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A SERIOUS DEFECT OF TWO CERVICAL VERTEBRAE FROM A MEDIEVAL CEMETERY IN POLAND; KLIPPEL-FEIL SYNDROME?

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Abstract

A probable case of Klippel-Feil syndrome is presented along with a short review of studies on congenital fusion of cervical vertebrae in contemporary and ancient populations. Such disturbances are probably of either genetic or paragenetic origin and seem to be on the increase.

Key words: KLIPPEL-FEIL syndrome?, Middle Ages, Poland.

At a medieval cemetery (12th-13th c.) in Czarna Wielka in north-eastern Poland, about 250 human skeletons were unearthed together with some separate bones that proved difficult to ascribe to the skeletons from the site. Among the separates bones, two cervical vertebrae fused together were found; they were classified subsequently as C4 and C5 or possibly C5 and C6. They were fused by their articular processes and partially by their arches, and had their shafts underdeveloped. The shaft of the upper vertebra had only the poorly developed posterior part in the form of a plate of varying thickness, while the lower vertebra had it in its anterolateral portion. The vertebral foramen and the intervebral and transverse foramina were unchanged (Fig. 1).

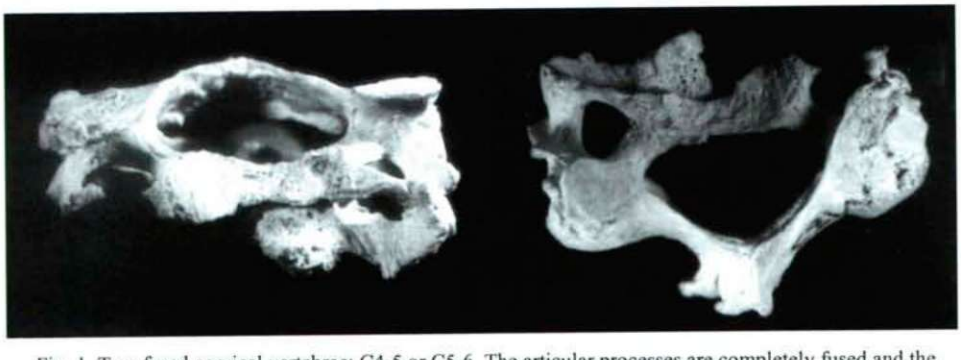

Fig. 1. Two fused cervical vertebrae: C4-5 or C5-6. The articular processes are completely fused and the arches only partially. The shafts are underdeveloped as incomplete plates: there is only an incomplete plate as the posterior part of C4 (or C5) and anterior-lateral (left) part of C5 (or C6).
a) top view, b) anterior-superior view.

Developmental disturbances of the skeleton are among those manifesting most commonly. In Poland, one newborn in every thousand is affected by this type of disorder, which generally occurs in the lower portion of the vertebral column (80%) (OSUCHOWSKI et al., 1974). GRAY et al. (1964) claim that the disorder appears more often in the cervical portion of the vertebral column. By 1961, 418 cases of this disorder had been reported in the available literature; the data seem to vary substantially, making the incidence of the defect difficult to determine accurately. Interestingly, fusion of the cervical vertebrae has also been found in animals.

Classification of developmental disorders of the cervical vertebrae is difficult, since the condition is symptomless in many of its forms, or manifests most often in patients over 40-50 years of age and is frequently accompanied by widely varying clinical symptoms (OSUCHOWSKI et al., 1974; ELSTER, 1984; PRUSICK et al., 1985).

Several forms of cervical segment developmental disorders have been distiguished: 1) atlanto-occipital vertebra fusion, 2) KLIPPEL-FEIL syndrome, 3) odontoid process deformation, 4) spina bifida, 5) hemivertebrae (ZAKRZEWSKI, 1956; KOZINA et al., 1965; PRUSEK et al., 1972).

The KLIPPEL-FEIL syndrome is the most frequently described such defect it can be found, among others, in the works of ZAKRZEWSKI (1956), KOZINA et al. (1965), GUNDERSON et al. (1969), TRAUENFELLNER (1968), MCLAY and MARAN (1969), BAGA et al. (1969), RASSUMOWSKA and MALINOWSKI (1970), PRUSEK et al. (1971), RAMSEY and BLIZNAK (1971), PALANT and CARTER (1972), JUZWA et al. (1973), ROZENBLAT and UTZIG (1973), PRUSZEWICZ and OTREMBOWSKI (1974), HENZINGER et al. (1974), MOORE et al. (1975), and in my other studies cited in this paper.

According to MCLAY and MARAN (1969) and their citation of the work of P. A. and L. GJØRUP from 1964, the condition of a congenital cervical vertebra anomaly was first indicated by HALLER following a post mortem examination in 1745. Only one year later, MORGAGNI discovered in an old man an atlanto-occipital fusion, together with one between the atlas and the third vertebra, and the condition was further characterized by some deformation of ligaments of the occipitovertebral joints (GRAY et al., 1964). The latter source also cites others who described this defect in a newborn (GOODHARD, 1874), in a dwarf (Varaglia), in a patient with the concurrent defect of SPRENGEL (HITSCHINSON, 1894), and in a woman who also had the seventh cervical vertebra fused with the first thoracic vertebra (WILLETT and WALSHAM, 1880; cited by GRAY et al., 1964).

The first cases of the condition in living persons were reported in 1902 by SICK, and by CLARK in 1906, and then other cases gradually became known (GRAY et al., 1964). In 1912, KLIPPEL and FAIL published several articles with a detailed description of cervical vertebra defects in a 46-year-old man. The outstanding feature in the patients was a very short, almost immobile neck, with prominent wing-like soft tissue on both sides thereof (pterigium colli) and a low-lying posterior hairline (MCLAY and MARAN, 1969; PALANT and CARTER, 1972). In 1919, FEIL presented 14 cases of the congenital deformation of cervical vertebrae and categorized the condition into three types:

Type I: a block fusion of all the cervical and upper thoracic vertebrae;

Type II: a fusion of one or two pairs of cervical vertebrae, frequently C1 to C3, or C3 to C4, and accompanied by hemivertebrae, occipito-atlantis;

Type III: combines the anomalies of type I or II with either lower thoracic or lumbar intervertebral fusion (PALANT and CARTER, 1972; MOORE et al., 1975).

In 1921, DUBREUIL-CHAMBARDEL first used the now familiar eponym of KLIPPEL-FEIL.

The classic triad of KLIPPEL-FEIL syndrome features has become known under various synonymous labels: cervical synostosis, congenital brevicollis, Kurzhals, l'homme sans cou, Froschhals (TRAUNFELLENER, 1968; URUNUELA and ALVAREZ, 1994).

Among the material of 418 cases presented by GRAY et al. (1964), the most frequent defect was of type II, block C2-3, and less commonly that of C6-7. The KLIPPEL-FEIL syndrome was equally common in both sexes, and in several cases manifested in the family, e. g. in two brothers.

Congenital deformation of the cervical vertebrae can be accompanied by other defects within the same system. In the bone structure, the concurrence of the following deformations have been discovered: skull and face asymmetry, cleft palate, abnormal development of the inner ear, basilar impression, dental anomalies, micrognathia, atlanto-occipital fusion, deformation of the odontoid process, torticollis, spina bifida, narrowness of the medullar channel, stenosis of the intervertebral foramen, SPRENGEL deformity, cervical ribs, fused ribs, and scapula-vertebral column union (RASSUMOWSKA and MALINOWSKI, 1970; RAMSEY and BLIZNAK, 1971; PALANT and CARTER, 1972; JUZWA et al., 1973; OSUCHOWSKI et al., 1973; PRUSEWICZ and OTRĘBOWSKI, 1974; MOORE et al., 1975; ELSTER, 1984; PRUSICK et al., 1985).

The etiology of developmental defects of the vertebral column is not yet fully known. Most likely, the process takes place in the first weeks of embryonic life, when the developmental differentiation of the blastodermic layers and the isolation of the proto-segments or somites occur, which then differentiate into dermatomes and sklerotomes. The latter surround the notochord and the medullary tube, and thus constitute the mesenchymal buds of the vertebrae. The defective developmental factor is most probably genetically or epigenetically determined (KOZINA et al., 1965; HILLER, 1968).

FEIL pointed out that intrauterine injuries and endometritis may provoke irregular segmentation in the first weeks of intrauterine life (DEMJÉN and MARCINKOVÁ, 1965). According to STRAX and BARAN (1975) (cited by URUNUELA and ALVAREZ, 1994), type II may be inherited as a dominant or recessive trait, depending on the vertebrae affected, while type I and type III are apparently of a recessive nature.

In bone material from ancient cemeteries, only a few cases of KLIPPEL-FEIL syndrome have been recorded. The first cervical block was reported by SMITH in an Egyptian mummy dating back to about 500 B. C. (GRAY et al., 1964). In 1923, MCCURDY described another case of the defect, which was classified by JARCHO as KLIPPEL-FEIL syndrome only in 1965. It was observed in an adult man from Poricarcancha in Peru. C6-7 and Th1 were fused, and within the thoracic vertebrae Th3-4-5 were hemivertebrae; on the right, there were 13 ribs, of which the first four were fused at their proximal ends (ORTNER and PUTSCHAR, 1981). A third case, involving the fusion of five cervical vertebrae, was described by UBELAKER in 1978; whose material

came from a local cemetery in Modbridge Side, South Dakota. Two cases dating back to pre-Columbian times were discovered in Tanacah, Quintana Roo, and described by SAUL in 1982. Both presented a fusion of the 2nd and 3rd cervical vertebrae (ORTNER and PUTSCHAR, 1981).

In 1994, a very rare case of type I KLIPPEL-FEIL syndrome was described by URUNUELA and ALVAREZ, who presented material dating back to 1450-80 from pre-Hispanic Cholula, Puebla, Mexico. The cases revealed a complete vertebral fusion from the second cervical to the first thoracic vertebrae, and the hypoplastic development of vertebral bodies. The investigated skeleton came from a 30-40-year-old woman, whose other bones presented other anomalies.

The case discovered in Czarna Wielka seems to be the first dating back to the Old World and probably represents type II KLIPPEL-FEIL syndrome, though the diagnosis may not prove absolutely accurate, as it is based only on the presence of developmental defects in two vertebrae (no other bones remained). A differentiation must be made between this defect and the neck deformation resulting from a vertebral column injury, sub-occipital POTT's disease and NELSON syndrome (MOLDENHAUER, 1964; DEMJÉN and MARCINKOVÁ, 1965).

The sporadic incidence of KLIPPEL-FEIL syndrome in the material from ancient cemeteries and the relatively high occurrence of the condition in our contemporary material (e. g. 1:1000) seem to suggest that the factor responsible for the defect has manifested so broadly only in modern times.

Interestingly, in the Polish material obtained from cemeteries such a dramatic developmental defect was discovered in a medieval burial ground, where the incidences of variation and of the defects themselves are particularly high vis a vis the revelant data from other cemeteries, including those from the Neolith (GLADYKOWSKA-RZECZYCKA,1980, 1989).

Conclusion

A rare case of a congenital cervical vertebra defect was discovered in the material from a medieval burial ground. Vertebrae C4-5 or C5-6 constitute a block and exhibit an underdeveloped shaft in the form of rudimentary plates. This is the first defect of this kind that has been reported from ancient Polish cemeteries so far. Other disturbances of the kind have come exclusively from the New World. A short review of studies on KLIPPEL-FEIL syndrome indicates that the defect occurs relatively often in contemporary Poland (1:1000).

References

BAGA, N., CHUSID, E. L. and MILLER, A. (1969): Pulmonary disability in the KLIPPEL-FEIL Syndrome. - Clin. Orthop. and Related Research *67*, 105-110.

DEMJÉN, Š. and MARCINKOVÁ, V. (1965): KLIPPEL-FEIL syndrome and cleft palate. - Acta Chirurg. Plastic. *7*, 297-302.

ELSTER, A . D. (1984): Quadriplegia after minor trauma in the Klippel-Feil syndrome. - J. Bone Joint Surg. *66 A (9)*, 1473-1474.

GLADYKOWSKA-RZECZYCKA J. (1980): Developmental disturbances in the skeletal material from ancient Polish cemeteries. - Przegl. Antrop. *46*, 347-362 (in Polish).

GLADYKOWSKA-RZECZYCKA, J. (1989): Congenital anomalies of early Polish man. - Advances in paleopathology, Monographic publication *1*, - J. Paleopath., 111-116.

GRAY, S. W., ROMAINE, C. B., and SKANDALAKIS, J. E. (1964): Congenital fusion of the cervical vertebrae. - Surg. Gynec. Obst. *118*, 373-385.

GUNDERSON, C. H., GREENSPAN, R. H, GLASER, G. H. and LUBS, H. A. (1969): The KLIPPEL-FEIL syndrome: Genetic and clinical reevaluation of cervical fusion. - Medicine *46*, 491-512.

HENSINGER, R. N., LANG, J. F. and MAC EWEN S. D. (1974): KLIPPEL-FEIL syndrome, a constellation of associated anomalies. -J. Bone Joint Surg. *56 a (6)*, 1246-1253.

HILLER, S. (1968): Histology and embryology. In: BOCHENEK A., REICHER, M. (eds): Human Anatomy. - Warsaw, 55-155 pp, (in Polish).

JUZWA, E. and WINIARSKA-MAJCZYNO, M. (1973): Facial asymmetry, disturbances in masticatory organ and maxillo-orthopedic management of KLIPPEL-FEIL syndrome in children. - Czas. Stom. *26*, 10, 1089-1094 (in Polish).

KOZINA, W., SAPER, J. and LEO, W. (1965): Remarks on neurologic disturbances and on X-ray diagnosis of congenital malformations of the cervical spine. - Rheumatol. *3*, 2, 135-145 (in Polish).

MCLAY, K. and MARAN, A. G. D. (1969): Deafness and KLIPPEL-FEIL syndrome. - J. Laryng. Otology *83*, 175-184.

MOLDENHAUER, E. (1964): Zur Klinik des NIELSON-Syndrome. -Dermatol. Wochenschrift *150*, 594-601.

MOORE, W., MATTHEWS, T. J. and RABINOWITZ R. (1975): Genitourinary anomalies associated with KLIPPEL-FEIL syndrome. - J. Bone Joint Surg. *57 A*, 355-357.

ORTNER, D. J., and PUTSCHAR, W. G. J. (1981): Identification of pathological condition in human skeletal remains. - Smithsonian Institution Press, Washington, Skeletal malformations, 346 pp.

OSUCHOWSKI, K., AUGUSTYŃSKI, W. and BŁACH, A. (1974): KLIPPEL-FEIL syndrome. -Wiad. Lek. *27*, 21, 1881-1883 (in Polish).

PALANT, D., I. and CARTER, B. I. (1972): KLIPPEL-FEIL syndrome and deafness. - Am. J. Diseas. Children *123*, 3, 218-221.

PRUSEK, W., CHILMAN, A. and NAWROCKA, E. (1972): Development anomalies of cervical spine. - Pol. Tyg. Lek. *46*, 1809-1813 (in Polish).

PRUSICK, V. R., SAMBERG, I. C. and WESOLOWSKI, D. P. (1985): KLIPPEL-FEIL syndrome associated with spinal stenosis. - J. Bone Joint Surg. *67A (1)*, 161-164.

PRUSZEWICZ, A. and OBRĘBOWSKI, A. (1974): KLIPPEL-FEIL syndrome with coexistent submucosal cleft palate and hypoacusia. - Czas. Stomat. *27*, 3, 251-255 (in Polish).

RAMSEY, J. and BLIZNAK, J. (1971): KLIPPEL-FEIL syndrome with renal agenesis and other anomalies. - Am. J. Roentg. Radium Therapy Nuclear Med. *113*, 3, 460-463.

RASSUMOWSKA, D. and MALINOWSKI, A. (1970): Anthropomorphological characteristics of KLIPPEL-FEIL syndrome with cleft palate. - Czas. Stomat. *23*, 9, 1075-1081 (in Polish).

ROZENBLAT, J. and UTZIG A. (1973): KLIPPEL-FEIL syndrome associated with multiple developmental anomalies. - Pol. Tyg. Lek. *28*, 22, 832-833 (in Polish).

TRAUNFELLNER, Z. (1968): Case of KLIPPEL-FEIL syndrome. - Wiad. Lek. *21*, 6, 521-522 (in Polish).

UBELAKER, D. H. (1978): Human skeletal remains: excavation, analysis, interpretation. - Aldine Publishing Company, Chicago.

URUNUELA, G. and ALVAREZ R. (1994): A report of KLIPPEL-FEIL syndrome in pre-Hispanic remains from Cholula, Puebla, Mexico. - J. Paleopath. *6(1)*, 63-67.

ZAKRZEWSKI, T. (1956): Case of congenital block of cervical vertebrae (KLIPPEL-FEIL syndrome). - Pol. Tyg. Lek. *11*, 842-44 (in Polish).