irreversible.

Letts and Jawadi (2004), Yochum and Rowe (2005) and other researchers have reported clinical features that are thought to be associated with congenital spinal anomalies but some of these reports are inconsistent and this could possibly lead to confusion with respect to recognizing clinical features associated with congenital thoracic and lumbar spinal anomalies amongst practitioners involved in spinal health care.

1.2 SCOPE OF THE STUDY
The current study took place at the Chiropractic Day Clinic at the Durban University of Technology (DUT) where thoracic and lumbar spine radiographs contained at the clinic for the period 1 January 1997 to 31 December 2005 were re-evaluated for any congenital
spinal anomalies and their corresponding patient files were looked at and all the information collected was recorded on data collection sheets (Appendix 1).

1.3 AIM AND OBJECTIVES

1.3.1 Aim

To determine the period prevalence (1 January 1997 to 31 December 2005) of congenital thoracic and lumbar spine anomalies and their association, if any, with the subjects presenting clinical features.

1.3.2 Objectives

1) To determine the period prevalence (1 January 1997 – 31 December 2005) of congenital thoracic and lumbar spine anomalies.

2) To determine if there was any association between the congenital thoracic or lumbar spine anomalies and the presenting clinical features of patients in general.

3) To determine if there is any association between individual congenital thoracic or lumbar spine anomalies and the presenting clinical features.

4) To compare subjects’ presenting clinical features with reported clinical features from literature.

1.4 THE HYPOTHESES

The Null Hypothesis (H₀) was set with respect to:

The period prevalence of the congenital thoracic and lumbar spine anomalies;

The association between the thoracic and lumbar spine anomalies and the presenting clinical features in general;

The association between the subjects’ presenting clinical features with reported clinical features

and stated that:

There shall not be a significant number of congenital thoracic or lumbar spine anomalies present for the chosen period of 1 January 1997 to 31 December 2005.

There shall be no significant association between the congenital thoracic or lumbar spine anomalies and the presenting clinical features in general.

There shall be little/no significant association between the subjects’ presenting clinical features with reported clinical features associated with thoracic or lumbar spinal congenital anomalies from the literature.
With respect to the association between the individual congenital thoracic or lumbar spine anomalies and the presenting clinical features, the Alternate Hypothesis ($H_a$) was, however, set, which stated that there shall be a significant association between the individual congenital thoracic or lumbar spine anomalies and the presenting clinical features. This was based on the findings of Hodges and Peck (1937); Winter et al. (1968) and Yochum and Rowe (2005).
CHAPTER 2
LITERATURE REVIEW

2.1 INTRODUCTION

Congenital spinal anomalies are defined as “structural defects of the vertebral column present at birth” (Berkow et al., 1997) that result from anomalous vertebral development in the embryo (Letts and Jawadi, 2004). These anomalies are physical defects that occur in an infant at birth irrespective of whether the defect was caused by a genetic factor or by a prenatal defect (Nace, 1999). The anomalies that affect the thoracic and lumbar spine maybe classified as either vertebral body anomalies or anomalies of the posterior arch (Yochum and Rowe, 2005).

2.2 OVERVIEW OF THE RELEVANT CLINICAL ANATOMY OF THORACIC AND LUMBAR VERTEBRAE

For the purpose of this study, the embryology of the thoracic and lumbar vertebra shall be outlined with incorporation of relevant congenital thoracic and lumbar spine anomalies where possible.

2.2.1 Embryology of the thoracic and lumbar vertebrae

A detailed sequence of embryologic steps must occur to result in the proper formation of both the bony and neural elements of the spine. Alterations in these steps can result in one or more congenital anomalies of the spine (Kaplan et al., 2005).

Vertebrae begin to develop during the embryonic period as mesenchymal condensations around the notochord. Later, this mesenchymal bone models chondrify and cartilaginous vertebrae form. Typical vertebrae begin to ossify toward the end of the embryonic period (8th week) and ossification continues throughout the fetal period. There are three primary ossification centers that develop in each cartilaginous vertebra, one in the centrum and one in each half of the vertebral arch. The halves of the vertebral arch begin to fuse with each other, from the lumbar to the cervical region, at approximately two years of age. The vertebral arches fuse to the centre of the vertebra in sequence, from the cervical to the lumbar regions, at approximately seven years of age (Moore and Dalley, 1999).

Five secondary ossification centers develop during puberty in each typical vertebra: one at the tip of the spinous process, one at the tip of each transverse process (TVP), two annular epiphyses (ring epiphyses), one on the superior and one on the inferior edge of
the centrum. The ring epiphyses usually unite with the vertebral body early in the adult period. All secondary ossification centers usually unite with the vertebra by the 25th year; the times of their union, however, may be variable. When there is a failure of half of a vertebral body to develop, the result is a hemivertebra. Another congenital anomaly, spina bifida occulta (SBO), occurs when the laminae of L5 and/or S1 fail to develop normally and fuse (Moore and Dalley, 1999).

The vertebral column in adults has four curvatures: cervical, thoracic, lumbar and sacral. The thoracic and sacral curvatures are primary curvatures that develop during the fetal period and are concave anteriorly, whereas the cervical and lumbar curvatures are secondary curvatures that begin to appear during the fetal period but do not become obvious until infancy and these are concave posteriorly. The cervical curvature becomes prominent when an infant begins to hold its head erect. The thoracic curvature results from the slightly wedge-shaped thoracic vertebral bodies and the lumbar curvature becomes obvious when an infant begins to walk and assumes an upright posture (Moore and Dalley, 1999).

2.2.2 Functional overview of the thoracic spine

The body of a typical thoracic vertebra (T2 to T8) is heart-shaped with both the horizontal and vertical dimensions of equal length. The pedicles and the laminae of the thoracic vertebrae are short when compared to the long slender spinous processes, which point in an oblique inferior direction. The transverse processes (TVPs) are thick, strong and relatively long with a concave facet on the anterior side. The intervertebral foramina in this region are essentially circular in shape and small when compared to the other areas of the spine (Bergmann et al., 1993).

The thoracic atypical vertebrae include T1 and T9 to T12. The vertebral body of T1 resembles that of C7 and possesses a whole facet for articulation with the first rib. The T9 vertebra may have one or two demifacets on either side. The T10 vertebra has one full rib facet located partly on the body of the vertebra and partly on the tubercle whereas the T11 vertebra has complete costal facets, but no facets on the TVPs for the rib tubercle. This vertebra also begins to take on characteristics of a lumbar vertebra, which includes a short and almost completely horizontal spinous process. The T12 vertebra has complete facets for articulation with the ribs but otherwise resembles a lumbar vertebra. The TVPs are replaced by superior, inferior and lateral tubercles. The normal thoracic spine has a kyphotic curve with a range of 20 to 50° [average of 45° degrees] (Bergmann et al., 1993).
It is a structural curve present from birth and maintained by the wedge-shaped vertebral bodies that are approximately 2 mm higher posteriorly than anteriorly (Bergmann et al., 1993). A vertebra, in which both the superior articular surfaces and the inferior articular surfaces were flat and facing posterolaterally and anteromedially, respectively was classified as a thoracic vertebra (Pal and Routal, 1999).

**2.2.3 Functional overview of the lumbar spine**

The typical lumbar vertebra has a large, kidney-shaped vertebral body designed to withstand the heavy loads imposed by the erect posture. It is wider from side to side than from anterior to posterior. The thick and broad spinous processes are hatchet-shaped structures and point straight posteriorly. The lumbar pedicles originate from the upper part of the body of the vertebra and extend horizontally and posteriorly. The pedicles are short and strong and the lumbar laminae are short, broad and strong and run in a vertical plane. The TVPs are long, slender and flattened on their anterior and posterior surfaces with L3 having the longest of the lumbar TVPs. The intervertebral foramina are large and triangular. The lumbar facets lie primarily in the sagittal plane, but become more coronal at the lumbosacral junction (Bergmann et al., 1993).

L5 is considered an atypical lumbar vertebra as it has the largest circumference of all the vertebrae and its body is thicker at its anterior aspect when compared with the posterior part. The TVPs are shorter and thicker and the spinous process is shorter and rounder than the other lumbar vertebrae. The secondary lordotic lumbar curve begins to develop when the child is about 9 to 12 months of age and is established at approximately 18 months. The lordotic curve begins at the L1-2 level and gradually increases at each level caudally to the sacrum, with an apex around the L3-4 intervertebral disc (IVD). The normal range for the lumbar lordosis is 20 to 60º (Bergmann et al., 1993). A vertebra, in which both superior articular surfaces and inferior articular surfaces were curved and facing posteriomedially and anterolaterally, respectively were classified as a lumbar vertebra (Pal and Routal, 1999).

**2.3 THE EPIDEMIOLOGICAL DATA OF CONGENITAL THORACIC AND LUMBAR SPINAL ANOMALIES**

A summary of the available epidemiological data of congenital thoracic and lumbar spinal anomalies is presented in **Table 2.1**.
Table 2.1 Epidemiological data of congenital thoracic and lumbar spinal anomalies

<table>
<thead>
<tr>
<th>Anomaly</th>
<th>Reference</th>
<th>Population/Sample</th>
<th>Incidence</th>
<th>Prevalence</th>
</tr>
</thead>
<tbody>
<tr>
<td>LSTV</td>
<td>Tilley (1970)</td>
<td>7,236 radiographs</td>
<td>n = 1,103 (14%)</td>
<td>N/A</td>
</tr>
<tr>
<td></td>
<td>Hahn et al. (1992)</td>
<td>200 consecutive clinic outpatients; 112 males and 88 females; 5-85 years old</td>
<td>n = 24 (12%)</td>
<td>N/A</td>
</tr>
<tr>
<td></td>
<td>Esses (1995)</td>
<td>N/A</td>
<td>N/A</td>
<td>N/A</td>
</tr>
<tr>
<td></td>
<td>Kamanli and Genc (2002)</td>
<td>503 male athletes</td>
<td>n = 37 (7.4%)</td>
<td>4.8%</td>
</tr>
<tr>
<td></td>
<td>Hughes and Saifuddin (2004)</td>
<td>General population</td>
<td>N/A</td>
<td>4.2%</td>
</tr>
<tr>
<td></td>
<td>Beck et al. (2004)</td>
<td>847 clinic outpatients</td>
<td>N/A</td>
<td>9.8%</td>
</tr>
<tr>
<td></td>
<td>Treble et al. (2005)</td>
<td>109 patients with isthmic spondylolisthesis</td>
<td>N/A</td>
<td>n = 10 (9.2%)</td>
</tr>
<tr>
<td>SBO</td>
<td>Yochum and Rowe (2005)</td>
<td>General population</td>
<td>4-10%</td>
<td>N/A</td>
</tr>
<tr>
<td></td>
<td>Esses (1995)</td>
<td>N/A</td>
<td>N/A</td>
<td>0.5%</td>
</tr>
<tr>
<td></td>
<td>Kamanli and Genc (2002)</td>
<td>503 male athletes</td>
<td>n = 107 (21.4%)</td>
<td>N/A</td>
</tr>
<tr>
<td></td>
<td>Beck et al. (2004)</td>
<td>847 clinic outpatients</td>
<td>N/A</td>
<td>6.7%</td>
</tr>
<tr>
<td></td>
<td>Yochum and Rowe (2005)</td>
<td>General population</td>
<td>10-22%</td>
<td>N/A</td>
</tr>
<tr>
<td></td>
<td>Treble et al. (2005)</td>
<td>109 patients with isthmic spondylolisthesis</td>
<td>N/A</td>
<td>n = 34 (31.1%)</td>
</tr>
<tr>
<td>Facet tropism</td>
<td>Kamanli and Genc (2002)</td>
<td>503 male athletes</td>
<td>1%</td>
<td>N/A</td>
</tr>
<tr>
<td></td>
<td>Beck et al. (2004)</td>
<td>847 clinic outpatients</td>
<td>N/A</td>
<td>0.7%</td>
</tr>
<tr>
<td></td>
<td>Yochum and Rowe (2005)</td>
<td>General population</td>
<td>20-35%</td>
<td>N/A</td>
</tr>
<tr>
<td></td>
<td>Beck et al. (2004)</td>
<td>847 clinic outpatients</td>
<td>N/A</td>
<td>1.4%</td>
</tr>
<tr>
<td>Block vertebra</td>
<td>Davies (1975)</td>
<td>10,000 births</td>
<td>N/A</td>
<td>0.05 - 0.1%</td>
</tr>
<tr>
<td>Hemivertebra</td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
</tbody>
</table>

LSTV = Lumbosacral transitional vertebra; SBO = Spina bifida occulta; N/A = not available

Tilley (1970) reported the incidence of lumbosacral transitional vertebra (LSTV) to be 14%. Hahn et al. (1992) reported a similar finding in a prospective study of 200 patients referred for the evaluation of symptoms related to the lumbosacral spine (Table 2.1). They identified 24 cases of LSTV, of which 15 were sacralisation of L5 and nine were lumbarisation of S1. Hughes and Saifuddin (2004) have reported a higher prevalence of LSTV than Esses (1995) whereas Treble et al. (2005) reported a similar prevalence of LSTV as Beck et al. (2004) (Table 2.1).

The prevalence of hemivertebra was reported by Davies (1975) to be very low (Table 2.1). No other literature on the epidemiological data of this anomaly could, however, be found. Kamanli and Genc (2002) reported on the overall incidence of lumbosacral congenital anomalies using radiographic interpretation in healthy and active young male athletes. The spinal congenital anomalies that were identified included LSTV, spina bifida occulta (SBO) and facet tropism (Table 2.1). Yochum and Rowe (2005) reported similar findings with respect to LSTV and SBO but not of facet tropism (Table 2.1).

Beck et al. (2004) conducted a study at the New Zealand College of Chiropractic to determine the occurrence rates for radiographically detectable abnormalities (i.e. any condition that could have affected the spine and not merely confined to congenital anomalies) of the spine. Their findings show a low prevalence of SBO, LSTV, facet tropism and block vertebra (Table 2.1).
Treble et al. (2005) conducted a retrospective study on radiological and clinical analysis of cases with isthmic spondylolisthesis. A total of 109 of isthmic spondylolisthesis were found from three student outpatient clinics. Their reported prevalence of SBO conflicts with the earlier reports of Esses (1995) and Beck et al. (2004) (Table 2.1).

In conclusion, it is difficult to report on established incidences and prevalences of congenital thoracic and lumbar spinal anomalies, as there are inconsistent reports in the literature as shown in Table 2.1. Furthermore, despite an exhausting research of the current literature (journals, books and the Internet), no epidemiological data on spinal anomalies such as butterfly vertebra, agenesis of lumbar pedicle, pig snout vertebra, Cupid’s bow sign, clasp knife syndrome, hypoplastic TVPs and congenital Schmorl’s node could be found.

2.3.1 Location of individual congenital spinal anomalies
The vertebral location of SBO as reported in the literature is presented in Table 2.2.

**Table 2.2 Location of spina bifida occulta**

<table>
<thead>
<tr>
<th>Reference</th>
<th>Level</th>
<th>Prevalence</th>
</tr>
</thead>
<tbody>
<tr>
<td>Kamanli and Genc (2002)</td>
<td>S1</td>
<td>17%</td>
</tr>
<tr>
<td></td>
<td>S1 &amp; S2</td>
<td>2%</td>
</tr>
<tr>
<td></td>
<td>L5</td>
<td>5%</td>
</tr>
<tr>
<td>Yochum and Rowe (2005)</td>
<td>L5 &amp; S1</td>
<td>0.1%</td>
</tr>
<tr>
<td></td>
<td>S1</td>
<td>15-17%</td>
</tr>
<tr>
<td></td>
<td>L5</td>
<td>1-6%</td>
</tr>
</tbody>
</table>

The most likely location of SBO is S1 followed by L5 (Kamanli and Genc, 2002; Yochum and Rowe, 2005). The prevalence of SBO existing in two levels concurrently (i.e. S1 and S2 or L5 and S1) is very low (Table 2.2). Taylor and Resnick (2000) and Yochum and Rowe (2005) reported that facet tropism was most commonly seen at the L5-S1 level followed by the L4-L5 level. Taylor and Resnick (2000) also stated that block vertebrae in the thoracic or lumbar spine commonly occurred at the T12-L1 and L4-L5 levels and Cupid’s bow sign was most commonly seen at the L4 and L5 level.

2.4 IDENTIFYING CONGENITAL THORACIC AND LUMBAR SPINE ANOMALIES
The studies investigating the use of diagnostic imaging modalities for identifying thoracic or lumbar spinal anomalies are summarised in Table 2.3.
Table 2.3 Diagnostics imaging modalities used for identifying thoracic or lumbar spinal anomalies

<table>
<thead>
<tr>
<th>Reference</th>
<th>Radiographs</th>
<th>CT</th>
<th>MRI</th>
</tr>
</thead>
<tbody>
<tr>
<td>Wigh and Anthony, (1981)</td>
<td>Thoracolumbar transitional vertebra</td>
<td>N/A</td>
<td>N/A</td>
</tr>
<tr>
<td>Garcia et al. (1993)</td>
<td>Butterfly vertebra</td>
<td>N/A</td>
<td>N/A</td>
</tr>
<tr>
<td>Garcia et al. (1993)</td>
<td>Butterfly vertebra</td>
<td>N/A</td>
<td>N/A</td>
</tr>
<tr>
<td>Hughes and Saifuddin, (2004)</td>
<td>LSTV</td>
<td>N/A</td>
<td>N/A</td>
</tr>
<tr>
<td>Yochum and Rowe (2005)</td>
<td>Block vertebra, butterfly vertebra, hemivertebra, SBO, LSTV, lumbar pedicle agenesis</td>
<td>Congenital Schmorl's node, SBO, Facet tropism, lumbar pedicle agenesis</td>
<td>Congenital Schmorl's node, facet tropism, lumbar pedicle agenesis</td>
</tr>
</tbody>
</table>

N/A = not available; CT = computed tomography; MRI= magnetic resonance imaging

Frymoyer et al. (1984) recommended the use of plain film radiographs to evaluate the radiographic features of spinal anomalies. Plain film radiographs should be used to analyse the thoracolumbar junction so that the L1 vertebral body can be correctly identified by differentiating the large TVPs from hypoplastic ribs (Wigh and Anthony, 1981). Garcia et al. (1993) reported that the best radiographic view for identifying a butterfly vertebra is the anterior-posterior (AP) radiograph of the lumbar spine. Hughes and Saifuddin (2004) reported that the plain film radiograph is the imaging modality of choice if an LSTV is suspected. An MRI may, however, be utilized to confirm the diagnosis of LSTV if it is not apparent on the plain film radiograph. Despite Bush and Kalen (1999) reporting that CT was used for identifying hemivertebrae and unsegmented bars, these anomalies are usually easily identified on plain film radiographs (Yochum and Rowe, 2005).

In conclusion, plain film radiographs appear to be the diagnostic imaging modality of choice to identify congenital spinal anomalies. Fordham (2004), however, suggested that in addition to plain film radiographs, CT and MRI have a role in imaging the primary abnormality, the monitoring of treatment affects and potential complications that may develop over time.

2.5 CLINICAL FEATURES ASSOCIATED WITH CONGENITAL THORACIC AND LUMBAR SPINE ANOMALIES

Congenital anomalies of the spine may be simple and benign, causing no spinal deformity, or they may be complex, resulting in severe spinal deformity or even paraplegia (Letts and Jawadi, 2004). While Hollingworth (1996) states that the most common clinical presentation of congenital defects of the spine are painless deformities, Drolet (2004) is of
the view that it is imperative that congenital anomalies be detected and corrected before symptoms arise, as neurological impairment associated with some of these defects maybe irreversible.

A summary of the studies that reported clinical features associated with congenital thoracic and lumbar spinal anomalies is presented in Table 2.4.

**Table 2.4 Reported clinical features associated with congenital thoracic and lumbar spinal anomalies**

<table>
<thead>
<tr>
<th>Reference</th>
<th>Spina bifida</th>
<th>LSTV</th>
<th>Facet tropism</th>
<th>Block vertebra</th>
<th>Hemi-vertebra</th>
<th>Butterfly vertebra</th>
<th>Lumbar pedicle agenesis</th>
<th>Clasp knife syndrome</th>
</tr>
</thead>
<tbody>
<tr>
<td>Hodges and Peck (1937) DeAnguin (1959)</td>
<td>Back pain, sciatica</td>
<td>N/A</td>
<td>Back pain</td>
<td>N/A</td>
<td>N/A</td>
<td>N/A</td>
<td>N/A</td>
<td>N/A</td>
</tr>
<tr>
<td>Farfan and Sullivan, (1967)</td>
<td>N/A</td>
<td>N/A</td>
<td>LBP, Sciatica, DN</td>
<td>N/A</td>
<td>N/A</td>
<td>N/A</td>
<td>N/A</td>
<td>N/A</td>
</tr>
<tr>
<td>Winter et al. (1968)</td>
<td>Scoliosis</td>
<td>N/A</td>
<td>N/A</td>
<td>N/A</td>
<td>N/A</td>
<td>N/A</td>
<td>N/A</td>
<td>N/A</td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td></td>
<td>Scoliosis</td>
<td>N/A</td>
<td>N/A</td>
<td>N/A</td>
<td>N/A</td>
</tr>
<tr>
<td></td>
<td>Scoliosis</td>
<td>N/A</td>
<td>N/A</td>
<td>N/A</td>
<td>N/A</td>
<td>N/A</td>
<td>N/A</td>
<td>N/A</td>
</tr>
<tr>
<td></td>
<td>Lipoma, HT pigmentation, sacral dimples, Scoliosis</td>
<td>N/A</td>
<td>N/A</td>
<td>N/A</td>
<td>N/A</td>
<td>N/A</td>
<td>N/A</td>
<td>N/A</td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td></td>
<td>DN, sciatica</td>
<td>N/A</td>
<td>N/A</td>
<td>N/A</td>
<td>N/A</td>
</tr>
<tr>
<td></td>
<td>Clubfoot</td>
<td>N/A</td>
<td>N/A</td>
<td>N/A</td>
<td>N/A</td>
<td>N/A</td>
<td>N/A</td>
<td>N/A</td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td></td>
<td>Lower limb reflexes, MW, lumbosacral lipoma.</td>
<td>N/A</td>
<td>N/A</td>
<td>N/A</td>
<td>N/A</td>
</tr>
<tr>
<td></td>
<td>Urinary and fecal incontinence</td>
<td>N/A</td>
<td>N/A</td>
<td>N/A</td>
<td>N/A</td>
<td>N/A</td>
<td>N/A</td>
<td>N/A</td>
</tr>
</tbody>
</table>

**Notes:**
- LSTV = Lumbosacral transitional vertebra; N/A = not available; ATR = Achilles tendon reflex; ROM = range of motion; S-lysis = spondylolysis; S-listhesis = spondyloolisthesis; MW = muscle wasting; DN = disc herniation; HT = Hair tufts.
2.5.1 MUSCULOSKELETAL FEATURES ASSOCIATED WITH CONGENITAL THORACIC AND LUMBAR SPINAL ANOMALIES

Low back pain / Back pain
- Despite Hodges and Peck (1937) reports of an increased likelihood of low back pain (LBP) in individuals with SBO (Table 2.4), other researchers have not corroborated these findings (O'Reilly, 1921; Breck et al., 1944; Yochum and Rowe, 2005).
- Tilley (1970) reported that individuals with sacralisation had a greater occurrence of LBP than those without the condition.
- Farfan and Sullivan (1967) stated that individuals with facet tropism are likely to suffer LBP, as there is a decreased ability of that vertebral joint to resist rotational stresses. Fischer et al., (1958) had, however, earlier reported a higher incidence of facet asymmetry in a group of 100 asymptomatic individuals compared to 200 individuals with LBP.
- Paillas et al. (1977) reported that back pain and sciatica were twice as common in individuals with transitional vertebrae compared to those who did not have these anomalies. This conflicts with the report of Tini et al. (1977) that individuals with a transitional vertebra were not more likely to experience back pain than controls.

Neck Pain
Yochum and Rowe (2005) reported that occasionally certain cervical congenital anomalies, such as block vertebrae, may co-exist with thoracic and lumbar anomalies and could possibly be a cause of neck pain. No other literature reported an association between neck pain and any congenital thoracic or lumbar spine anomaly.

Headaches
Headaches may result from hypertonic cervical muscles (Simons et al., 1999). Hypertonic cervical muscles have been shown to be associated with certain anomalies of the cervical spine e.g. block vertebra/e and this could account for the headache experienced by individuals with these anomalies (Ganasram, 2006). It is possible that cervical congenital anomalies may co-exist with thoracic and/or lumbar spine anomalies. No literature has reported thoracic or lumbar spinal anomalies to be directly associated with headaches.

Decreased thoracic and/or lumbar range of motion
Reduced mobility or decreased range of motion may be detected clinically, in patients with block vertebra especially if the block occurs at the thoracolumbar junction (Yochum and Rowe, 2005). No other literature could be found to associate decreased range of motion (ROM) with other thoracic or lumbar spinal anomalies.
Scoliosis
Congenital scoliosis is a lateral curvature of the spine that occurs as a result of congenital anomalies of vertebral development. The vertebral abnormalities are present at birth, but clinical deformity may not be evident until later in childhood, when progressive scoliosis is evident (Letts and Jawadi, 2004). Winter et al. (1968) distinguished congenital scoliosis from other causes of scoliosis by associated anomalies of the vertebrae or ribs. The most frequently observed vertebral anomalies were hemivertebrae, block vertebrae, spina bifida and bridging vertebral bars. Yochum and Rowe (2005) found that individuals with an SBO had a high incidence of congenital scoliosis (Table 2.4). Mohanty and Kumar (2000) reported that the most common type of deformity was a right-sided thoracolumbar curve in 66 cases of congenital scoliosis, with the hemivertebra identified as the most likely cause.

Kyphosis
Congenital kyphosis is less common than congenital scoliosis. Progression of this deformity may result in paraplegia (Letts and Jawadi, 2004). Two types of congenital kyphosis exist:

- Defects of segmentation occur often in midthoracic or thoracolumbar regions and may involve 2-8 levels. They tend to produce a round kyphosis rather than a sharp angular gibbus deformity; therefore, paraplegia rarely is a problem. The main clinical symptom is LBP caused by a compensatory lumbar hyperlordosis (Letts and Jawadi, 2004).

- Defects of formation are more common and may involve one level, but multiple defects are possible. The failure of the formation can be purely anterior, resulting in kyphosis or can be anterolateral with a posterior hemivertebra, resulting in kyphoscoliosis (Letts and Jawadi, 2004).

McMaster and Singh (1999) reported that progressive thoracolumbar kyphosis is usually due to a butterfly vertebra located in the same region.

Lordosis
Congenital lordosis is the least common of the three major patterns of congenital spinal deformities (Letts and Jawadi, 2004). Three factors may contribute to the development of congenital lordosis in the thoracic spine: firstly, the posterior defect of segmentation with concomitant normal anterior development, secondly, abnormal or lack of formation of the posterior elements, and finally, it may be a compensatory deformity as a result of a kyphosis at a lower vertebral level (Dubousset, 1994; Letts and Jawadi, 2004).
Foot deformities

Clubfoot, also known as talipes equinovarus (Moore and Dalley, 1999), is seen in approximately 1 in 1000 births, with men affected twice as commonly as women. Rasool et al. (1992) and Mazur (2005) have reported a strong association between SBO and clubfoot deformity in children.

2.5.2 Neurological Features

Muscle weakness and Hyporeflexia

Muscle weakness of the upper or lower extremity has been reported in individuals with SBO (Zatouroff, 1976; Berkow et al., 1997). Berkow et al. (1997) explained that damage of the nerves at the affected levels accounted for this clinical feature. Weatherall et al. (1985) reported an association between absent ankle tendon reflexes and SBO. Nadkarni et al. (2005) presented a case review of an 18 month-old male child with SBO who presented with delayed ability to sit, stand and walk and other neurological changes (Table 2.4).

Gait abnormalities

A study by Duffy et al. (1999) was designed to determine gait patterns in children with lumbar and sacral level spina bifida. A group of 28 children were examined: 10 had L4-level lesions, eight had L5-level lesions, and 10 had S1-level lesions. A group of 15 children with no SBO was used for comparison. Each child underwent a three-dimensional gait analysis. It was found that there were recognisable gait patterns for each level of spina bifida and that the abnormalities accurately reflected the muscle deficiencies present. The gait patterns approximated more closely to those of the normal group as the neurological level descended. The most important findings were of increased pelvic obliquity and rotation with hip abduction in stance (reflecting the gross Trendelenburg-type gait seen in these children) and persistent knee flexion throughout the stance as a result of the absence of the plantar flexion-knee extension couple.

Paraesthesia and Radicular pain/sciatica

DeAnguin (1959) reported that sacral nerve root compression from clasp knife syndrome may produce radiating pain or paraesthesia. Myelomeningocele, which is a complication of SBO, may cause neurological symptoms such as paraesthesia (Esses, 1995). Intervertebral disc herniations have been associated with clasp knife syndrome (DeAnguin, 1959), facet tropism (Hagg and Wallner, 1990) and less commonly with LSTV (Yochum and Rowe, 2005) giving rise to radicular pain or sciatica.
**Bowel and bladder disorders**
Verhoef *et al.* (2005) reported that urinary incontinence (61%) and fecal incontinence (34%) were associated with SBO. Bowel and bladder symptomologies were also reported in 95% of children with myelomeningocele, possibly due to damage to the nerve supply to these organs (Zickler and Richardson, 2004). Constipation and a slower intestinal transit time have been reported in individuals with lumbosacral SBO (Thorpe *et al*., 1994).

### 2.5.3 Cardiorespiratory symptoms
There is no literature that directly relates cardiorespiratory symptoms with congenital thoracic or lumbar spinal anomalies, but an association between dyspnea and coughing has been shown with congenital scoliosis and spondylocostal dysostosis (Mohanty and Kumar, 2000), which is a congenital disorder associated with hemivertebrae, block vertebrae, hypoplastic vertebrae and butterfly vertebrae (Nadkarni *et al*., 2005).

### 2.5.4 Cutaneous lesions
Zatouroff (1976), Drolet, (2004), Dickson (2004) and Yochum and Rowe (2005) have reported hair tufts in individuals with SBO. Weatherall *et al.* (1985) and Drolet (2004) reported an association between spina bifida and patches of pigmentation on the sacral area. Drolet (2004), Dickson (2004) and Yochum and Rowe (2005) have also found a close association between sacral dimples and spina bifida. Lipomas are common cutaneous lesions that have been reported in individuals with SBO (Drolet, 2004 and Nadkarni *et al*., 2005).

### 2.5.5 Other Clinical features
SBO was found to occur twice as commonly in patients with idiopathic epilepsy compared to patients with known causes of epilepsy and in the general population (Klepel and Freitag, 1992). Congenital spinal anomalies are also associated with an increased incidence of congenital anomalies in other systems, viz. the VATERL syndrome. V is vertebral, A is anal atresia TE is tracheo-esophageal, R is renal and L is limb (Dickson, 2004). Spinal anomalies that may be associated with the VATERL syndrome are hemivertebra, butterfly vertebra, block vertebra and SBO (meningocele) (Epstein, 1976). In the presence L5 SBO, there is a 13-fold greater incidence of L5 spondylolisthesis. SBO of S1 has been found in nearly 50% of L5 spondylolisthesis (Fischer *et al*., 1958).
2.6 CONCLUSION

The prevalence and incidence of congenital thoracic and lumbar spinal anomalies vary according to the specific anomaly. A review of the related literature reveals that little information exists regarding the true prevalence of certain individual anomalies. The epidemiological data pertaining to these anomalies vary depending on the sample of the population. This may explain the inconsistent reports in the literature (Table 2.1).

The debate still persists amongst researchers with respect to the clinical features thought to be associated with these spinal anomalies. Some are of the view that they may be clinically silent or result in painless spinal deformities (Hollingworth, 1996) while others are of the view that severe spinal deformities and other pronounced clinical features may be associated with congenital thoracic and/or lumbar spine anomalies (Letts and Jawadi, 2004). Furthermore, as shown in Table 2.4, the reports in the literature appear to be inconsistent with respect to the clinical features thought to be associated with these spinal anomalies.
CHAPTER 3
MATERIALS AND METHODS

3.1 INTRODUCTION
This chapter will include a detailed description of the study design, the selection of the thoracic and lumbar spine radiographs and patient files and data collection. The statistical procedures that were implemented in analyzing the data have been included in this chapter. Ethical clearance for this study was obtained from the Faculty of Health Sciences Research Committee, DUT.

3.2 STUDY DESIGN
This research study was designed in the form of a quantitative, non-experimental, empirical clinical survey. Data was obtained from the thoracic and lumbar spine radiographs and corresponding patient files that were present at the Chiropractic Day Clinic at the DUT from 1 January 1997 to 31 December 2005. This time frame was selected due to the availability of the radiographs as all radiographs taken prior to 1 January 1997 were either given to patients or used for teaching purposes.

3.2.1 Patient Confidentiality
Steps were undertaken to maintain patient confidentiality throughout the process of the study. All patients at the Chiropractic Day Clinic sign an informed consent form that stipulates that their clinical records may be inspected for research purposes only and strict guidelines (DUT policy) are followed. All information that was obtained from the radiographs and patient files were reduced to code form. To further maintain confidentiality only the researcher and the research supervisor examined the patient files and radiographs. Where required, an external qualified radiologist helped in verifying any radiograph when the diagnosis was uncertain.

3.2.2 Sampling and Sample Allocation
In this study, convenience sampling was used. To improve reliability and avoid confusion this study took place in four phases. All the information collected was recorded on data collection sheets (Appendix 1) and patient files were evaluated first to avoid researcher bias.
3.2.3 Inclusion and Exclusion Criteria

Inclusion Criteria
1) This study was limited to patients’ radiographs and files within the archives of the Chiropractic Day Clinic at the Durban University of Technology from 1 January 1997 to 31 December 2005.

2) All thoracic and lumbar spine radiographs and corresponding files, with the exception of those that satisfied the exclusion criteria, were used and those that did not have a congenital thoracic or lumbar spinal anomaly were utilized as a control.

Exclusion Criteria
1) Patient files that revealed a past or present history of trauma to the thoracic or lumbar spine area were excluded to avoid confusion that could have arisen with regards to the clinical manifestations.

2) Any radiographs that did not fall in the allocated time period (1 January 1997 to 31 December 2005) were excluded.

3.2.4 Data Collection
Data collection involved both primary data and secondary data.

3.2.4.1 The Primary Data
The primary data was obtained from patient files and from the thoracic and lumbar spine radiographs.

(A) File Selection
Patient files, corresponding to thoracic and lumbar spine radiographs that were present at the Chiropractic Day Clinic, were evaluated with focus on the following:
- Presenting complaint
- Medical history (including any history of spinal trauma)
- Orthopaedic examination findings of the thoracic or lumbar spine

(B) Radiograph Selection
All the thoracic and lumbar spine radiographs were evaluated according to the Alignment, Bone, Cartilage and Soft tissue (ABCS) guidelines by Yochum and Rowe (2005). For the purpose of this study the classification system of Yochum and Rowe (2005) was utilized. They are as follows:
1. Vertebral body anomalies:

**Block vertebra**
This refers to two vertebrae that are osseously fused. Embryologically, this is the failure of the segmentation process of the somites during the period of differentiation at 3 to 8 foetal weeks (Burgener and Kormano, 1997). Radiographically, the cardinal findings are small vertebral bodies, variable degrees of bony fusion, a small hypoplastic disc which is often calcified and associated anomalies of the posterior arch (Yochum and Rowe, 2005).

**Hemivertebra**
Failure of ossification of part of the vertebral body produces a hemivertebra. Three types are recognized on the basis of location: lateral, dorsal, and ventral (Yochum and Rowe, 2005). Vertebral body originally develops from paired chondral centers, which at a later stage form a single ossification focus that is separated transiently by the notochordal remnant into anterior and posterior centers. Lateral hemivertebra results from failure of development of one of the paired chondral centers. Dorsal and ventral hemivertebra result from agenesis of either the anterior or posterior portion of the growth center, respectively (Taylor and Resnick, 2000).

**Butterfly Vertebra**
When the lateral ossification centers fail to unite, the result is a butterfly vertebra (Yochum and Rowe, 2005). This occurs when two halves of vertebra assumes an appearance of a butterfly’s wings when viewed from front, when viewed from the center it resembles an “hourglass” (Burgener and Kormano, 1997). There will be a widened interpediculate distance and the pedicles of the involved vertebra may be larger than normal (Yochum and Rowe, 2005).

2. Posterior (neural) arch anomalies:

**Agenesis of a lumbar pedicle**
Absence of a lumbar pedicle is failure of ossification within the neural arch (Yochum and Rowe, 2005). Radiologically, it maybe accompanied by unusually large intervertebral foramen, absent lamina or articular processes (Epstein, 1962).

**Spina bifida occulta**
Spina bifida occulta (SBO) refers to the developmental failure of osseous union between the two halves of the posterior arch, typically resulting in a sagittal midline cleft of variable size without posterior herniation of the thecal sac or its contents. Radiographically, it
appears as a vertical or oblique radiolucent cleft between the lamina (Yochum and Rowe, 2005).

**Spina bifida vera**
This is a wide bony defect in the posterior arch of the lumbar vertebrae, usually over more than one segment, with the protrusion of the spinal cord contents (meninges, cerebrospinal fluid, nerve roots) beyond the confines of the spinal canal. Herniation of the CSF-filled sac covered with meninges is called a meningocele. A sac containing CSF and neural elements is called a myelomeningocele, and if neural elements project through the bony defect without thecal covering, then a myelocoele is present (Yochum and Rowe, 2005).

**Facet tropism**
Vertebral facet asymmetry (tropism) is a condition in which the facet joint surfaces of adjacent vertebrae are oriented in different spatial planes when comparing the right versus the left facets. Tropism at any level will manifest itself radiographically as nonvisualisation of the facet joint space on one side, while being clearly seen on the contralateral side (Yochum and Rowe, 2005).

**Transitional vertebrae**
An LSTV is either sacralisation (failure of segmentation of the L5 vertebra from the S1 segment) or lumbarisation (when the S1 segment develops partial or complete lumbar type morphology). Complete lumbarisation results in the presence of six lumbar-type vertebra. A characteristic of this type transitional vertebra is distinctive enlargement of the transverse processes (Yochum and Rowe, 2005).

According to Pal and Routal (1999), a transitional vertebra at the thoracolumbar junction is identified as any vertebra showing articular surface(s) orientation in a plane different to that described in Chapter 2.

**3. Other anomalies:**

**Pig snout vertebra**
An anomalous malformation of the transverse process of a lumbar vertebra that is orientated superiorly, creating the pig snout appearance of the “scotty dog” (oblique view of the lumbar spine) (Yochum and Rowe, 2005).
Cupid’s bow sign
A smooth parasagittal endplate concavity with a thickened cortex that is separated in the midline by a smooth convex hump is called the Cupid’s bow sign (Yochum and Rowe, 2005).

Clasp knife syndrome
Elongation of L5 spinous process that invaginates into an SBO (especially in lumbar extension) of at least the S1 segment (Yochum and Rowe, 2005).

Congenital Schmorl’s node
Herniation of disc material into the vertebral body. The nucleus pulposus breaches the cartilaginous endplate and occupies an intraosseous location. The age of initial occurrence is often indeterminate but is most likely during adolescence. Later onset is usually associated with significant trauma or bone pathology (Yochum and Rowe, 2005).

3.2.4.2 The Secondary Data
This was obtained from various sources that included journals, books, the Internet and discussions with radiologists and anatomists.

3.2.5 Research Procedure
This research took place in four phases to maintain order and to avoid any confusion that would have arisen.

3.2.5.1 Phase One
Radiographs contained in the confines of the Chiropractic Day Clinic at the Durban University of Technology were sorted. All the thoracic and lumbar spine radiographs from 1 January 1997 to 31 December 2005 were obtained and placed aside.

3.2.5.2 Phase Two
Patient names present on the radiograph envelopes were recorded to help assist the researcher in obtaining the corresponding patient files via the Chiropractic Day Clinic computer system. Once the patient files were drawn, patient names were converted to code form to maintain patient confidentiality. The files were evaluated with focus on the presenting complaint, medical history (including spinal trauma) and orthopaedic examination findings of the thoracic or lumbar spine.

All information that was gathered was recorded on Data Collection Sheet 1 (Appendix 1), which contained the following:
- Code
- History of trauma, present or not
- The presenting complaint
- Principle clinical features, including spinal (orthopaedic examination) features.

3.2.5.3 Phase Three
Patient files were evaluated and those that had a past or present history of trauma to the thoracic or lumbar spine area were excluded.

3.2.5.4 Phase Four
First the researcher evaluated all the radiographs using the ABCS guidelines, this was followed by confirmation of the findings by the researcher, supervisor and finally the radiograph report was looked at and the diagnosis was confirmed. A qualified and experienced radiologist was consulted in cases where there was doubt in the radiological diagnosis. It must be noted that all these radiographs had been evaluated and reported on by a qualified and experienced radiologist prior to the examination by this researcher.

All information that was gathered was recorded on Data Collection Sheet 2 (Appendix 2) and contained the following:
- Code
- Date
- The type of spinal anomaly that is present or not
- The location of the anomaly
- The description of the anomaly
- Whether it was identified by the researcher, radiologist or both.

3.3 STATISTICAL ANALYSIS
The statistical package SPSS, version 13 (SPSS Inc, Chicago, Illinois, USA) was used in the analyses of the data in this study.

3.3.1 Method of data analysis
The period prevalence of congenital thoracic and lumbar spine anomalies was determined using descriptive analysis, which included proportions, and counts that were presented for categorical variables, and means and standard deviations that were presented for quantitative variables. The α level of significance was set at 0.05.
In determining the associations between clinical features and presence of any congenital spinal anomalies, Fischer’s exact tests were used. Sensitivity, specificity and 95% confidence intervals were calculated using EpiCalc 2000 (Gilman and Myatt, 1998).
CHAPTER 4
RESULTS

4.1 THE PERIOD PREVALENCE OF CONGENITAL THORACIC AND LUMBAR SPINE ANOMALIES

Through the research procedure, 519 thoracic and lumbar spine radiographs were located within the confines of the Chiropractic Day Clinic at the DUT from 1 January 1997 to 31 December 2005. One hundred and forty seven radiographs were excluded according to the guidelines of the exclusion criteria.

Of the remaining 372 radiographs meeting the inclusion criteria for this study, 98 were positive for any congenital thoracic or lumbar spinal anomaly, which equated to a period prevalence of 26.3%. The number of congenital thoracic and lumbar spinal anomalies per subject ranged from zero to three and are shown in Table 4.1.

Table 4.1 Number of congenital thoracic and lumbar spine anomalies per subject (n = 372)

<table>
<thead>
<tr>
<th>Frequency</th>
<th>Percent</th>
</tr>
</thead>
<tbody>
<tr>
<td>0</td>
<td>274</td>
</tr>
<tr>
<td>1</td>
<td>73</td>
</tr>
<tr>
<td>2</td>
<td>23</td>
</tr>
<tr>
<td>3</td>
<td>2</td>
</tr>
<tr>
<td>Total</td>
<td>372</td>
</tr>
</tbody>
</table>

The prevalence of the individual congenital thoracic and lumbar spine anomalies is depicted graphically in Figure 4.1. The three most common anomalies identified were SBO in the sacrum (6.5%), complete lumbarisation (5.7%) and thoracolumbar transitional vertebra (5.4%). The prevalence of “other” anomalies [pig snout vertebra, Cupid’s bow sign, clasp knife syndrome, hypoplastic TVP, and congenital Schmorl’s node] was 3.8%.
4.2 LOCATION OF SPINAL ANOMALIES

4.2.1 Regional location

The regional location of the spinal anomalies is depicted graphically in Figure 4.2. There were no thoracic congenital anomalies identified in this study. Most of the spinal anomalies were located in the lumbar region (12.1%), followed by the sacral region (10.8%) and then the thoracolumbar area (5.6%).

Figure 4.2 Number of cases of each category of anomaly
4.2.2 Location of individual congenital anomalies

The vertebral levels of SBO, facet tropism, hypoplastic TVP’s, Cupid’s bow sign and congenital Schmorl’s node are shown in Tables 4.2 and 4.3. The most common site identified for the location of SBO was S1 followed by L5. Facet tropism was identified mostly at L5 followed by L4. There was a single case of facet tropism at L2. Hypoplastic TVPs were identified at L1 and L4 levels. Cupid’s bow sign was identified in all levels of the lumbar spine except L1. Four cases of congenital Schmorl’s nodes were identified in the upper lumbar spine i.e. L1 and L2.

Table 4.2 Vertebral levels of SBO and facet tropism

<table>
<thead>
<tr>
<th>Vertebral level</th>
<th>SBO</th>
<th>Facet tropism</th>
</tr>
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<tbody>
<tr>
<td>T12</td>
<td>0</td>
<td>0</td>
</tr>
<tr>
<td>L1</td>
<td>0</td>
<td>0</td>
</tr>
<tr>
<td>L2</td>
<td>0</td>
<td>1*</td>
</tr>
<tr>
<td>L3</td>
<td>0</td>
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<tr>
<td>L4</td>
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<tr>
<td>L5</td>
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<td>S1</td>
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<td>S2</td>
<td>0</td>
<td>0</td>
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<tr>
<td>Total</td>
<td>30</td>
<td>100</td>
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</table>

* = L2-L3; # = L4-L5; ¥ = L5-S1

Table 4.3 Vertebral levels of other anomalies

<table>
<thead>
<tr>
<th>Type</th>
<th>Level</th>
<th>n</th>
<th>%</th>
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<tr>
<td>1. Hypoplastic TVP</td>
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<td>1</td>
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</tr>
<tr>
<td></td>
<td>L4</td>
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<td>50</td>
</tr>
<tr>
<td>Total</td>
<td></td>
<td>2</td>
<td>100</td>
</tr>
<tr>
<td>2. Cupid’s bow</td>
<td>L2</td>
<td>1</td>
<td>12.5</td>
</tr>
<tr>
<td></td>
<td>L3</td>
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<td>25</td>
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<tr>
<td></td>
<td>L4</td>
<td>3</td>
<td>37.5</td>
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<tr>
<td></td>
<td>L5</td>
<td>2</td>
<td>25</td>
</tr>
<tr>
<td>Total</td>
<td></td>
<td>8</td>
<td>100</td>
</tr>
<tr>
<td>3. Congenital Schmorl’s node</td>
<td>L1</td>
<td>2</td>
<td>50</td>
</tr>
<tr>
<td></td>
<td>L2</td>
<td>2</td>
<td>50</td>
</tr>
<tr>
<td>Total</td>
<td></td>
<td>4</td>
<td>100</td>
</tr>
<tr>
<td>4. Block vertebra</td>
<td>T12-L1</td>
<td>1</td>
<td>100</td>
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4.3 ASSOCIATION BETWEEN THE CONGENITAL LUMBAR SPINE OR LUMBOSACRAL ANOMALIES AND THE CLINICAL FEATURES

4.3.1 The association between congenital lumbar spine or lumbosacral anomalies and clinical features in general

The various clinical features identified in individuals with lumbar and/or lumbosacral anomalies are depicted graphically in Figure 4.3. The significant ($p < 0.05$) musculoskeletal clinical features identified were low back pain, scoliosis in the lumbar spine and decreased lordosis. Despite the high prevalence of radicular pain /leg pain and decreased ROM, these features were not statistically significant ($p > 0.05$) when compared to individuals with no congenital anomalies. Nonmusculoskeletal features
included dyspnea, lipoma, bowel abnormalities and bladder abnormalities. Figure 4.3 shows the prevalence of these clinical features in the 98 patients with anomalies.

Figure 4.3 Prevalence of clinical features in subjects with anomalies (n = 98)

* p < 0.05, Fischers exact test vs individuals with no anomalies

4.3.2 The association between individual congenital lumbar spine or lumbosacral anomalies and the presenting clinical features.

Only individual congenital spinal anomalies that were statistically associated with significant clinical features are shown in Table 4.4. Two significant clinical features that were associated with SBO were scoliosis and radicular/ leg pain. Individuals with thoracolumbar transitional vertebra were more likely to have decreased range of motion, scoliosis and decreased lumbar lordosis. A decreased lumbar lordosis was also significantly associated with complete lumbarisation compared to individuals with no spinal anomalies.

Table 4.4 The association between significant clinical features and individual anomalies

<table>
<thead>
<tr>
<th>Significant clinical features</th>
<th>SBO</th>
<th>Thoracolumbar transitional vertebra</th>
<th>Complete lumbarisation</th>
</tr>
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<tbody>
<tr>
<td>↓ ROM</td>
<td></td>
<td></td>
<td>p = 0.020*</td>
</tr>
<tr>
<td>Scoliosis</td>
<td>p = 0.017*</td>
<td></td>
<td>p = 0.012*</td>
</tr>
<tr>
<td>↓ Lordosis</td>
<td>p = 0.041*</td>
<td></td>
<td>p = 0.047*</td>
</tr>
<tr>
<td>Radicular /Leg pain</td>
<td>p = 0.028*</td>
<td></td>
<td></td>
</tr>
</tbody>
</table>

↓ = decreased, ROM = range of motion.
* p < 0.05, Fischers exact test vs individuals with no anomalies
4.4 RESEARCHER AND RADIOLOGIST SENSITIVITY

The results of the researcher and radiologist sensitivity are indicated in Table 4.5. Of the 98 anomalies, the researcher detected 92 and 51 were noted by the radiologist in the original radiologist report.

Table 4.5 Sensitivity of the researcher and the radiologist

<table>
<thead>
<tr>
<th></th>
<th>Sensitivity</th>
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<tbody>
<tr>
<td></td>
<td>Estimate</td>
<td>Mean %</td>
<td>% Range</td>
</tr>
<tr>
<td>Researcher</td>
<td>94</td>
<td>87 – 97</td>
<td></td>
</tr>
<tr>
<td>Radiologist</td>
<td>52</td>
<td>42 - 62</td>
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</table>
CHAPTER 5  
DISCUSSION OF RESULTS

5.1 RESEARCHER AND RADIOLOGIST SENSITIVITY
The data has been shown in Table 4.5. In this study, it appeared that the researcher had greater sensitivity when compared to the radiologists, on reporting congenital thoracic and lumbar spinal anomalies. However, the possible explanations for this include:

- The information requested on the request form by the student/intern or doctor may not have been related to a thoracic or lumbar spine congenital anomaly but rather to exclude other disorders of the thoracic or lumbar spine e.g. arthritic, infective, etc.
- The anomaly would have been most likely noted but not reported due to the possibility of it being considered clinically insignificant or an incidental finding.

It is worth noting that the researcher does not make any claims at being “more qualified” than a radiologist in the interpretation and reporting of x-rays.

5.2 THE PERIOD PREVALENCE OF CONGENITAL THORACIC AND LUMBAR SPINAL ANOMALIES
The data associated with the period prevalence of congenital thoracic and lumbar spinal anomalies was presented in Chapter Four. From the 372 radiographs that were included, 98 were positive for congenital thoracic or lumbar spine anomalies of any type, resulting in a total period prevalence of 26.3%. This may be assumed to be of significance as it is a large value, however, since it was obtained from a population formed by patients who were radiographed and managed at the Chiropractic Day Clinic at the DUT for the period 1 January 1997 to 31 December 2005, it is, therefore, not a reflection of the general population but more accurately associated to the population from which the sample was obtained. Since not all patients presenting at the Chiropractic Day Clinic are radiographed, it is not possible to state the true prevalence of the congenital thoracic or lumbar spine anomalies during the nine-year period.

5.2.1 Prevalence of individual congenital spinal anomalies
The majority of the subjects’ in the sample size presented with one anomaly, with a low prevalence for two or more coexisting (Table 4.1). One may extrapolate that the possibility of an individual having two or more lumbar spine anomalies is low. There were no thoracic anomalies (with the exception of thoracolumbar) identified in this study. It is, therefore, possible that the prevalence of anomalies in this region of the spine is very low.
With respect to the prevalence of the individual thoracolumbar, lumbar or lumbosacral anomalies:

**SBO:**
The overall prevalence of SBO in this study (Figure 4.1) was in keeping with the findings of Beck et al. (2004), but not with those of Esses (1995) and Treble et al. (2005) (Table 2.1). Although Treble et al. (2005) reported a high prevalence of SBO in individuals with isthmic spondylolisthesis, no such association was found in this study.

**LSTV:**
This was the most commonly identified anomaly in the selected sample with an overall prevalence of 11.9%. This finding lies within the range reported by Hughes and Saifuddin (2004) but is higher than that reported by Esses (1995), Beck et al. (2004) and Treble et al. (2005) (Table 2.1). The contribution of complete/partial lumbarisation/sacralisation to this overall prevalence is shown in Figure 4.1.

**Thoracolumbar transitional vertebra:**
Thoracolumbar transitional vertebra had a prevalence of 5.4% in the selected sample size. This prevalence, however, could not be compared to any data in the existing literature.

**Facet tropism:**
The prevalence of facet tropism in the selected sample size (Figure 4.1) was five-fold greater than that reported by Beck et al. (2004) (Table 2.1). Since the selected samples were similar in both studies (i.e. outpatients), it may, therefore, be possible that the prevalence of this condition may be higher than previously thought.

**Spina bifida vera:**
Spina bifida vera in the sacrum and lumbar spine had a prevalence of 0.5% and 0.3% respectively.

**Block vertebra:**
The prevalence of block vertebra in this study (Figure 4.1) was lower than that reported by Beck et al. (2004) (Table 2.1) indicating the rarity of this anomaly.

**Other anomalies:**
Hypoplastic TVP, clasp knife syndrome, congenital Schmorl’s node, pig snout vertebra and Cupid’s bow sign had a collective prevalence of 3.8%. No comparison could be made
with previous studies, as there was no available data in the literature with respect to these anomalies.

5.2.2 The prevalence of individual congenital spinal anomalies by location

SBO:
The majority of SBO lesions involved S1 followed by L5 (Table 4.2). Although this prevalence finding is much higher than that reported by Kamanli and Genc (2002) and Yochum and Rowe (2005) (Table 2.2), it does support their findings that the most likely location for SBO was S1 followed by L5.

Block vertebra:
There was one case of a thoracolumbar block vertebra. Taylor and Resnick (2000) had earlier reported that a possible site for this anomaly was the T12-L1 area.

Facet tropism:
This was found primarily in the L5-S1 level followed by L4-L5 level and there was one case at the L2-L3 level (Table 4.2). This finding is in keeping with those of Taylor and Resnick (2000) and Yochum and Rowe (2005).

Cupid’s bow sign:
This anomaly was identified at all lumbar vertebral levels except L1 (Table 4.3). The majority of the lesions were identified at the L4 level and this supported the findings of Taylor and Resnick (2000).

Congenital Schmorl’s node and Hypoplastic TVP:
There were four cases of congenital Schmorl’s node with two cases identified at the L1 and L2 levels respectively (Table 4.3). There was one case of hypoplastic TVP identified at the L1 and L4 levels respectively. No comparison could be made with previous studies, as there was no available data in the literature with respect to these anomalies.

5.3 ASSOCIATION BETWEEN THE CONGENITAL THORACIC AND LUMBAR SPINE ANOMALIES AND THE CLINICAL FEATURES

5.3.1 Association between congenital thoracic and lumbar spine anomalies and the clinical features in general

A comparison was done to determine any statistical significance in clinical features between the group that was positive for a congenital thoracic or lumbar spine anomaly
and the control group, which had no congenital spinal anomalies. By far the most common feature was low back pain ($p = 0.003$; **Figure 4.3**). This was followed by radicular/leg pain and decreased range of motion. These were, however, not significantly associated with the presence of any anomalies ($p > 0.05$). Scoliosis of the lumbar spine and decreased lordosis were more likely to found in those with anomalies ($p = 0.006$; $p = 0.005$). The clinical implication of this finding is that a congenital spinal anomaly should be considered as a differential diagnosis in a patient presenting with low back pain, scoliosis and decreased lordosis. Mild scoliosis and/or decreased lordosis may, however, not be clinically apparent in certain individuals e.g. obese patients. As a result, the physician may not be aware that a congenital lumbar spinal anomaly could be the cause of the LBP.

5.3.2 Association between individual congenital thoracic or lumbar spine anomalies and clinical features

**SBO:**
Scoliosis and radicular/leg pain were the two clinical features that were significantly associated with SBO (**Table 4.4**). This is similar to the findings of Hodges and Peck (1937), Winter *et al.* (1968) and Yochum and Rowe (2005). Spinal cord and/or nerve root lesions have been identified in individuals with SBO (Berkow *et al.*, 1997). This may lead to muscle weakness in some or all areas below the level of the lesion/s resulting in scoliosis (Esses, 1995). The defect in the posterior neural protective bony arch found in SBO may lead to a loss of mechanical stability resulting in an increased rate of IVD degeneration and the subsequent development of radicular/leg pain (Hodges and Peck, 1937).

**LSTV:**
The only clinical feature that was significantly associated with LSTV, more specifically complete lumbarisation, was a decreased lordosis. This finding is contrary to those of Hodges and Peck (1937), Tilley (1970), Paillas *et al.* (1977) and Yochum and Rowe (2005). Although no skeletal features have been definitively associated with LSTV, a decreased lordosis may occur as a result of altered biomechanics of the spine associated with this condition (Tini *et al.*, 1977).

**Thoracolumbar transitional vertebra:**
There was a significant association between decreased ROM, scoliosis and decreased lordosis with thoracolumbar transitional vertebra (**Table 4.4**). Patients were more likely to have these symptoms if they had thoracolumbar transitional vertebra. These features could be accounted for by variations in facet joint articulations at the thoracolumbar
junctions (Singer et al., 1988; Pal and Routal, 1999). No other literature could be found on the possible association between these findings and thoracolumbar transitional vertebra.

**Facet tropism**

Although there were reports of LBP and/or radicular/leg pain in individuals with this anomaly (data not reported), there was no significant association ($p > 0.05$) between facet tropism and any clinical feature. Earlier, Farfan and Sullivan (1967) reported that there was a likelihood of LBP and sciatica in individuals with facet tropism (Table 2.2). These features were attributed to a decreased ability of the facet joint to resist rotational forces and annular degeneration of the IVD leading to disc herniation.

**Spina bifida vera, block vertebra, pig snout vertebra, Cupid’s bow sign, clasp knife syndrome, hypoplastic TVP, and congenital Schmorl’s node:**

There were no specific clinical features that were significantly associated with these anomalies ($p > 0.05$). There was one case of a lipoma that was found in an individual with spina bifida vera. This feature was reported earlier by Drolet (2004) and Nadkarni et al. (2005) (Table 2.4).

The clinical features of decreased ROM, decreased lordosis, radicular pain and scoliosis (unless severe or to evaluate progression) do not necessarily warrant a radiographic examination. Often a clinical diagnosis is made and conservative treatment (manual and/or pharmacological) protocols may be instituted. A radiograph of the thoracic or lumbar spine may be taken if patient does not respond to such treatment and a congenital anomaly of the spine may be identified as an incidental finding or the cause of the patient’s symptoms and signs. Some congenital anomalies appear to be clinically silent (e.g. pig snout vertebrae, Cupid’s bow sign and hypoplastic TVPs) and may be identified as incidental findings on radiographs. The findings of this study indicate that these anomalies may not have any clinical significance.

The differences in the findings of this study and those reported in the literature may be attributed to variations in the population sample.
CHAPTER 6
CONCLUSION AND RECOMMENDATIONS

6.1 CONCLUSIONS

The primary aim of this study was to determine the period prevalence (1 January 1997 to 31 December 2005) of congenital thoracic and lumbar spine anomalies and their association, if any, with the subjects presenting clinical features. With regard to the primary aim of the study, from the 372 radiographs that were included, 98 were positive for congenital thoracic or lumbar spine anomalies of any type, resulting in a total period prevalence of 26.3%.

In terms of the specific objectives and associated hypothesis set at the onset of the study:

- The Null Hypothesis ($H_0$) that stated that there shall not be a significant number of congenital thoracic or lumbar spine anomalies present for the chosen period of 1 January 1997 to 31 December 2005 was partially accepted as no thoracic congenital anomalies were identified in this study while the prevalence finding of congenital anomalies of the lumbar spine was ambivalent.

- The Null Hypothesis ($H_0$) that stated that there shall be no significant association between the congenital thoracic or lumbar spine anomalies and the presenting clinical features in general was also partially accepted. No congenital thoracic anomalies were identified while there was a significant association between congenital lumbar spinal anomalies in general and LBP, scoliosis and decreased lordosis.

- The Alternate Hypothesis ($H_a$) that stated that there shall be a significant association between the individual congenital thoracic or lumbar spine anomalies and the presenting clinical features was partially accepted with respect to SBO, LSTV and thoracolumbar transitional vertebra.

- The Null Hypothesis ($H_0$) that stated that there shall be little/no significant association between the subjects presenting clinical features with reported clinical features associated with thoracic or lumbar spinal congenital anomalies from the literature was partially accepted with respect to scoliosis and radicular/leg pain that were found to be significantly associated with SBO.
6.2 RECOMMENDATIONS

Recommendations for future studies include the following possible investigations:

- The inclusion of demographics such as age and sex of the subjects so that any association between these factors and congenital anomalies of the spine could be determined.
- A similar study should be conducted in a state hospital/s so that a larger sample size and wider variation in the population may be obtained.
- A study to determine the management principles of (both conservative and surgical) of congenital spinal anomalies should be conducted to enable spinal health care professionals to become aware of the appropriate patient management principles.
REFERENCES


Appendix 1

DATA COLLECTION SHEET 1

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<tr>
<th>CODE</th>
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<th>INITIAL PRESENTING COMPLAINT</th>
<th>CLINICAL FEATURES RELATED TO THE ANOMALY</th>
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</thead>
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Appendix 2
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<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td>RESEARCHER</td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td>RADIOLOGIST</td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td>BOTH</td>
</tr>
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