

A124 Modern Variation in Vertebral Column Segmentation and Transitions

Janet E. Finlayson, MA*, University of Florida, 2033 Mowry Road, Rm G-17, Gainesville, FL 32610; and Amanda N. Friend, MA, C.A. Pound Human Identification Laboratory, 2033 Mowry Road, Gainesville, FL 32608

The goal of this presentation is to describe the variation observed in individuals accessioned at the University of Florida C.A. Pound Human Identification Laboratory (CAPHIL) from the modal human vertebral formula and discuss potential effects of this variation on forensic analyses. After attending this presentation, attendees will understand the value in assessing variation within the vertebral column.

This presentation will impact the forensic science community by highlighting the importance of understanding modern human biological variation and the potential of anomalous development to affect forensic identification processes.

The modal formula of the human vertebral column consists of 7 cervical, 12 bilaterally rib-bearing thoracic, 5 lumbar, 5 fused sacral, and 4 coccygeal vertebrae. Variation in the number of vertebrae and associated ribs can occur within each vertebral column region. These variations are generally attributed to embryological developmental irregularities, such as an amodal number of somites or abnormal patterns of genetic signaling.¹⁻³

This study accessed skeletal inventory and descriptive data of 55 ($F=14$, $M=41$) identified and unidentified individuals analyzed at the CAPHIL between 2007 and 2017. The individuals ranged in age at death from late adolescence to older adult, and the sample varied in ancestry. Sample selection required the presence of at least one developmental vertebral or rib anomaly. Selection of developmental anomalies was limited to differences from the modal vertebral column expression, including: (1) regional segment count; (2) segmental cranialization or caudalization; and, (3) improper segmentation. This criteria allows for identification of variance in vertebra/rib count, transition locations, and segment separation errors. Thoracic and lumbar vertebrae are typically defined morphologically, with the orientation patterns of Superior and Inferior Articular Facets (SAFs/IAFs) or articulation to ribs as defining criteria. For this study, designation of a thoracic vertebra is determined by SAF/IAF orientation patterns and possession of costal facets; designation of a lumbar vertebra is determined by SAF/IAF orientation, regardless of rib-bearing status.

Forty-one of the studied individuals varied in segmental count in at least one region (74.5%). Of these individuals, three had 11 bilaterally rib-bearing thoracic vertebrae (7.3%), 2 had 13 bilaterally rib-bearing thoracic vertebrae (4.9%), 16 had 6 lumbar vertebrae (39.0%), 21 had 6 sacral segments (51.2%), and 4 had 3 coccygeal segments (9.8%).

Thirty-seven individuals exhibited variation in at least one transitional region (67.3%). Of these individuals, four exhibited variation in two regions (10.8%). In one male, C7 articulated unilaterally to a rib (2.7%). One male had a unilateral supernumerary rib of indeterminate anatomical origin in addition to the 24 thoracic ribs (2.7%). Sixteen individuals exhibited bilateral lumbar ribs (43.2%), one of whom exhibited a bilateral set on L1 and a unilateral rib on L2 and two of whom exhibited a unilateral rib on L1. For 13 individuals (35.1%), the thoracolumbar transition occurred at the level of T11; in ten of these cases (76.9%), the vertebra inferior to T11 had lumbarized SAFs/IAFs and was rib-bearing, followed inferiorly by five non-rib-bearing lumbar vertebrae. Thirteen individuals displayed sacralization (35.1%), with seven instances at the level of L5 and six at the level of L6. Of individuals with a sixth lumbar vertebra, 75.0% exhibited sacralization of the element. Four individuals (10.8%) exhibited S1 lumbarization.

Three individuals had segmentation defects (5.4%), of whom there was separation failure of C2/C3 in one female (33.3%), of C7/T1/T2 in one male (33.3%), and of right ribs 3/4 in one male (33.3%).

In this study, the vast majority of developmental vertebral column anomalies were expressed in the caudal regions. This is likely due to abnormal genetic signaling during embryonic specification of individual vertebral identity in which the signaling code becomes caudally complex. Certain cervical anomalies can be associated with detrimental conditions that may result in perinatal or infantile death; thus, the cervical region has strong developmental constraints.^{4,5} This constraint may contribute to the relatively low frequency of cervical anomalies observed in this study.

This study describes relative frequencies of developmental vertebral anomalies observed in a forensic sample. Vertebral anomalies may affect forensic stature estimations, as the vertebral column majority directly contributes to height, and standard stature calculation techniques assume modal vertebral frequencies. Further, deviance from modal expressions can affect the condition of the vertebral column, which may have clinical implications and serve as potentially individualizing pathological features.^{6,7}

Reference(s):

1. Barnes E. 1994. Developmental defects of the axial skeleton in paleopathology. Niwot, CO: University Press of Colorado.
2. Giampietro F.P., Dunwoodie S.L., Kusumi K., Pourquié O., Tassy O., Offiah A.C., Cornier A.S., Alman B.A., Blank R.D., Raggio C.L., Glurich I., Turnpenny P.D. 2009. Progress in the understanding of the genetic etiology of vertebral segmentation disorders in humans. *Ann N Y Acad Sci.* 1151: 38-67.
3. Kmita M., Duboule D. 2003. Organizing axes in time and space; 25 years of colinear tinkering. *Science.* 301:331-333.
4. ten Broek C.M.A., Bakker A.J., Varela-Lasheras I., Bugiani M., van Dongen S., Galis F. 2012. Evo-devo of the human vertebral column: On homeotic transformations, pathologies and prenatal selection. *Evol Biol.* 39:456-471.
5. Furtado L.V., Thaker H.M., Erickson L.K., Shirts B.H., Opitz J.M. 2011. Cervical ribs are more prevalent in stillborn fetuses than in live-born infants and are strongly associated with fetal aneuploidy. *Pediatr Dev Pathol.* 14(6):431-437.
6. Bron J.L., van Royen B.J., Wuisman P.I.J.M. 2007. The clinical significance of lumbosacral transitional anomalies. *Acta Orthop Belg.* 73(6):687-695.
7. Konin G.P., Walz D.M. 2010. Lumbosacral transitional vertebrae: Classification, imaging findings, and clinical relevance. *Am J Neuroradiol.* 31(10):1778-1786.

Anomalous Variation, Vertebrae, Development