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Case Report

A Supernumeric Thoracic Vertebra Associated with Neural Tube Defects

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Abstract

When normal development is disrupted, it results in abnormal skeletal formation and may even affect the typical numeracy of bones. Numeric variation in the vertebral column may result from meristic or homeotic transformations. The presence of a supernumeric thoracic vertebra at the thoracolumbar junction is very rare. The prevalence of this rare anomaly remains unknown. Other congenital defects that may arise from disrupted development are neural tube defects (NTD) that result from defective closure of the posterior neural tube. The aim of this article is to report a case of meristic transformation associated with congenital defects in the vertebral column. The case was observed in the skeletal remains of a deceased individual from the Kirsten Collection at Stellenbosch University. Post-mortem evaluation of the skeletal remains revealed a thirteenth thoracic vertebra associated with neural tube defects in the vertebral column of an individual. Typically, there are only 12 vertebrae in the thoracic region and five in the lumbar region. In addition, severe degenerative joint disease in the vertebral column resulted in the fusion of vertebrae in both the cervical and the lumbar regions. As the vertebral column functions as the protective skeletal framework of the spinal cord, any structural malformations and biomechanical disruptions can lead to severe neural and musculoskeletal damage. Ultimately, this can decrease the quality of life in patients. This case study is significant as it is extraordinarily rare to observe a supernumeric vertebra in the thoracic region, which in addition, is associated with neural tube defects. All clinical procedures that consider the thoracolumbar junction will be complicated. This may further complicate clinical procedures performed the thoracolumbar junction.

Keywords: *Anatomy; Meristic Transformation; Vertebral Column; Congenital Anomalies*

Abbreviations

C1-7: Cervical Vertebrae; NTD: Neural Tube Defect; T13: Thoracic Vertebra 13; TLTV: Thoracolumbar Transitional Vertebra

Introduction

The vertebral column develops under regulation of the notochord from the two layers of pre-somatic mesoderm. During segmentation, the paraxial mesoderm is divided into approximately 44 pairs of somites [1-3]. The various somites develop into the respective vertebrae [2,4-8]. When normal development is disrupted, it results in abnormal skeletal formation and may even result in numeric variation.

According to Bateson (1894), numeric variation in the vertebral column may result from: (1) the addition of a segment known as a meristic transformation or (2) the change of one series identity at the expense of another known as homeotic transformation [9]. A supernumeric thoracic vertebra at the thoracolumbar junction that does not alter the number of vertebrae in adjacent regions, therefore, results from a meristic transformation. The presence of a supernumeric thoracic vertebra is very rare [5,10,11]. For this reason, a prevalence has not yet been recorded in published literature.

Other congenital anomalies that may arise from disrupted development are neural tube defects (NTD's). NTD's result from the defective closure of the posterior part of the neural tube between 21 and 28 days of prenatal development [12-14]. Infants who develop other NTD's have a high probability of developing severe lifelong disabilities, deteriorating their quality of life [15].

Individuals with NTD's are more likely to undergo surgical procedures in the vertebral column. The thoracolumbar junction is biomechanically the least stable area of the vertebral column and likely to be susceptible to vertebral fractures [16,17]. In cases of a supernumeric vertebra at the thoracolumbar junction, the biomechanical stability of the vertebral column is altered. The altered morphology may further complicate surgical procedures.

Aim of the Study

The aim of this article is to report a case of a supernumeric thoracic vertebra that resulted from meristic transformation and was associated with neural tube defects in the vertebral column.

Methods

This article is a report compiled from the evaluation of the skeletal remains of a deceased individual from the Kirsten Collection at Stellenbosch University.

The case was discovered during the data collection of an alternative study. The study was ethically cleared by the Health Research and Ethics Committee of Stellenbosch University which conforms to the principles within the Declaration of Helsinki (1964). The allocated ethics number is S13/05/100.

This case study discusses the presence of a supernumeric thoracic vertebra with corresponding costal articulation. The anomaly was associated with disrupted neural tube closure in the thoracic and cervical regions of the vertebral column. In addition, the specimen in the case study suffered from degenerative joint disease. Various multifocal lytic and sclerotic lesions are visible at joints in the axial and appendicular skeleton.

It was critical to differentiate whether the numeric variation was homeotic or meristic. Differentiation was performed by sequencing the vertebral column and evaluating the number of vertebrae in every region. Meristic variation in a region results in variability of the affected region without affecting adjacent regions and alters the total number of vertebrae present in the vertebral column.

Vertebrae were sequenced according to Bron., et al (2007). The last vertebra with costal articulation is numbered as the last thoracic vertebra. The subsequent vertebra is numbered as the first lumbar vertebra [18].

Results

Case Study

Specimen AN291, from the Kirsten Collection at the University of Stellenbosch, is the skeletal remains of a 78 year old male (1894-1972). According to medical records, the patient was admitted to Karl Bremer Hospital, in Elsies River South Africa with terminal emphysema. Post-mortem evaluation of the skeletal remains revealed several congenital defects and pathology present in the vertebral column of this individual.

The most significant congenital anomaly observed during evaluation of the skeletal remains is thirteen thoracic vertebrae with corresponding costal facets for rib articulation on the vertebral bodies and transverse processes. Typically, there are 12 thoracic and 5 lumbar vertebrae present in the vertebral column. In figures 1-3, the first 12 thoracic vertebrae can be observed.

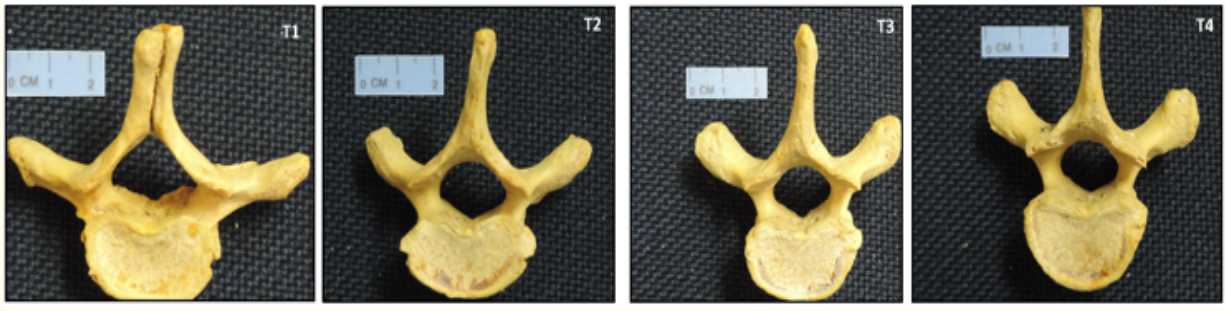

Figure 1

Figure 2

Figure 3

The supernumeric thoracic vertebra is located at the thoracolumbar junction (Figure 4). The facets for costal articulation were present on the vertebral body. Evaluation suggests that the individual had thirteen pairs of ribs corresponding to thirteen thoracic vertebrae.

It was, observed that the T13 vertebra retained partial features characteristic of the lumbar region, such as mammillary bodies and an accessory process on the right side. This suggests that vertebra is a thoracolumbar transitional vertebra (TLTV) as it is a vertebra located at a regional junction with morphological traits that are intermediary of the two adjacent regions [19].

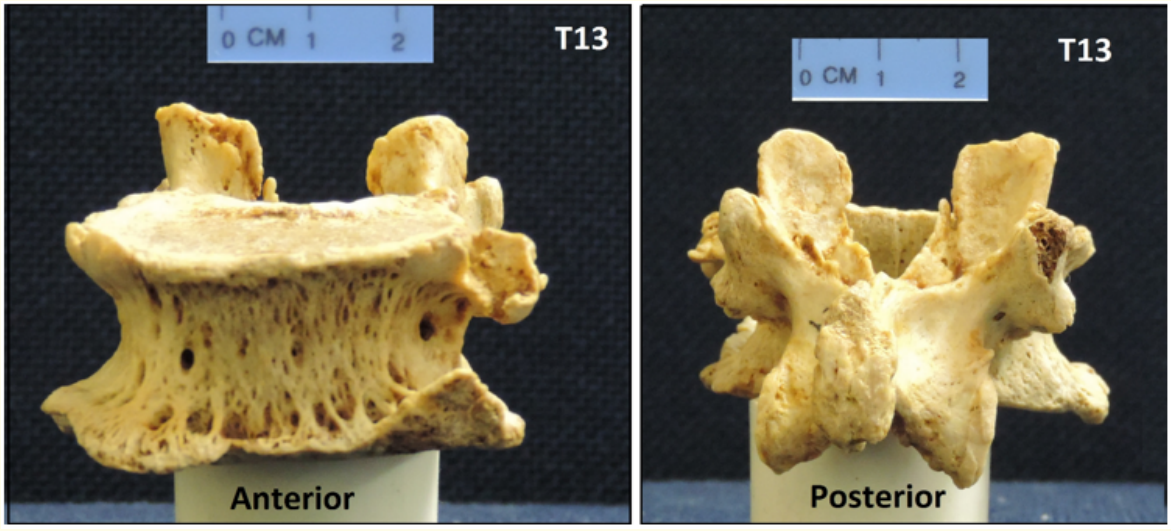

Figure 4

The additional thoracic segment present did not result from a homeotic transformation. The vertebral segments in the other regions of the vertebral column were unaltered with seven cervical, five lumbar and five sacral vertebrae present. It can, therefore, be confirmed that this anomaly resulted from a meristic transformation during development.

Other congenital defects observed were NTD's. The first NTD was present in the cervical region. The first cervical vertebra (C1) exhibited disrupted closure of the anterior and posterior neural arches, thus dividing the atlas into two segments (Figure 5). The second NTD observed was present in the thoracic region (Figure 1). The first thoracic vertebra (T1) in the vertebral column exhibited disrupted fusion of the spinous posterior neural arch forming the spinous process. This resulted in a bifurcating neural arch.

The axis (C2) and third cervical vertebra (C3) are fused by lytic lesions and ossification of the ligamentum flavum (Figure 6). Additional fusion resulted from degenerative joint disease of the articular surfaces between the inferior articular facet of C2 and the superior articular facet of C3. Despite minor osteophyte and syndesmophyte formation, there is no indication of fusion between the vertebral bodies of C2 and C3.

In the lumbar region, the first and second lumbar vertebrae were fused at the vertebral bodies resulting from severe lytic lesions and degenerative joint disease (Figure 7).

Figure 5

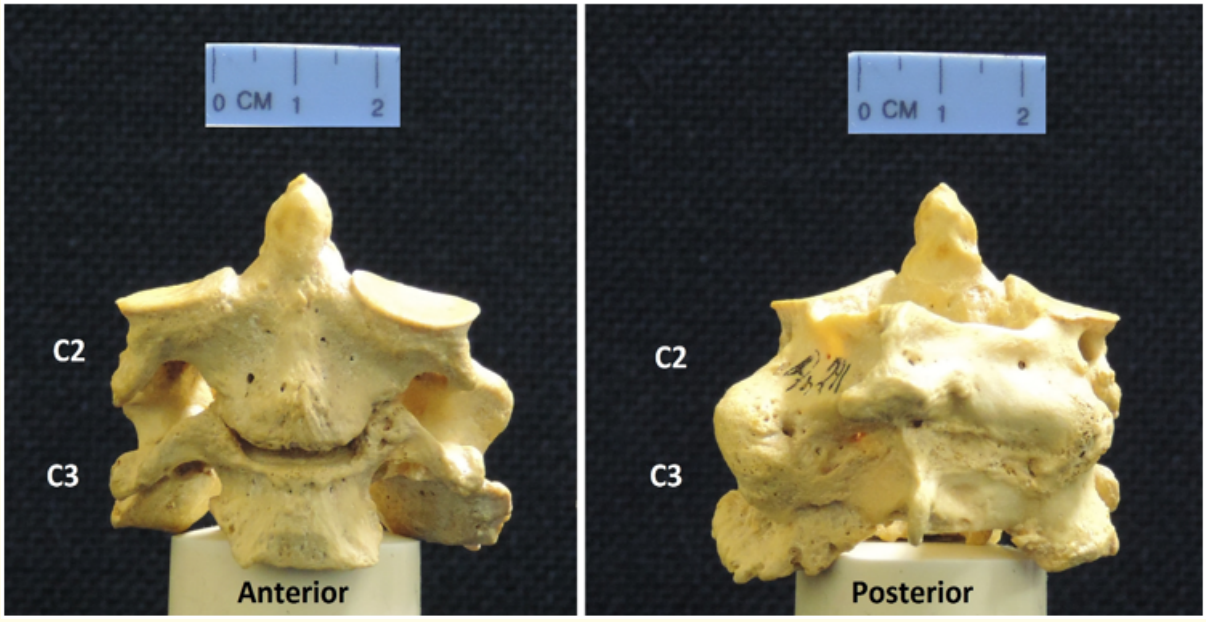

Figure 6

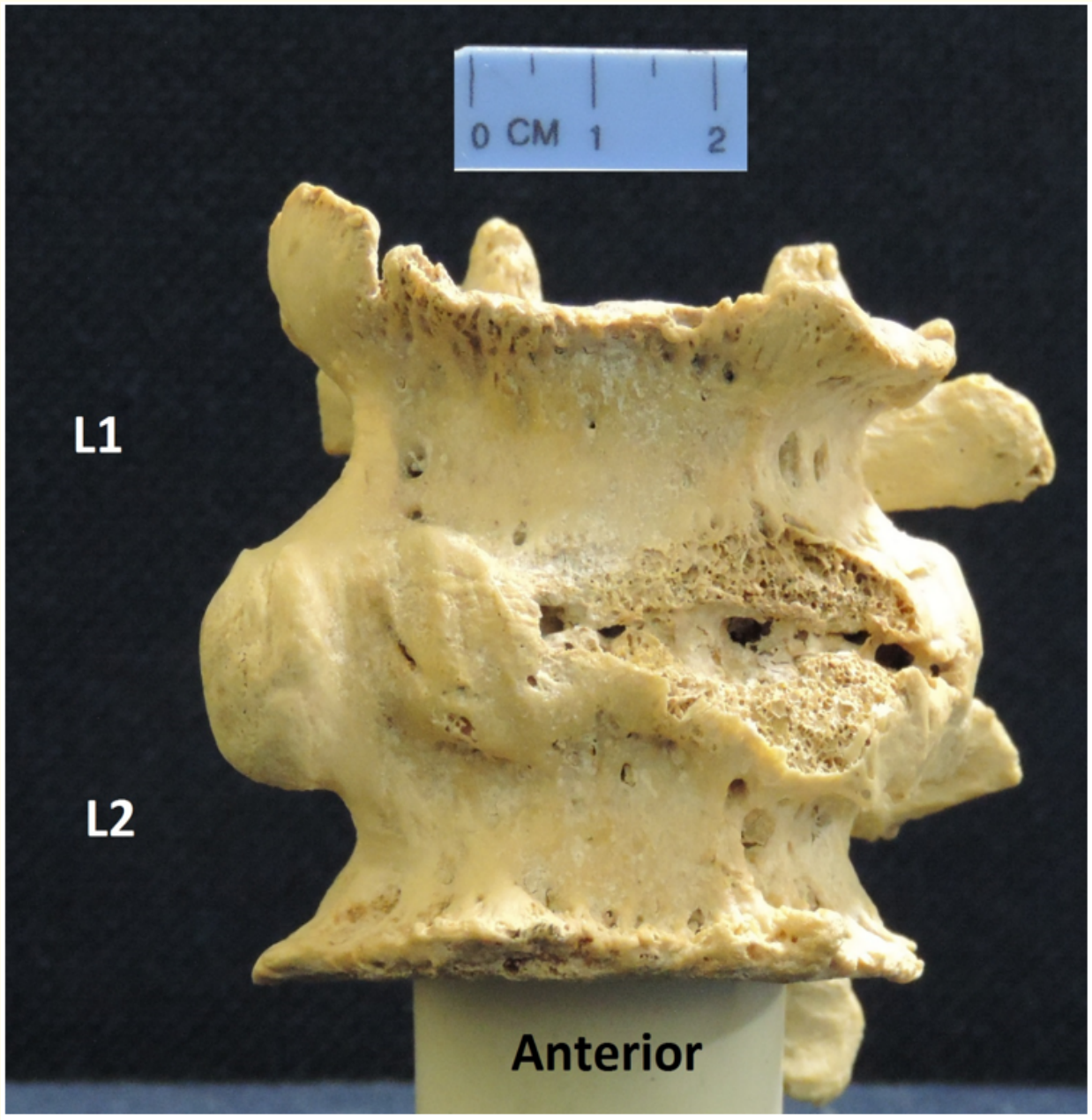

Figure 7

Discussion

As the vertebral column functions as the protective skeletal framework of the spinal cord, any structural malformations and biomechanical disruptions can lead to severe neural and musculoskeletal damage. Ultimately, this can decrease the quality of life in patients.

Most patience born with neural tube defects suffer from life-long disabilities such as sexual dysfunction, bowel dysfunction and motor neuron paralysis.

Patients with congenital defects in the vertebral column are more likely to undergo reparative surgical procedures to help increase biomechanical security and decrease further potential structural damage to surrounding tissue.

Deviation from typical vertebral anatomy can result in confusion by clinicians that lead to significant clinical errors [3,19]. There is not much published literature based on the T13 vertebra or the clinical relevance regarding its presence at the thoracolumbar junction. The thoracolumbar junction is a significant anatomical region for health care professionals. It is a structurally weak biomechanical region in the vertebral column that frequently leads to spinal instability and injury. According to Kim., *et al.* [17], majority (90%) of spine fractures are related to this region. The article further states that 24% of the fractures at the thoracolumbar junction are associated with neurological injury that may results in paralysis and require surgical intervention [16,17,20,21].

Severe instability, structural damage and post-traumatic deformity is the most frequent indication for surgery [20]. The variation of the spinal cord length associated with the additional vertebra at the thoracolumbar junction is unknown in this case, however, should be considered by clinicians during surgical intervention prospectively. As the thoracolumbar junction is the end-point of the spinal cord and start of the corda equine, structural damage of the spinal cord may occur during surgical procedures. The presence of an additional vertebra within the region, where spinal fractures are more likely, further complicates procedures and increases the probability of spinal damage and paralysis.

There are several biomechanical factors that contribute to the instability of the thoracolumbar junction [16,17,21]. It is the transitional site from the kyphotic thoracic to the lordotic lumbar region. This results in strenuous transition of weight between vertebral bodies and articular facet joints [21]. The additional vertebra observed in this case alters the transition of weight at the thoracolumbar junction and may further destabilize this region, leading to injury.

Conclusion

The vertebral column functions as the protective skeletal framework of the spinal cord, any structural malformations and biomechanical disruptions can lead to severe neural and musculoskeletal damage. This case study is significant as it is rare to observe a supernumeric vertebra in the thoracic region, which in addition, is associated with neural tube defects. All clinical procedures that consider the thoracolumbar junction must be performed with care, taking into account the atypical morphology. Clinicians should be aware that cases such as these may be encountered in order to avoid clinical errors or surgical complications.

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