



Pathology/Biology Section - 2014

G92 Skeletal Pathology With Associated Soft Tissue Defects Inform Physiognomy

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After attending this presentation, attendees will understand how skeletal pathology can be used to infer associated soft tissue pathology to better provide a physical description of a decedent to aid in identification of nearly skeletonized remains. Presented are the characteristics of Klippel-Feil Syndrome and a case study of identification aided by a profile of the common presentation of this syndrome.

This presentation will impact the forensic science community by serving as an example of interdisciplinary collaboration for developing a biological profile and identification.

When conducting a postmortem evaluation, pathologists note skeletal abnormalities that are readily visible on radiograph or noted in dissection; however, their significance may be overlooked unless related to cause of death. However, when examining unidentified skeletal remains, these abnormalities may provide clues related to the soft tissue physiognomy of the decedent and/or associated defects. These findings can then be combined with other aspects of the biological profile to provide a better description of the decedent, including possible conditions that would be apparent in the decedent's appearance that law enforcement can then use to search for possible putative decedents.

This study presents a case of Klippel-Feil syndrome, a rare skeletal abnormality (1 in 42,000), marked by congenital failure of vertebral segmentation, resulting in fusion of at least two cervical vertebrae with other skeletal changes often noted. The most typical presentation of this syndrome is fusion of C2 and C3, but fusions of any sub-set of cervical vertebrae are possible and, in some instances, fusion of thoracic or lumbar vertebrae have been noted. Additional skeletal changes include the increased incidence of scoliosis, transitional vertebra, rib abnormalities, cleft palate, cranial asymmetry, and short stature. Aside from the skeletal presentation of this syndrome, there are numerous soft-tissue or physical presentations that may have been noticeable to family and acquaintances of the decedent. Signs of Klippel-Feil most often include a low posterior hair line, a shortened neck, and limited range of motion of the neck. Additional conditions that may be noted include hearing problems, kidney and heart conditions, shortened stature, webbed neck and digits, and elevated scapulae. The common presentations of this syndrome were used to develop a profile that was utilized by law enforcement in their investigation to establish a presumptive identity and to secure medical records for a putative decedent. Once medical records are located, personal identification is accomplished via antemortem/postmortem radiographic comparison. In these cases, the radiographic comparison is greatly aided by the unusual morphology secondary to the pathological condition.

Skeletal Pathology, Klippel-Feil Syndrome, Forensic Identification