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THE EFFECT OF LUMBOSACRAL TRANSITIONAL VERTEBRAE ON THE FIFTH LUMBAR VERTEBRA AND FIRST SACRAL BODY AND ITS PREVELANCE IN THE OSTEOLOGICAL COLLECTION OF THE UNIVERSITY OF TEXAS AT ARLINGTON

Stephanie Dolenz

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by

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ABSTRACT

THE EFFECT OF LUMBOSACRAL TRANSITIONAL VERTEBRAE ON THE FIFTH LUMBAR VERTEBRA AND FIRST SACRAL BODY AND ITS PREVALENCE IN THE OSTEOLOGICAL COLLECTION OF THE UNIVERSITY OF TEXAS AT ARLINGTON

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Lumbosacral transitional vertebrae, or LSTV, is generally considered to be a congenital anomaly affecting the fifth lumbar vertebra and first sacral body. It is presented as sacralization, characterized by a caudal shift of fifth lumbar vertebra, and lumbarization, characterized by a cranial shift of the first sacral body. This research focused on the prevalence of LSTV in the osteological collection (overall and by sex), on nonmetric observations to describe the anomaly, and on metric observations to observe the effect of LSTV on bone. It was found that LSTV occurs in 20% of the sample. In addition, LSTV is more common in females and sacralization is more common than lumbarization. The areas affected by LSTV on the fifth lumbar vertebrae and first sacral bodies showed metric differences when compared to the control sample; however, there is not a distinguishable

pattern. Overall, it appears that LSTV is highly variable in its expression on bone.

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CHAPTER 1

INTRODUCTION

Lumbosacral transitional vertebrae, or LSTV, are generally considered to be congenital anomalies affecting the L5 vertebra and the S1 vertebra of the sacrum, presented as sacralization or lumbarization. Sacralization refers to the caudal shift of the fifth lumbar vertebra, in which the L5 may fully or partially fuse with the first sacral vertebra and develops sacral characteristics. Lumbarization refers to cranial shift of the first sacral vertebra, in which it does not fuse to the second sacral vertebra and rather acts as a sixth lumbar vertebra. LSTV are caused by shifts to the L5 and S1 borders during development, most likely due to the mutation or overexpression of *Hox* genes (Adibatti & Asha, 2015; Carapuço et al., 2005). The occurrence of this anomaly begins in the early stages of embryonic development.

Because this congenital anomaly is not apparent or particularly incapacitating, it is likely that there never existed biological pressures to select against it. Therefore it has become curiously common in the general population, with a frequency of around 4-30% (Apazidis et al., 2011; Konin & Walz, 2010). Due to the relatively common presence of LSTV in the general human population, it seems likely that this anomaly is not a pathology, but rather a product of human variation. Furthermore, it seems possible that this variation may be present in our deep ancestral past. Paleoanthropologists have suggested that a certain specimen of *Australopithecus africanus,* STS 14, dating around 2.5 million years ago, has a sixth lumbar vertebra (Haeusler, et al., 2002). Through analysis of the 2.5 million paleoanthropological fossil record, it is postulated that most early hominids typically had five lumbar vertebrae, meaning that this individual had a transitional vertebra. Regardless, it is evident that lumbarization of the S1 vertebra and sacralization of the L5 vertebra, or LSTV, is quite common in the general population as well as in the archaeological record.

While LSTV are fairly common in the general population, there are discrepancies in relating factors such as biological sex and ancestry to this anomaly. Due to the congenital nature of LSTV, it seems likely that biological sex and ancestry may affect the prevalence of this anomaly, but that has not been sufficiently proven by current research. Overall, it seems that the most contentious subject surrounding LSTV research is the possible correlation between this anomaly and lower back pain. A large number of researchers suggest that the presence of LSTV can be linked to some cases of lower back pain, termed Bertolotti Syndrome (Eyo et al., 2001; Jancuska et al., 2015; Konin & Walz, 2010). However, it is not certain whether the relationship between these two factors is causal or coincidental.

Although it is strongly debated whether LSTV are responsible for lower back pain, this anomaly can create problems for affected individuals and those treating them. The most pressing issue in regard to the identification of LSTV in individuals is the possibility of surgical error due to spinal surgery occurring at incorrect levels of the spine (French et al., 2014). This can occur due to improper numeric identification of the vertebrae and result in complications or the need for a second surgical procedure. In addition, the failed assessment of LSTV can lead to an incorrect sex assignment of the sacrum or diagnosis of spinal pathologies in a bioarchaeological context. Although incorrect assignment of sex is not as distressing as surgical errors, correctly identifying LSTV in both an osteological and clinical setting is highly important.

This study attempts to address the classification of LSTV, the prevalence of this anomaly in males and females, and the effect of this anomaly on the metric dimensions of the L5 and S1 vertebrae. This research has three main components. First, the research will examine the prevalence of this anomaly in the total sample as well as separately in both males and females. Second, a qualitative analysis of the affected specimens at the lumbosacral region will serve to classify the specimens by Castellvi type. Third, a metric assessment of both the affected and unaffected specimens will allow for the direct comparison of vertebral dimensions. This will provide a better understanding of how LSTV affect the morphology of L5 and S1 vertebrae. Findings will be integrated with existing research. Overall, by collecting data on the skeletal collection at the University of Texas at Arlington, a better understanding of this anomaly can be reached.

CHAPTER 2

LITERATURE REVIEW

2.1 Lumbar and Sacral Development

In order to understand how this anomaly is presented on the L5 and S1 vertebrae, it is important to understand the development of both the lumbar and sacral vertebrae. Lumbar vertebrae typically ossify at three primary centers (the centrum and the left and right vertebral arches) as well as at secondary centers at the tips of the left and right transverse processes and the spine (White et al., 2012). The secondary ossification centers for the lumbar vertebrae typically appear during puberty and fuse between 25 and 30 years of age. Additionally, the vertebrae, as well as the intervertebral discs, ligaments, and periosteal coverings, are mesodermal in their developmental originations (O'Rahilly & Meyer, 1979). These features of the spinal column begin development in the early embryonic stages; the vertebrae continue to ossify and fuse during postnatal maturation into adulthood.

In the sacrum, there are between 58 and 60 ossification centers: 5 in the centra, 10 in the posterior neural arches, 8 in the costal processes, 10 at the vertebral endplates, about 8 to 10 in the transverse processes, 8 in the anterior costal epiphyses, 4 in the posterior epiphyses, 3 in the spinous processes, and 2 in the mammillary processes (Broome et al., 1998). Of course, this is assuming that the individual has 5 sacral vertebrae. However, if the sacrum features a sixth vertebra, then it is likely that L5 fusedto the sacrum. Regardless, these ossification centers begin fusing as a fetus and continue to do so during postnatal maturation. The primary ossification centers generally fuse before the age of 7 (Broome et al., 1998). Moreover, the sacral vertebrae continue to fuse up until the final fusion of the S1 and S2 at around 25 years of age, with the possible fusion of the S5 and coccygeal vertebrae occurring as late as 27 years of age (Broome et al., 1998). In addition, it has been suggested that the fusion timing is sexually dimorphic in males and females, with females typically fusing earlier than males, due to an association with childbearing (Ríos et al., 2008). However, typically most of the fusion of the sacrum occurs before puberty in both males and females.

Hox genes are integral to the development of the lumbar and sacral vertebrae. In particular, *Hox* group 11 genes are essential for the development of sacral and lumbar vertebrae (Carapuço, et al., 2005). These expressions initially occur in the presomitic mesoderm phase of fetal development. In studies of *Hoxa11*, it was observed that overexpression of this gene led to sacralization and, in addition, anteriorly projecting protuberances were likely the result of a residual expression of *Hoxa11* (Carapuço et al., 2005). Also, it was found that overexpression of this gene led to a caudal shift in the lumbar vertebrae (Carapuço et al., 2005). *Hox* group 11 genes are not only significant in the development of the sacrum and vertebral column, but also in part responsible for anomalies affecting these regions such as LSTV. This further supports that this anomaly is congenital in nature, and provides a cause for the presence of lumbarization or sacralization in an individual.

2.2 Prevalence in Populations

Generally, LSTV is presented as either lumbarization of the S1 or sacralization of the L5 vertebra. Though these two classifications are used to describe LSTV, expression on the lumbar vertebrae and sacrum is rather variable. Sacralization occurs when the transverse process of the L5 overgrows and fuses to the S1, which can appear either bilaterally or unilaterally, and lumbarization occurs when the S1 does not fully fuse to the S2, morphologically presenting itself as a sixth lumbar vertebra. LSTV are highly prevalent in the general population, with 4-30% of individuals showing some degree of LSTV (Apazidis et al., 2011; Konin & Walz, 2010). The wide range reported is most likely due to differing degrees of prevalence between certain populations.

In attempts to understand the presence of this anomaly in the general population, previous studies have been carried out on whether the ancestry of an individual is correlated with the presence of LSTV. In one study, Savage found no significant difference of the presence of sacralization or lumbarization when comparing ancestry within the skeletal sample (Savage, 2005). Furthermore, another researcher determined that there was no significant difference observed by ancestry in clinical patients with LSTV (Nardo et al., 2012). As previously stated, LSTV are highly prevalent in the general population; however, it is suggested that this anomaly does not have a higher frequency in certain ancestral populations. Although more research is necessary in understanding this aspect of LSTV, especially due to its genetic origins, this suggests that this anomaly is more indicative of general human variation rather than population-specific variation. Unfortunately, the research undertaken in this paper is unable to explore this facet of LSTV research due to the lack of biographical information of the sample. Therefore, the focus of this research will be placed upon biological sex.

It has been suggested that lumbarization is more prevalent in females, whereas sacralization is more prevalent in males (Eyo et al., 2001; Mahato, 2011). This could in part be due to the congenital nature of the anomaly, pertaining to the expression of *Hox* genes. Furthermore, it has been proposed that gene expression is more variable in males (Itoh & Arnold, 2015). This could explain the higher rate of sacralization, or the caudal shift in the lumbar vertebrae, being more prevalent in males. Perhaps the *Hox 11* genes are more likely to overexpress in males, resulting in sacralization. However, that does not account for the possible higher prevalence of lumbarization in females. More research is needed to fully understand the effect of the *Hox* genes and biological sex on the presence of LSTV in individuals, as well as the generalization of variation in male gene expression, meaning that perhaps only certain genes are more variably expressed in males.

In analyzing the anomaly in general, one clinical study suggested that females had a higher prevalence, about 1.3 times, of LSTV compared to males (Sekharappa et al., 2014). However, an osteological study showed no statistically significant correlation between these LSTV and biological sex (Savage, 2005). This discrepancy opens debate about whether lumbarization and sacralization, or perhaps LSTV in general, are indeed more prevalent in a particular biological sex. Although it seems likely that biological sex could have an effect on the presence of LSTV due to its genetic origin, more statistical analysis of this research could elucidate the inconsistency of results in these studies.

2.3 Previous Clinical Research

The presence of LSTV has been linked to lower back pain, termed Bertolotti Syndrome (Eyo et al., 2001; Jancuska et al., 2015; Konin & Walz, 2010). This means that this anomaly could directly impact the biomechanics of the individual. Although lower back pain can occur from alternative sources even when LSTV is present, it is suggested that the pain originates at the pseudoarticulation of the transverse process(es) and sacrum or the osseous fusions (Jancuska et al., 2015). The pseudoarticulation creates a joint surface, which can be further subjected to degenerative conditions known for pain or discomfort such as arthritis. These morphological anomalies could be the source of lower back pain in individuals with LSTV, with the possibility that level of fusion or pseudoarticulation may correlate with the degree of pain. Furthermore, it has been suggested that of patients suffering from lower back pain in the lumbar region, around 23.6% also had a LSTV (Uçar et al., 2012). This shows a possible correlation between lower back pain and this anomaly in a clinical setting. Lower back pain is significant in not only modern populations, but also past populations, in determining quality of life and possible workload of an individual.

Another concern with the presence of LSTV is its effect on disc degeneration or herniation. Through the analysis of radiographic and x-ray imaging, it has been suggested that this anomaly is common in symptomatic patients suffering from disc degeneration (Apazidis et al., 2011; French et al., 2014). This may, in part, also be a factor in the individual's lower back pain. Furthermore, it is postulated that the presence of LSTV decreases the height of the intervertebral disc of that correlating segment, especially in sacralized cases where the transverse process fused to the sacrum (Hsieh et al., 2000). Due to the border shift of LSTV, it is possible for the intervertebral discs to undergo degeneration or displacement. In addition to pseudoarticulation joint surfaces that can be subjected to arthritic changes, this anomaly also affects the cushioning between the vertebrae, which may also cause lower back pain.

Apart from lower back pain, another topic of interest in LSTV research is the effect of this anomaly on biomechanics of the individual. In order to understand how this anomaly affects the individual, it is beneficial to consider lumbarization and sacralization separately. Lumbarization of the first sacral vertebrae is characterized by the lack of fusion of the S1 to the S2, in which the S1 assumes the characteristics of a sixth lumbar vertebra. It has been suggested that the presence of a sixth lumbar vertebra can actually be quite favorable in bipedalism, creating a more elongated spine; this not only increases lumbar lordosis, but can also reduce the vertical displacement of the center of gravity during walking as well as the stress placed upon the intervertebral discs (Haeusler et al., 2002). Although the difference between five and six lumbar vertebrae is not drastic, it seems that the lumbarization does not have any debilitating effects and may be partially advantageous.

In analyzing the effect of sacralization on biomechanics, it seems evident that this anomaly may cause discomfort. Although the connection to lower back pain is contentious, due to the pseudoarticulations and possible disc degeneration, it seems possible that sacralization of the L5 to the sacrum may cause lower back pain. The presence of lower back pain could certainly affect the quality of life of the individual; however, it is suggested that due to the load-bearing function of the sacrum, a sacralized L5 may actually be beneficial. The size and surface area of the first sacral vertebrae is essential in the sacrum's ability to handle the weight of the upper body. Due to the decrease in the height of sacra (S1-S5) with LSTV, it is suggested that the smaller sacrum may incorporate the L5, through sacralization, to enhance the load-bearing capacity of the first sacral vertebrae, which in these cases has an insufficient joint surface (Jancuska et al., 2015). Although this postulation provides a clear interpretation for the advantage of sacralization, it does not address the timing of development of LSTV. It is possible LSTV develop to address a biological issue relating to load-bearing capacity of the sacrum, but it is also possible that this anomaly develops prior to load bearing, creating the issue of a smaller and insufficient S1 joint surface. More research is needed on the timing of development of this anomaly to fully understand the possible advantage of sacralization.

2.4 Previous Osteological Research

In describing the presence of LSTV, nonmetric methods classify this anomaly into four types. Type I includes a unilaterally or bilaterally enlarged transverse process; type II includes a unilateral or bilateral pseudoarticulation of the enlarged transverse process and sacrum; type III includes a bilateral or unilateral complete osseous fusion of the transverse process and sacrum; and type IV includes a type II unilateral transition with a type III fusion on the opposing side (Jancuska et al., 2015; Konin & Walz, 2010). As shown below in Figure 2.1, these nonmetric classifications, termed the Castellvi Classification, describe how LSTV are presented in the L5 and sacrum. This classification system not only describes the variable ways this anomaly can be presented, but also acknowledges the extreme variability of LSTV. This anomaly can be presented unilaterally or bilaterally, and the degree of fusion and contact is also highly variable. In addition, the nonmetric method used to describe LSTV is important in understanding how this anomaly can affect the joints between the lumbar and sacral vertebrae.

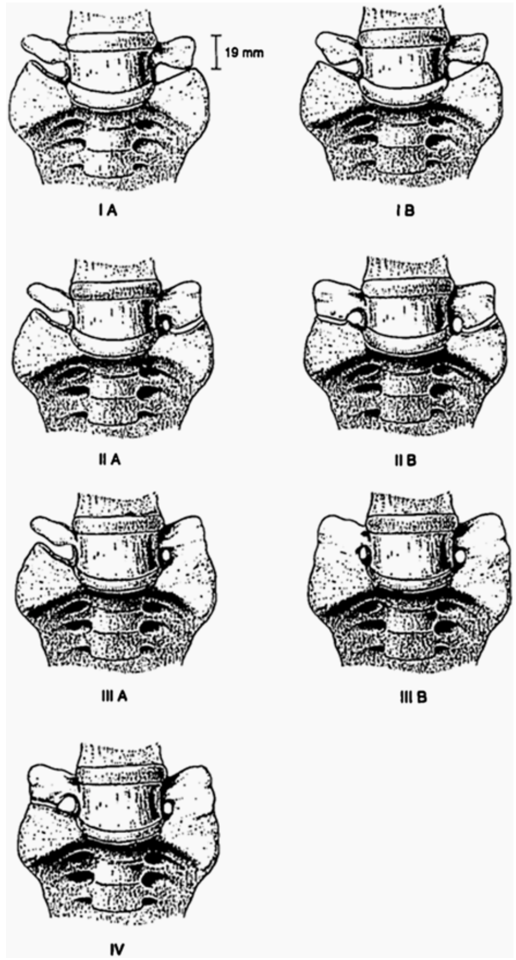

Figure 2.1: Castellvi Classification of LSTV Illustration (Apazidis et al., 2011)

Although the nonmetric observations are significant for studying LSTV, metric methods are equally important. Measurements of the structures in the lumbar and sacral region are a direct way to observe the effect of this anomaly on the L5 and S1 vertebrae. Through previous studies, osteologists have attempted to measure the effect of LSTV on lumbar vertebrae and sacra. Measurements of the pedicle width, body height, and canal width of lumbar vertebrae suggested that the body height of the L5 vertebrae was more asymmetrical in specimens with LSTV, and in sacralized specimens the L5 assumed more sacral characteristics, including a wider spinal canal (Savage, 2005). Overall, the results of that study were quite inconsistent, reflecting a uniqueness of each LSTV case. Although these features metrically differed from unaffected specimens, the differences were not consistent enough to develop a pattern, suggesting that perhaps this anomaly is individually variable.

In addition, measurements of the sacrum of sacralized and unaffected individuals suggest that the overall length of the sacrum is shortened in individuals with a fused L5 (not including the L5 in measurements) (Jancuska et al., 2015; Mahato, 2010a). In addition, there is a notable change in distance between facets, and occasionally the auricular surface is situated lower (Mahato, 2010a). This could mean a widening or narrowing of the spinal canal of the sacrum as well as the adjustment of the auricular surface position to accommodate the transitional vertebra. Also, measurements of variation in individuals with and without lumbarization show no significant difference (Mahato, 2010b). Although these anomalies affect the bone, it is generally inconsistent for each specimen. This indicates a variable expression and uniqueness in the presentation of sacralization and lumbarization, or LSTV, on the L5 and S1.

CHAPTER 3

METHODOLOGY

This research project is structured to address three main components of LSTV. First, the prevalence of LSTV in the overall collection, as well as by sex, was calculated. Second, nonmetric characters were observed for specimens with this congenital anomaly. Third, measurements were taken to determine the effect of LSTV on bone.

3.1 Specimens and Samples

In discussing the methodology of this research, it seems important to note that the osteological collection at the University of Texas at Arlington was donated from a medical supply company. The specimens within the collection have no biographical information and are littered with drill and cut marks. Therefore, all of the biographical information was deduced using sex assignment methods. Moreover, these individuals are composite in nature, meaning that articulated specimens may not necessarily have belonged to the same individual during life. However, an effort was made to ensure that the articular facets of the lumbar vertebrae and sacrum connected, providing some evidence that the articulated vertebral columns used matched the sacra by which they were accompanied. While some specimens seemed to belong to differing individuals in life, it is doubtful that this invalidates the results of this study; however, it seems necessary to acknowledge these possible concerns relating to the collection. This research focused on the presence of LSTV within the collection. Overall, 54 specimens were observed and measured to determine the effects of this anomaly on bone as well as to find a possible association between LSTV and biological sex. The total of 54 is comprised of 42 articulated vertebral columns and sacra, eight isolated sacra, and four isolated fifth lumbar vertebrae. The sample of LSTV specimens was chosen based upon the absence or presence of certain characteristics of this anomaly including a fused L5, an overgrown transverse process of the L5 creating a pseudoarticulation with the S1, and the presence of a sixth lumbar. This allowed for the grouping of the specimens into two samples, those with LSTV and those without. The sacra and fifth lumbar vertebrae without LSTV within the collection were used as control sample. It is important to note that specimens within the control sample do have pathologies, including osteoporosis and osteoarthritis; however, this should not affect the findings of this research. The presence of the control sample allows for the direct comparison of measurements, mentioned below, with the sample with LSTV.

3.2 Sample Observations

In the first phase of this research, the prevalence of LSTV was determined in the overall collection and separately by sex. Percentages were established for the overall prevalence of specimens with and without LSTV within the collection. These percentages were then compared to determine the prevalence of LSTV within the collection. In addition, the prevalence of sacralization and lumbarization was also separately calculated in the overall sample using percentages.

Sex was assigned based on standard calculations for the specimens within this collection. Measurements of the sacra included the anterior length of the sacrum (Figure A.A.1), the superior breadth of the sacrum (Figure A.A.2), the width of S1 (Figure A.A.3), and the anterior-posterior length of S1 (Figure A.A.4).

These measurements, were then expressed in two ratios of sacral index:

(breadth / length) * 100

and

(ant. –post. S1 length / S1 width) * 100

These measurements and indices were then compared to those of the Yadav et al. (2015) study on the determination of sex using sacra. Once sex was assigned, the prevalence of LSTV was calculated by sex using percentages. These percentages were then compared (focusing on the occurrence of LSTV, sacralization, and lumbarization by sex) to determine a possible association between biological sex and LSTV. Sex was not assigned to isolated fifth lumbar specimens.

3.3 Nonmetric Observations

For the morphological portion of research, nonmetric and metric methods were used to measure and discuss the extent of LSTV using methods in previous studies (Konin & Walz, 2010; Mahato, 2010a; Savage, 2005). First, nonmetric variables were observed and recorded on the specimens with LSTV and described using the Castellvi classification system (Jancuska et al., 2015; Konin & Walz, 2010). The sample with LSTV was then grouped by the presence of sacralization and lumbarization. Using the Castellvi classificiation system, the specimens with sacralization were categorized as type Ia, Ib, IIa, IIb, IIIa, IIIb, and IV as seen in Figure 2.1. In specimens with sacralization, recorded observations included whether the sacralization of the specimen was presented unilaterally or bilaterally. Additionally, it was noted which side of the specimen was affected, as well as the degree of connection, described as contact, partial ossification, and complete ossification. In cases of lumbarization, the presence of a sixth lumbar vertebra and only four sacral bodies was used to characterize the anomaly. These nonmetric observations were then compared throughout the specimens with LSTV to determine the possible variation of its expression within the collection.

3.4 Metric Observations

In the final phase of the research, metric observations were used to determine the possible effect of LSTV on bone. The instrument used for measurements was a standard sliding caliper, with 0.5 precision, and each measurement was taken three times. The average of the measurements was calculated and used for the sex assignments and morphological comparisons. The specific points of measurements taken on the sacra and fifth lumbar vertebrae can be viewed in Appendix A; the averages of the values for each measurement can be found in Appendix B. Measurements were taken on the L5 including the width of the L5 spinal canal (Figure A.A.5), the width of the right and left pedicles (Figure A.A.6), and the height of the right and left bodies (Figure A.A.7). Measurements were also taken on the sacrum, including the anterior length of the S1 (without the L5 in cases of fusion; Figure A.A.1) and the width of the sacral canal, or distance between the articular facets of S1 (Figure A.A.8).

The measurements of the individuals with LSTV were compared to measurements taken on the control specimens, or specimens with an absence of LSTV, to determine the degree of variation in these features due to the presence of lumbosacral transitional vertebrae. Also, the measurements of the LSTV specimens were compared with each other to determine variation within the expression of lumbosacral transitional vertebrae. In comparing the asymmetry of right and left pedicle width, differences above 1 mm were considered notable. In comparing the asymmetry of the right and left body heights, differences above 1.7 mm were considered notable. Differences in canal widths of the fifth lumbar vertebra and first sacral body were not considered notable unless greater than 3 mm. These values were determined by comparing the control sample and affected sample. In the control sample, few specimens exceeded these values, and the most of the affected specimens did.

CHAPTER 4

RESULTS

4.1 Sample Observations

The overall sample consists of 54 specimens including 42 articulated vertebral columns and sacra, eight isolated sacra, and four isolated fifth lumbar vertebrae. Of the 54 specimens, 11 have LSTV. This means that 20% of the specimens within the collection have LSTV, as shown below in Figure 4.1. Furthermore, of the 11 specimens with LSTV, nine have characteristics of sacralization and two have characteristics of lumbarization. This means that 82% of the specimens with LSTV have sacralization and have 18% lumbarization, as shown below in Figure 4.2. To distinguish the fully fused sacralized specimens from lumbarization, the presence of four lumbar vertebrae and six sacral bodies was noted. Conversely, the presence of five lumbar vertebrae and five sacral bodies was used to distinguish partial lumbarization from fully fused sacralization.

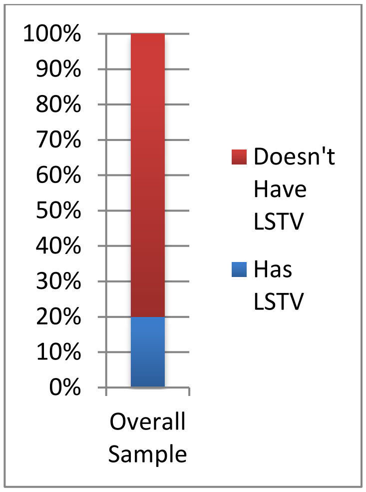

Figure 4.1: Percentage of Sample With and Without LSTV

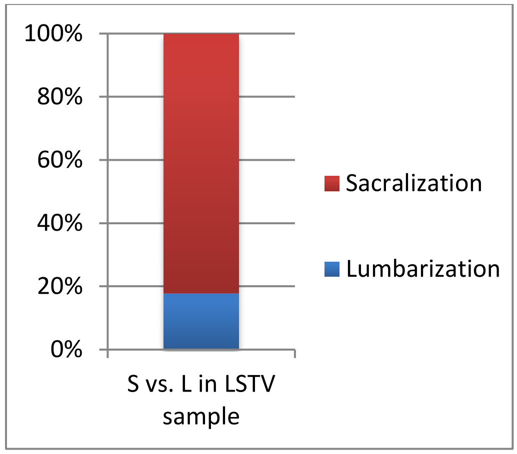

Figure 4.2: Percentage of Sacralization vs. Lumbarization

Of the 54 specimens in this study, biological sex was assigned to 50 using the sacral indices and measurements mentioned in the methodology section. The other four specimens could not be assessed because they are isolated vertebrae, lacking a sacrum. Of these 50, 18 (36%) of the specimens were assigned to “male” and 32 (64%) were assigned to “female”. In the sample with LSTV, seven are female and four are male. This means that of the 11 specimens with LSTV, about 64% are female and 36% are male, as shown below in Figure 4.3. In addition, five of the female specimens and four of the males showed characteristics of sacralization, meaning that of the nine specimens with sacralization, 56% are female and 44% are male. Furthermore, the two specimens with characteristics of lumbarization are both female, as shown below in Figure 4.4. This means that 100% of the specimens with lumbarization are female.

<u>4.2 Nonmetric Observations</u>

The presence of five lumbar vertebrae and five sacral bodies was used to distinguish partial lumbarization from fully fused sacralization. Conversely, to distinguish the fully fused sacralized specimens from lumbarization, the presence of four lumbar vertebrae and six sacral bodies was noted. Photographs of the specimens with LSTV are available in the appendix for convenience and clarification.

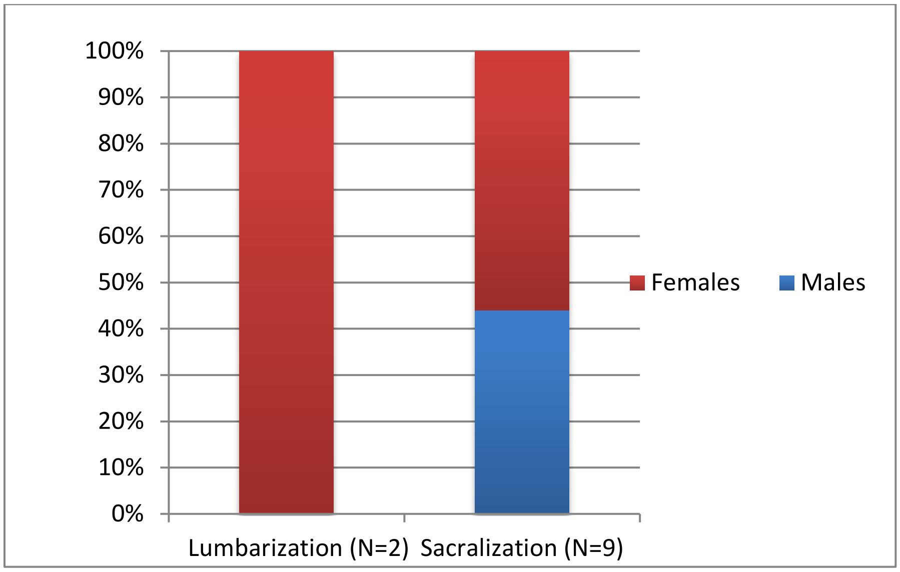

Figure 4.3: Percentage of Females and Males With LSTV

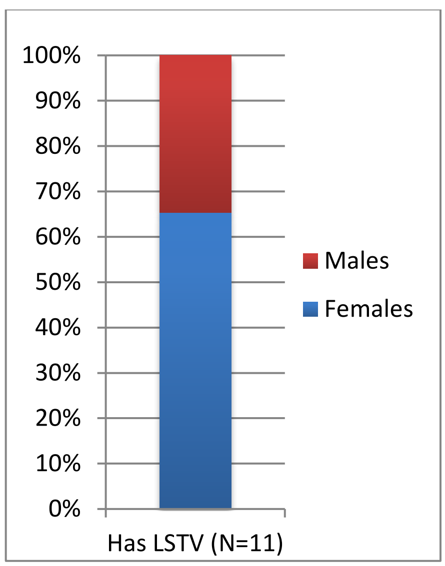

Figure 4.4: Lumbarization and Sacralization Prevalence in Males and Females

Of the 11 specimens with LSTV, nine exhibit characteristics of sacralization, with the transverse process(es) either fully or partially fused to the sacral ala(e). The sacralized specimens can be described using the Castellvi system. Two (P1643 and P1616) are type Ia, with a slightly overgrown transverse process of the fifth lumbar vertebra making contact with the first sacral body. P1643 is affected on the left side, whereas P1616 is affected on the right. Five specimens (CI10, CI13, P1373, P1594, and P2048) are type IIa, meaning there is a pseudoarticulation between the transverse processes of the fifth lumbar and the first sacral body. Of these specimens, two (CI10 and P1594) are affected on the right and three (CI13, P1373, and P2048) are affected on the left transverse process. In addition, two specimens (CI2 and P1912) are type IIIb, which is characterized by fusion of the transverse processes of the fifth lumbar vertebra to the first sacral body on both sides.

In observing the 11 LSTV specimens, it was determined that two (P1400 and CI3) have characteristics of lumbarization. In both, the lumbarization is only partial. P1400 has five lumbar vertebrae and five sacral bodies. In addition, the area between the first and second sacral bodies is not fused anteriorly or posteriorly, and the left ala is not fused. CI3 also has five lumbar vertebrae and five sacral bodies. The space between the first and second sacral bodies is not fused anteriorly or posteriorly, and ossified ligament is visible in the cavity.

4.3 Metric Observations

To determine the effect of LSTV on fifth lumbar vertebrae, the right and left pedicle widths and the right and left body heights were measured. When observing right and left pedicle widths, the affected side is larger. In four specimens (P2048, P1643, CI13, and P1373) the left pedicle is wider. In three specimens (P1616, P1594, and CI10) the right pedicle is wider. In both P1912 and CI2, the specimens with fully fused sacralization, the left pedicle is wider, although CI2 is only asymmetrical by 1 mm. One specimen with characteristics of lumbarization (CI3) does not have pedicle width asymmetry, whereas the other lumbarized specimen (P1400) has a slight asymmetry of 1 mm. However, the metric difference in the widths is variable among the specimens with LSTV, as shown in Table 4.1 below. The pedicle width asymmetry (>1 mm) ranges from absolute values 3 mm-6 mm, with an average of ca. 4.3 mm in the six sacralized specimens displaying substantial asymmetry. Including the three specimens with a width asymmetry of 1 mm, the average declines to 3.2 mm. In the control sample, there are some specimens with a slight difference in pedicle width (between 1 mm-1.7 mm); however none have an asymmetry greater than 1.7 mm.

In addition to pedicle width, the body height is asymmetrical in some of the specimens with LSTV. Five specimens (P1373, P1594, CI13, CI10, and P2048) have a distinguishable difference, between 1.7 mm-3.7 mm, in body heights of the fifth lumbar vertebra. In the fully fused sacralized specimens (P1912 and CI2) and the lumbarized specimens (P1400 and CI3), there is either a slight or no difference (0 mm-1 mm). In addition, P1616 and P1643 do not have a distinguishable difference (0.3 mm and 1 mm respectively) in right and left body heights. In the specimens with a difference in right and left body heights, the affected side has a larger body height than the unaffected side. The asymmetry in right and left body height ranges from 1.7 mm-3.7 mm in five of the nine sacralized specimens, with an average difference of ca. 2.7 mm. In the control sample, there are some specimens with an asymmetrical body height, but the difference is typically less than 1.7 mm.

Table 4.1: Measurements of LSTV Specimens

Specimen	P2048	P1912	P1643	P1400	CI 2	CI 3	CI 10	CI 13	P1594	P1616	P1373
Side affected	left	both	left	Lumb.	both	Lumb.	right	left	right	right	left
L5 canal width	30	28.7	30	21	24	21.3	28	28	25	29	25
R pedicle width	20.3	17.3	14.7	15	14.3	13.3	21.7	18	23	17	17.7
L pedicle width	23.7	20.3	15.7	16	15.3	13.3	16.3	22.3	17	16	21.3
R body height	25.3	25	21	23. 7	23. 7	24. 7	25. 7	24	25	25	22
L body height	28	26	22	22.7	23	24.7	22	25. 7	22.7	24.7	25
Sacral Length (w/o L5)	104.3	83. 3	91. 7	84.3	77	88	111	87	83	74.7	92. 7
Sacral canal width	28		24	22.3		23	29	27. 7	23	31.3	25

In metrically analyzing the sacrum, the width of the first sacral body canal in respect to the fifth lumbar vertebra canal as well as the length of the sacrum in specimens with fully fused sacralization were the focus. In comparing the width of the fifth lumbar vertebra canal with the first sacral body canal in specimens with LSTV, there is little change in the width between them, with a range usually between 0.3 mm- 2.3 mm. The only specimen with a distinguishable difference is P1643, with a difference of 6 mm between the width of the fifth lumbar canal and first sacral body canal. In the control group, the difference in width between the two canals is also slight with few specimens with a difference greater than 3 mm.

CHAPTER 5

DISCUSSION

5.1 Sample Observations

In the osteological collection at the University of Texas at Arlington, LSTV occurs in 20% of the sample. This is expected, as the range of LSTV in previous sample studies has been between 4 and 30% (Apazidis et al., 2011; Konin & Walz, 2010). Although the range proposed in previous literature is large, perhaps it exemplifies the variability of LSTV. Furthermore, it is suggested that due to the high occurrence of this anomaly in this sample, and in several other previous studies, LSTV may be explained as normal human variation rather than a pathological condition. In addition, in this study sacralization was more prevalent than lumbarization in specimens with LSTV: 82% of the specimens with LSTV displayed characteristics of sacralization, and 18% had characteristics of lumbarization.

Previous research suggests that lumbarization is more common in females, while sacralization is more common in males (Eyo et al., 2001; Mahato, 2011). However, this research showed a higher occurrence of both lumbarization and sacralization in females. In the sample with sacralized fifth lumbar vertebrae, 56% are female and 44% are male. This opposes previous research as it is expected that males have a higher prevalence of sacralization. However, 100% of the specimens with lumbarization (N=2) are female. Although based on a very limited number of cases, this result is in agreement withprevious research. In general, of the 11 specimens with LSTV, 64% are female and 36% are male.

Although it is possible that these higher occurrences could be due to a higher number of females in the sample, there is still a substantial difference in the prevalence between females and males with LSTV. This favors the Sekharappa et al. 2014 study, in which it is suggested that females had a higher prevalence, of about 1.3 times, of LSTV in comparison to males. Although there is discrepancy in previous research regarding sex and LSTV, this study favors a higher occurrence in females than males.

5.2 Nonmetric Observations

In describing the sacralized fifth lumbar vertebrae, the Castellvi system was used. It was noted that two specimens, P1643 and P1616, are type Ia, meaning that there is a slight unilateral overgrowth of the transverse process. The transverse processes are not fully sacralized, but feature a more caudal shift which is indicative of overexpression of the *Hox 11* gene group (Carapuço, et al., 2005). The overexpression of *Hox 11* genes is known to cause a general caudal shift in vertebrae. Also, five specimens are type IIa, characterized by a unilateral pseudoarticulation of the transverse process and first sacral body. Although contested, this is the category that is often associated with lower back pain due to the joint issues at the pseudoarticulation.

In addition, type II and III are also associated with lower back pain due to the fusion of the transverse process(es) to the sacral ala(e). Two specimens in the sample, CI2 and P1912, show characteristics of type IIIb, or a bilateral fusion of the transverse processes. This fusion is caused not only by a caudal shift of the transverse processes, but also from a slight cephalic shift of the sacral alae. In this study, it is very common for type I and II sacralized specimens to be affected unilaterally and type III to be affected bilaterally. However, it cannot be proposed with high confidence that this observation applies to LSTV in other samples. Instead, the observation can be made that, within the specimens with a sacralized fifth lumbar vertebra, there is high variation in the presentation of the anomaly. There is not a specific side that is affected more often or a pattern of measured asymmetry across specimens with LSTV. In addition, the range of types from the Castellvi system in this sample further indicates that the presentation of sacralization is variable among individuals.

In the two specimens with characteristics of lumbarization, P1400 and CI3, the first sacral bodies were partially fused to the remaining sacrum. There were no instances of full lumbarization, or the presence of a sixth lumbar vertebra with a "missing" first sacral vertebra. Instead, the partially lumbarized specimens are characterized by a relatively wide cavity between the first and second sacral bodies at the joint area and alae. It should be noted that these specimens are older and therefore the cavity is not due to a lack of fusion because of young age. It is suggested that similar to sacralization, the expression of lumbarization is also variable on bone.

5.3 Metric Observations

Through this research it was found that bone size is affected by the presence of LSTV, characterized by asymmetry of pedicle width and body height of the fifth lumbar vertebrae. In the metric portion of this research, the left and right pedicle widths of the fifth lumbar vertebra of both LSTV and Non-LSTV specimens were measured. It was found that the pedicle width was asymmetrical in specimens with LSTV, specifically with sacralization. In addition, specimens with sacralization of the fifth lumbar vertebra had a wider pedicle on the affected side. It is suggested that the overgrown transverse process creates a thickening of bone at the pedicle. This shows that LSTV affects the pedicle width.

However, when comparing the measurements of the asymmetry, individuals did not exhibit a distinguishable pattern. This shows that although LSTV alters the thickness of bone, it is individualistically variable.

In addition to asymmetry in the widths of the right and left pedicle, the right and left body height of the fifth lumbar vertebrae also varies. It was found that the in specimens with LSTV there exists body height asymmetry, in which the affected side has a larger body height. This is another indicator that LSTV affects the fifth lumbar vertebrae in cases of sacralization. However, similar to the pedicle width, the metric difference between the asymmetrical body heights varies. It is suggested that there is no measurable pattern to the effect of sacralization on the fifth lumbar vertebra, meaning that the anomaly is highly variable in its expression on bone.

In metrically analyzing the effect of LSTV on the sacrum, both the difference in canal width between the fifth lumbar vertebra and first sacral body, as well as the length of the sacrum, not including the fused fifth lumbar vertebra, were studied. There was only one specimen with LSTV that displayed a distinct difference in canal widths. Also, there were a few specimens in the control sample with a relatively large difference in canal widths (between 3 mm-6 mm). However, it seems likely that this is due to the composite nature of the osteological collection studied and is not indicative of a possible alteration to the canal width due to the presence of LSTV. Finally, measuring the affected sacrum with the fifth lumbar vertebra included can make the specimen appear more robust or male, while a measurement without the fused fifth lumbar vertebra can result in a more gracile or female appearance. Due to this possible issue, more care should be taken when measuring and assigning sex to specimens with type IIIb sacralization.

CHAPTER 6

CONCLUSION

LSTV is generally considered to be a congenital anomaly caused by an overexpression of the *Hox 11* gene group. This creates a caudal shift of the fifth lumbar vertebra (referred to as sacralization) or a cranial shift of the first sacral body (referred to as lumbarization). It is possible that LSTV is linked to lower back pain due to joint problems from pseudoarticulations and fusion of the transverse processes of the fifth lumbar vertebra to the sacral alae. Termed Bertolotti Syndrome, lower back pain associated with LSTV is still a strongly contested topic.

In this study, LSTV was studied in three main phases: its occurrence in the sample (overall and by sex), nonmetric descriptions of the presentation of LSTV, and the anomaly's effect on bone through metric analysis. It was found that LSTV is fairly prevalent in the sample, with a 20% occurrence, and that LSTV, in both the form of sacralization and lumbarization, has a higher occurrence in females. Nonmetric descriptions exhibited the variation in presentation of LSTV in the specimens. In addition, the metric analysis further indicated the high variation within this anomaly. Although it was noted that LSTV created asymmetry in pedicle widths and body height of the fifth lumbar vertebra, there was no distinguishable pattern to the degree of variation in specimens with LSTV.

Overall, this study contributes to the debate of whether LSTV is correlated to biological sex, or more prevalent in one sex. This research favors the conclusion that LSTV is more common in females; however, due to the greater number of females in this sample it would be fruitful to reexamine this issue in more populations. In addition, to fully understand the origin of LSTV, more research should be conducted on the *Hox* 11 gene group, focusing on possible causes for overexpression. This would provide a better understanding of the genetic component of the anomaly and provide more insight into whether, or why, LSTV is more prevalent in certain groups.

APPENDIX A

PHOTOGRAPHS OF MEASUREMENTS AND LSTV SPECIMENS

Figure A.1: Measurement of Anterior

Figure A.2: Measurement of Superior

Figure A.3: Measurement of S1 Width

Figure A.4: Measurement of Ant-Post S1

Figure A.5: Measurement of L5 Canal

Figure A.6: Measurement of L5 Pedicle Width

Figure A.7: Measurement of L5 Body

Figure A.8: Measurement of S1 Canal Width

Figure A.9: Anterior View of

Figure A.10: Posterior View of P1643

Figure A.11: Anterior View of

Figure A.12: Posterior View of P2048

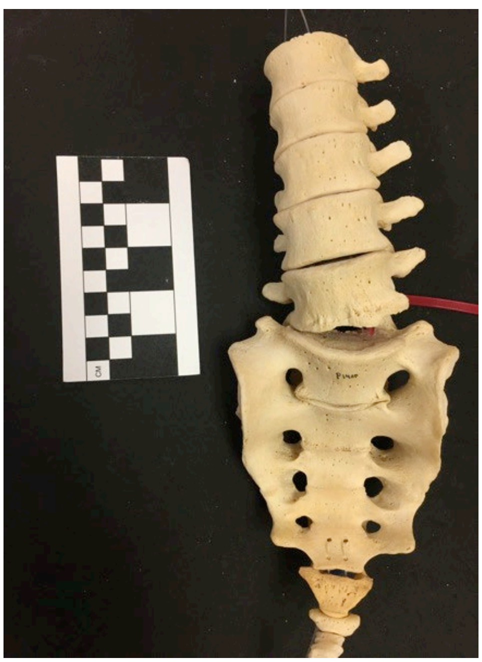

Figure A.13: Anterior View of

Figure A.14: Posterior View of

Figure A.15: Anterior View of P1912

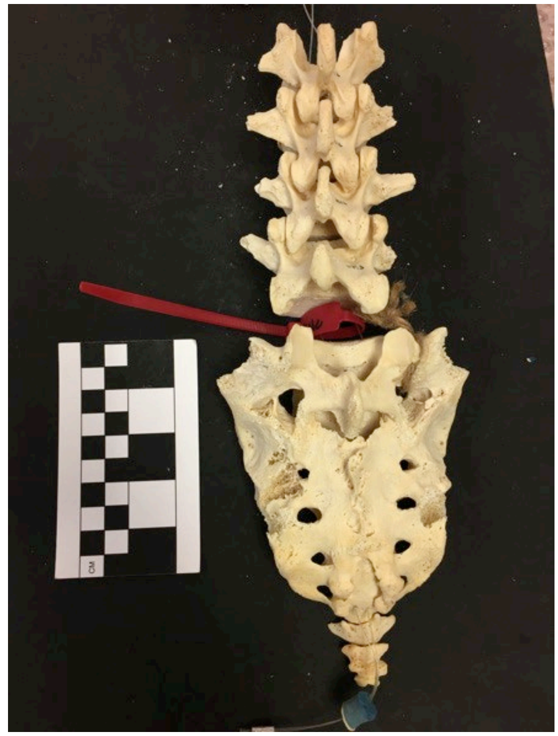

Figure A.16: Posterior View of P1912

Figure A.17: Anterior View of P1616

Figure A.18: Posterior View of P1616

Figure A.19: Anterior View of P1594

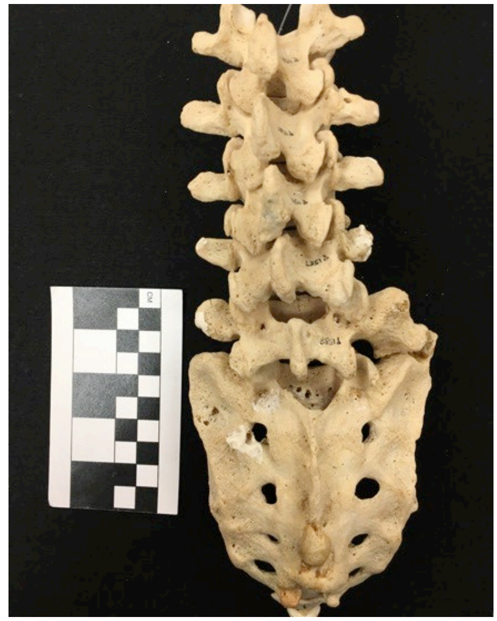

Figure A.20: Posterior View of P1594

Figure A.22: Posterior View of CI13

Figure A.22: Posterior View of CI13

Figure A.23: Anterior View of P1373

Figure A.24: Posterior View of P1373

Figure A.25: Anterior View of CI2

Figure A.26: Posterior View of CI2

Figure A.27: Anterior View of CI3

Figure A.28: Posterior View of CI3

Figure A.29: Anterior View of CI10

Figure A.30: Posterior View of CI10

APPENDIX B

MEASUREMENT DATA

Table B.1: Measurement Data (Highlighted Have LSTV)*

Specimens	P2048	P1912	P1643	P1400	P1805
L5 canal width	30	28.7	30	21	23
R pedicle width	20.3	17.3	14.7	15	16.7
L pedicle width	23.7	20.3	15.7	16	18. 3
R body height	25.3	25	21	23.7	27
L body height	28	26	22	22.7	23.3
Sacral Length (w/o L5)	104.3	83.3	91.7	84.3	99.7
Sacral canal width	28	27	24	22.3	26

Specimens	P2102	P1346	P2129	P1319	P1778
L5 canal width	25	24.7	22	22.3	27.3
R pedicle width	16	17	17. 7	14.7	16.3
L pedicle width	16. 3	17.3	17. 7	16	17
R body height	22.3	26.3	24.3	27	25
L body height	22	27	24	25	23.7
Sacral Length (w/o L5)	101	93	90.7	111.3	82
Sacral canal width	21.3	23	23	24.7	24.7

Specimens	P1698	P2021	P2075	P1994	P1724
L5 canal width	27.7	24.3	22.7	27	21
R pedicle width	15.3	19.3	13.7	13.7	17.3
L pedicle width	15.3	19.3	15.7	14.7	18.3
R body height	26.3	26	26	22	25
L body height	23.3	26	24	20	23
Sacral Length (w/o L5)	88.3	95.7	113	84.3	103.3
Sacral canal width	24	22.7	24	28	22.7

Specimens	P1562	P1481	P1751	P1832	P1886
L5 canal width	22	24	27.3	29	27
R pedicle width	15.3	14	14	15	17. 7
L pedicle width	15.3	14	14	15	18.3
R body height	21.7	23	23.7	22	23.3
L body height	20	22.7	24.7	22	23
Sacral Length (w/o L5)	89.7	91	94	87.7	98.3
Sacral canal width	26	28.3	25	27.7	28

Specimens	P1966	P1939	CI 1	CI 2	CI 3
L5 canal width	21	22		24	21.3
R pedicle width	15	16		14.3	13.3
L pedicle width	14.3	15		15.3	13.3
R body height	22	24.7		23.7	24.7
L body height	21.3	24.7		23	24.7
Sacral Length (w/o L5)	80	97	92	77	88
Sacral canal width	19	21	22	27	23

Specimens	CI 4	CI 5	CI 6	CI 8	CI 9
L5 canal width	21	25	22	25	
R pedicle width	14.3	18.7	15.7	23	
L pedicle width	14.3	18.3	15.7	24	
R body height	22.3	25.7	23.3	25.3	
L body height	23.7	26	23	25	
Sacral Length (w/o L5)	102	130.7	103.7	91.3	102.3
Sacral canal width	22	25	22.7	25	27

Specimens	CI 10	CI 11	CI 12	CI 13	CI 15
L5 canal width	28	25	23.3	28	24
R pedicle width	21.7	15.7	15	18	15
L pedicle width	16.3	17.3	15.3	22.3	14
R body height	25.7	24	19	24	27
L body height	22	23	19	25.7	27
Sacral Length (w/o L5)	111	91.3	82.7	87.3	106
Sacral canal width	29	19.7	22	27.7	26.7

Specimens	P524	P1453	P1426	P1120	P1373
L5 canal width	27	27	25	25	25
R pedicle width	18	18.3	16.3	14.7	17.7
L pedicle width	17	18.3	16.7	15.7	21.3
R body height	23	26	23	23	22
L body height	22	25	22	22	25
Sacral Length (w/o L5)				81.3	92.7
Sacral canal width				22.3	25

Specimens	P1535	P2156	P548	P1180	P1454
L5 canal width	26	23	24		
R pedicle width	17	13	13.7		
L pedicle width	17.3	14.7	12.7		
R body height	25	21.3	23		
L body height	25	20.7	22.3		
Sacral Length (w/o L5)	103.7	72.7		84.7	113.7
Sacral canal width	25	20		29	24

Specimens	Specimen 9	P1178	P1179	P1177	P1670
L5 canal width					25
R pedicle width					14.3
L pedicle width					15
R body height					25
L body height					23
Sacral Length (w/o L5)	88.7	96.7	83.3	91.3	89.7
Sacral canal width	32	30.3333333	22	27.7	23.7

Specimens	P1427	P1147	P1594	P1616
L5 canal width		26.3	25	29
R pedicle width		18.7	23	17
L pedicle width		18.3	17	16
R body height		24.3	25	25
L body height		23	22.7	24.7
Sacral Length (w/o L5)	90.3	102.3	83	74.7
Sacral canal width	25	28	23	31.3

*Average of three measurement attempts

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BIOGRAPHICAL INFORMATION

Stephanie Dolenz graduated *suma cum laude*, in May 2018 with an Honors Bachelor of Arts in Anthropology and a minor in German. Throughout her undergraduate career she has focused her studies on archaeology. In the summer prior to her junior year, she conducted fieldwork in Drawsko, Poland, through the Slavia Foundation, where her passion for bioarchaeology flourished. In the following semesters she shifted her academic focus to bioarchaeology, finding that the study of human remains is the most intimate approach in understanding past peoples. This summer, Stephanie will be traveling to South Africa and Italy to aid in the excavation of two cave sites.

In the past year, Stephanie has developed a keen interest in archaeogenetics. She will continue to pursue an M.S. in Bioarchaeology at Durham University in the United Kingdom in October of 2019. In her gap year, she hopes to work as a research assistant at a genetic research lab to become familiar with the equipment. She hopes to use archaeogenetics to better understand movement and migration during the medieval period, but is also keeping an open mind to other possible future research opportunities.