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Case Report

Klippel–Feil Syndrome: The Curious Case of the Skeleton of a Young Slavic Soldier Who Died in 1946

Alessia Leggio [1,*], Massimo Gallieni [2], Pasquale Puzo [3], Francesco Introna [1] and Sara Sablone [1]

[1] Department of Interdisciplinary Medicine, Section of Legal Medicine, Policlinico di Bari Hospital, University of Bari, Piazza Giulio Cesare 11, 70124 Bari, Italy; francesco.introna@uniba.it (F.I.); sara.sablone@policlinico.ba.it (S.S.)
[2] Department of Neurosurgery, International Neuroscience Institute, Rudolf Pichlmayr Str. 4, 30625 Hannover, Germany; gallieni@ini-hannover.de
[3] Department of Medical Science, Neuroscience and Sense Organs, Section of Ophthalmology, Bari Policlinico Hospital, University of Bari, 70124 Bari, Italy; puzopasquale@gmail.com
* Correspondence: alessialeggio11@gmail.com

Abstract: This paper describes the curious case history of the famous and rare Klippel Feil syndrome type II, identified in the skeleton of a young Slavic soldier who died in 1946. It is a very interesting case given the fusion of the C1 and C2 cervical vertebrae, which prevented the young soldier from rotating his skull while alive. Klippel–Feil syndrome is a rare osteopathology and involves fusion of the vertebrae of the spine and is linked to other pathologies that indicate the presence of this pathological condition. In the present study, several basic investigations were carried out: a macroscopic observation to document the abnormalities throughout the rest of the skeleton, a morphological one to determine the identifying anthropological analysis, a pathological one to determine the pathologies present and a radiographic one to diagnose and confirm the pathology. Studying the pathologies of the past is fundamental in order to know the evolution and behaviour of the disease today, and the investigations carried out in this case study determined what the limitations of the young soldier were, how this disease may have influenced his activities as a soldier during World War II and through which therapies the syndrome may have been treated in such an important historical period.

Keywords: paleopathology; Klippel–Feil syndrome; forensic anthropology; Slavic soldier; World War II

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1. Introduction

In the summer of 2019, the Institute of Forensic Medicine of Bari and the Forensic Anthropology Laboratory of the same department undertook research work within the Monumental Cemetery of Bari that led to the discovery of 93 skeletal remains of Yugoslavian origin, most belonging to soldiers missing for a very long time who died during the Second World War in the famous Prison Camp of Torre Tresca (Bari, Puglia). Among these remains was the skeleton of a young soldier who died in 1946, probably from complications of a rare disease. This disease had been sporadically documented in anthropological literature as Klippel–Feil syndrome, in this case type II with fusion of the first and second cervical vertebrae. The detection of skeletal pathology and trauma provides important information that can help identify an unknown subject and may provide possible evidence of the circumstances or cause of death. Skeletal pathology can be very important in forensic contexts in cases where other aspects of the osteological profile are very similar [1].

Klippel–Feil syndrome was first described by Klippel and Feil (1912) in a clinical case in which massive fusion of the cervical vertebrae was observed. The classic clinical triad of symptoms is short neck, low back hairline and restriction of movement in the neck [2]. The different manifestations due to the different genetic pathways made it possible to define three degrees of cervical fusion (I–III):

Type I: the classic Klippel–Feil syndrome with several cervical and upper thoracic vertebrae embedded in a bony block showing a complex abnormal appearance and often associated with other major defects [3].

Type II: only two or three vertebral segments are involved, with the second and third cervical segments most commonly affected, and there is evidence of autosomal dominant inheritance; the fifth and sixth cervical segments are the next most commonly involved and are usually expressed as an autosomal recessive trait. If thoracic vertebrae are involved, the defect most commonly occurs between T2 and T5. In some cases, hemivertebral or atlanto–occipital fusions are present [2,4].

Type III: fusion of cervical vertebrae coexisting with segmental vertebral defects in the thoracic and lumbar regions [2].

Only a few old cases of Klippel–Feil syndrome have been reported so far, particularly in Europe [5]. It is very important to study and document rare osteopathies in individual cases to gather information that can be used in population studies [6]. The bones of the young soldier's skeletal remains were subjected to a preliminary investigation, which at first only revealed Klippel–Feil syndrome. A second, more specific analysis allowed all the anomalies associated with this pathology to be examined: bilateral and symmetrical occipital condylar hypoplasia, cranial asymmetry, deviation of the nasal septum, spina bifida occulta type III and mild cervical dextroscoliosis [7–9].

2. Case Report

In 2019, the Institute of Forensic Medicine of Bari (Apulia, Italy) undertook research that led to the discovery of 93 skeletal remains of Slavic origin, abandoned in the ossuary of the Monumental Cemetery of Bari. The skeletal remains were placed inside small steel boxes, and during the first general inspection, war badges were found (Figure 1) that allowed the identification and historical contextualizing of these skeletal remains. After a thorough historical analysis of the badges, it was determined that the skeletal remains belonged to young Slavic soldiers of the Royal Yugoslav Army, Cetnico of the Ravna Gora national movement in exile in 1941, under the command of Serbian General Dragoljub "Draža" Mihailović [10]. These soldiers were most likely, given the historical evidence, deported, and then, died in the Torre Tresca prison camp (Bari, Apulia) during World War II [11].

Figure 1. Objects placed in coffin No. 33. Frieze of the Royal Yugoslav Army, bearing the monogram of Peter II Karadordevic (**a**), and military badge of Ravnogorksi, representing the movement's struggle of Ravna Gora under the leadership of General Dragoljub Draža Mihailovi'c (**b**).

3. Materials and Methods

The remains of Private Chetnic were not subjected to AMS radiocarbon dating [12] because there is historical documentation on paper that certifies the subject's personal data such as date of birth and death: 1921–1946; there is also an identification tag engraved inside the iron box in which the skeleton is stored, with personal data such as first and last name and military rank, which also confirms the origin of these remains. The skeleton was recovered almost 90% intact [13], completely skeletonised without soft tissue and with green pigmentation on the front of the skull due to excessive humidity in the area where the box was stored. Pathologies can influence the results of the anthropological investigation during the reconstruction of the biological profile and for these reasons, in the present case, different methods were applied during the investigation. The skeletal remains were subjected to several stages of investigation: macroscopic, morphological, dental and pathological analysis. Given the known subject, age at death was confirmed biologically, using epiphyseal fusion, age-related tooth wear and the degree of obliteration of cranial sutures [14,15].

Dental analysis for age determination was applied to the entire dental system through morphological and macroscopic observation of dental wear [16]. Finally, suture obliteration is at level 1 [17]. Congenital anomalies could influence sex determination based on morphological data; therefore, even in the case of a known subject, it was necessary to determine sex by applying different methods. Sex determination was mainly based on the characteristics of the skull and pelvis, the skeletal districts with the greatest sexual dimorphism [18–22]. After reconstructing and confirming the subject's biological profile, a pathological analysis revealed multiple abnormalities in the skeletal system under investigation. The first and most obvious is the fusion of the C1 and C2 vertebrae. The skull shows symmetrical and bilateral condylar hypoplasia and deviation of the nasal septum. The cervical vertebrae are affected by mild dextroscoliosis and the sacrum shows spina bifida occulta type III. However, the morphology of these bones is apparently normal. A radiographic examination confirmed the actual C1–C2 fusion, allowing the diagnosis of Klippel–Feil syndrome type II and affirming the correlation of the additional pathologies found (Figure 2c).

Figure 2. Klippel–Feil Syndrome, fused vertebral block of C1 and C2. (**a**) Front projection, (**b**) rear projection. X-ray front projection (**c**), evident fusion between the C1 vertebra and the C2 vertebrae.

4. Results

The epiphyseal fusion process suggested an age at death between 18 and 25 years [10,11]. Dental analysis for age determination based on dental wear indicates an age at death between 20 and 24 years [14]. Finally, the degree of obliteration of the suture is stage 1 [13]. Overall, the different methods applied indicate an age at death between 23 and 25 years, confirming the age data indicated on the plaque found inside the box containing the young soldier's remains.

Klippel–Feil syndrome is a rare skeletal disease mainly characterised by an abnormal union or fusion of two or more vertebrae that concentrates specifically on the cervical vertebrae, as in the present case. Some affected individuals have an abnormally short

neck and limited neck and head movement due to fusion of the vertebrae. The syndrome is congenital but can only be diagnosed late in life when symptoms become worse or evident. This condition may sometimes be related to a variety of other symptoms and physical anomalies and may manifest as an isolated anomaly or as a syndrome with associated anomalies.

In many individuals, the disease seems to occur randomly for no apparent reason. In other cases, the syndrome is inherited as an autosomal dominant or autosomal recessive trait or may be associated with genetic mutations [23]. Klippel–Feil syndrome type II is characterized by localised fusion of one or two cervical vertebrae, hemivertebrae and atlanto–occipital fusion; in the case at hand, there is total fusion of the C1 and C2 vertebrae, causing severe limitation in flexion and extension of neck rotation (Figure 2a,b). The entire skeletal system presents several anomalies associated with this syndrome as bilateral and symmetric occipital condylar hypoplasia, cranial asymmetry, nasal septum deviation, spina bifida occulta type III and mild cervical dextroscoliosis.

Macroscopic observation of the skull was performed to identify abnormalities associated with Klippel–Feil syndrome. The skull shows a mild parietal and occipital asymmetry causing an abnormal deformity on the lower part of the left occipital portion (Figure 2c). The subject presents a cranial index of 76, indicating an intermediate shaped mesocranial skull [24]. A further abnormality is the presence of the nasal septum deviation from its correct orientation.

During the analysis, a bilateral and symmetric occipital condylar hypoplasia of the foramen magnum was found: the condyles are complete but asymmetrical in the dorso-ventral direction (Figure 3a,b). This anomaly is due to the fusion of the C1 and C2 vertebrae, which are bilaterally symmetrical between the lower right and upper left portions, thus creating a difference in height that causes the skull and the entire skeletal system to tilt to the left. Asymmetry has been found in the left occipital portion of the skull (Figure 3c). Deviated septum can occur due to various aetiologies such as irregular bone development, trauma or overgrowth of the turbinates; this contributes to nasal obstruction (Figure 3d) and can lead to a level of facial deformity [25].

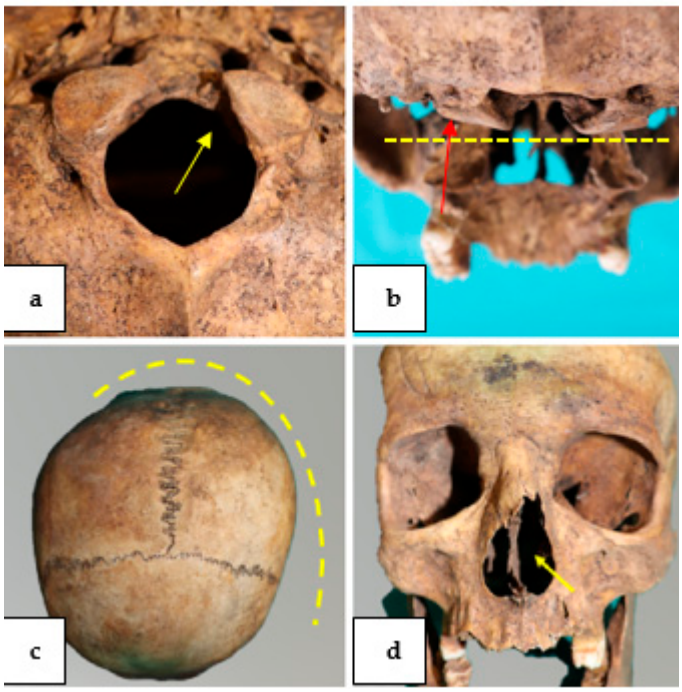

Figure 3. Bilateral asymmetry of the occipital condyles with hypoplasia of the left condyle. (**a**) skull base in axial projection, (**b**) skull base in postero-anterior coronal projection); (**c**) asymmetry in the left occipital portion of the skull; (**d**) deviation of the nasal septum. (Frontal skull projection.)

This skeleton shows an example of mild cervical dextroscoliosis affecting the C1–C2 vertebral blocks, probably caused by the posture induced by the fusion of these vertebrae (Figure 4a). Scoliosis is a deviation of the normal vertical line of the spine that presents a lateral curvature with a rotation of the vertebrae within the curve. Typically, for scoliosis to be considered, there should be at least 10° of spinal angulation on the posterior–anterior radiograph associated with vertebral rotation [26]. In addition to the right cervical scoliosis, the skeleton shows another related pathology: spina bifida occulta (Figure 4b). Spina bifida is a developmental defect of the spinal column in which the laminae do not fuse, and the spinal cord remains relatively unprotected due to the absence of a bony back wall on the spinal cord.

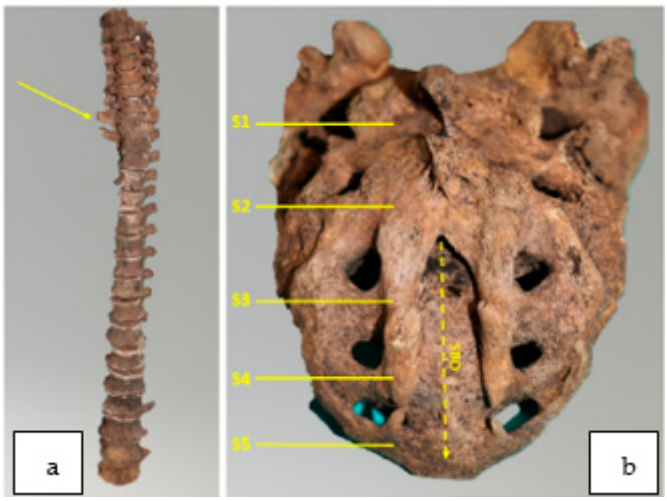

Figure 4. Cervical dextroscoliosis (**a**), front projection; shows type-III SBO (**b**), front projection. Here the level of apex of sacral hiatus is at S2 vertebra. SBO—sacral spina bifida occult, D—dorsal surface of sacrum, S1, S2, S3... are the levels of sacral spines.

This condition can be classified into two types depending on its condition: exposed or occult [27]. The presence of spina bifida in the sacral region spreading from S1 to S5 has been termed to be sacral spina bifida occulta (SSBO). Our case study specifically shows a spina bifida occulta type III with correct fusion of the S1–S2 vertebrae and non-fused opening of the S3, S4 and S5 sacral vertebrae. These two diseases are often related to Klippel–Feil syndrome.

5. Discussion

Klippel–Feil syndrome is a rare complex congenital pathology, and its main feature is the fusion of two or more cervical vertebrae associated with other possible skeletal abnormalities mainly affecting the spine [28]. Congenital fusion of two vertebrae into one block (C1, C2), as in the present case, occurs when there is an abnormal division of the somites, resulting in a disorder known as Klippel–Feil syndrome [29,30]. According to Feil's classification, the case described here is consistent with type II in which fusion of one or two intermediate spaces occurs [31].

Type II is characterised by the fusion of a few spinal segments, although it also includes the phenomenon of hemivertebrae and/or atlanto–occipital fusion. Klippel–Feil syndrome type II is probably dominant, while types I and III are recessive. Often, this syndrome is accompanied by other serious anomalies, such as atlanto–occipital fusion, basilar impression, spina bifida occulta, scoliosis, eight cervical vertebrae, Sprengel's deformity, hearing problems, deafness or deaf-mutism, hypodontia, renal anomalies, meningocele, neck pain and vascular, cardiac, central nervous system or skeletal system diseases [32].

Epidemiology on a global scale of KFS is difficult to determine due to the inherent difficulty in documenting cases and the consequent lack of studies identifying its true

prevalence. According to some authors, its incidence in the modern population is estimated at between 0.0025% and 0.5%, this means approximately 1:42,000 births [33].

The present case presents Klippel–Feil syndrome type II, and five anomalies were noted: deviated septum, cranial occipital symmetry, bilateral occipital condylar hypoplasia, spina bifida occulta type III and cervical dextroscoliosis. Type II occurs more frequently with a prevalence of 7.3/1000 [34,35]. Both sexes are equally affected [2,4].

Regarding treatment, most patients receive non-surgical management unless there is a neurological deficit, chronic neurological problems, cervical instability, in which case the recommendation is for surgical management. Overall, treatment is conservative and symptom driven. For patients with 1 or 2 level fusions below C3, monitoring and conservative management are sufficient. Those with a fusion above C3, particularly at the occiput, are more likely to be symptomatic and prone to the risk of spinal injury [36–40].

The syndrome is generally caused by mutations in the GDF6 or GDF3 genes, belonging to the family of bone morphogenetic proteins involved in regulating the growth and maturation of bone and cartilage; this condition is inherited as an autosomal dominant character, i.e., only one copy of the altered gene in each cell is sufficient to cause the disorder [41–44]. The robustness index and pilastric index of the femur were calculated in addition to macroscopic observation, detection of abnormalities and radiographic investigation that diagnosed the pathology under investigation [44].

The findings indicate muscle weakness in the thighs and confirm a reduced biomechanical activity of the individual during his life (Table 1).

Table 1. Anthropometric indices applied to the bones of the young Slavic soldier for the evaluation of aspects of the individual's biomechanical activity during his lifetime.

Femur Strength Index	**Result**
WEAK (X-12.5)	
STRONG (12.6-X)	INDEX 8
PILASTRIC FEMUR INDEX	
NULL (X-99) WEAK (100–109) MEDIUM (110–119) STRONG (120-X)	INDEX 102
TOTAL ANALYSIS	**WEAK**

These results play an important role in determining how much the syndrome may have affected the entire skeletal system, especially the activity of the lower limbs. In the present case, it is possible to affirm the presence of the symptoms in the living subject because the fusion of the C1 and C2 vertebrae did not allow a lateral rotation of the skull, with consequent cervical dextroscoliosis that inevitably caused an imbalance between skeletal and muscular development, particularly in puberty when bone growth is high.

6. Conclusions

The palaeopathological study of Klippel–Feil syndrome in anthropological archaeology is well known but underdeveloped, as there is no rich documentation to describe the origin of this pathology in past populations. This study investigated type II pathology and observed a number of related anomalies including deviated septum, cranial occipital symmetry, hypoplasia of the bilateral occipital condyles, spina bifida occulta type III and cervical dextroscoliosis. Considering the clinical presentation of the living subject, it is a mystery how he could have been part of a platoon of Slavic soldiers during the Second World War. Given the clinical condition of the living subject, it is speculated that he may have been a young, disabled volunteer who enlisted during the war.

7. Impact Statement

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