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CASE REPORT

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Klippel-Feil anomaly with associated rudimentary cervical ribs in a human skeleton: case report and review of the literature

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Anomalies of the cervical spine are intriguing anatomically and often come to clinical attention. Fusion of one or more cervical vertebral segments, the Klippel-Feil anomaly (KFA), often causes increased motion at the vertebral segments superior to and inferior to the fused level with a resultant premature wear of these joints. We report an adult male skeleton with fusion of his C6 and C7 vertebral bodies (Type II KFA). A remnant of the intervertebral disc space was noted and bilateral rudimentary cervical ribs were observed emanating from the C7 vertebrae. Excessive joint degeneration was noted between the vertebral bodies of C5 and C6. Following our review of the literature and case report, it appears that there is an increased incidence of the presence of cervical ribs in KFA. We review the literature for coexistent KFA and cervical ribs and discuss their dysembryology.

Key words: anatomy, spine, congenital, anomaly, fusion, cervical

INTRODUCTION

In 1912 Maurice Klippel and Andre Feil were the first to describe congenital fusion of the cervical spine, the Klippel-Feil anomaly (KFA) [7]. However, congenital fusion of the cervical spine was first noted in an Egyptian mummy (circa 500 BC.) in 1908 [13]. It has been noted that patients with KFA often have the triad of a short neck, a decreased range of motion (ROM) in the cervical spine and a low hairline. Fewer than 50% of patients have all three elements of this triad and, therefore, in its current usage the term KFA refers to congenital fusion of the cervical vertebrae [13]. Subsequently KFA has been classified into 3 types. Type I is described as a massive fusion of the cervical spine. Type II is present when the fusion of one or two vertebrae occurs. Type III occurs when thoracic and lumbar spine anomalies are associated with type I or type II KFA. Raas-Rothschild et al. [14] have recently introduced the Type IV KFA, which includes Types I–III combined with sacral agenesis. The most commonly fused vertebral level in the KFA is that of the C5 and C6 vertebrae. The aetiology of KFA and its associated conditions is currently unknown.

CASE REPORT

We report two unusual findings in an adult male skeleton found in our anatomy laboratory at the University of Alabama at Birmingham. No other osteological anomalies such as scoliosis or Sprengel's deformity were observed. Congenital fusion was found between the sixth and seventh cervical vertebrae (Fig. 1). Specifically, the fusion was between the bodies and zygapophyseal joints. No fusion was noted between the posterior elements of these two vertebrae. A rudimentary disc space was seen anteriorly and measured 1 mm in height and 2.5 cm in width (Figs. 1, 2). Additionally, rudimentary cervical ribs

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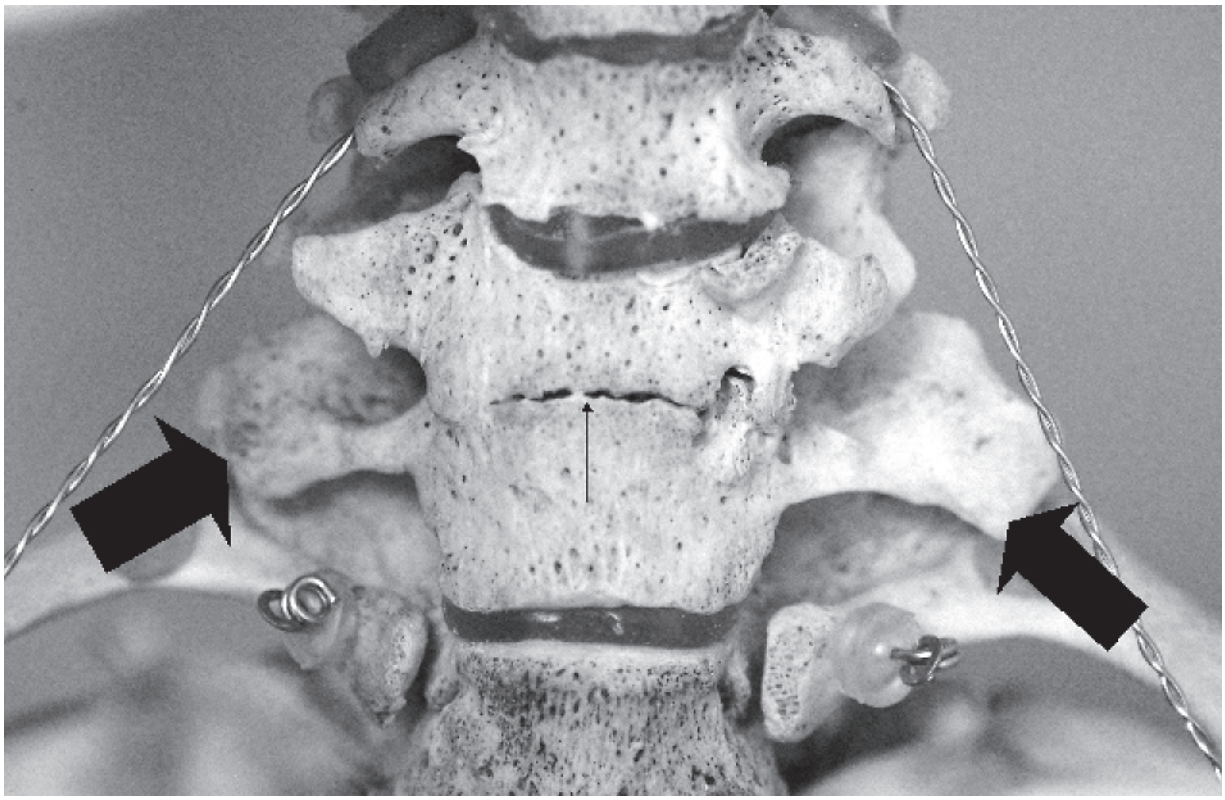

Figure 1. Anterior view of our skeletal specimen noting the fused cervical vertebral Segment (centre of photo between vertebral bodies C6 and C7) with a rudimentary intervertebral disc space. Note the rudimentary cervical ribs associated with C7.

Figure 2. Magnified view of Figure 1. Note the osteophyte formation between C5 and C6 (left side).

were seen on the left and right sides (the left larger than right) of the seventh cervical vertebrae (Fig. 1). Increased arthritic change (i.e. osteophyte formation) was noted between the vertebral bodies of C5 and C6 (Fig. 2).

DISCUSSION

Patients with KFA usually present during childhood but may present later in life. Patients presenting with upper cervical spine involvement tend to present clinically at an earlier age than those whose involvement is lower in the cervical spine, but irrespective of this a decreased range of motion of the cervical spine is the most frequent clinical finding. Rotational loss is usually more pronounced than is the loss of flexion and extension of the cervical spine.

The true incidence of KFA is unknown as no one has yet studied a cross-section of healthy people to determine this. However, Gjorup and Gjorup [5] reviewed all the radiographic cervical spine films in a single hospital in Copenhagen. From these radiographs they determined an incidence of 0.2 cases per 1000 people.

Neurological problems may develop in 20% of patients [13]. Occipitocervical abnormalities were the most common cause of neurological problems. Synkinesia (mirror movement of the upper or lower extremities) is often seen. Patients with this finding are, for example, unable to perform a conscious movement of the right hand without performing the same movement of the left hand subconsciously.

One hypothesis is that mirror movements are due to accessory corticospinal tracts [16]. KFA may be seen more commonly in patients with Chiari I malformation (i.e. cerebellar tonsillar ectopia through the foramen magnum) [18].

Aside from cervical vertebral fusion, other orthopaedic findings in KFA include scoliosis, which was not seen in our specimen and occurs in approximately 60% of patients [6]. In some patients this is congenital scoliosis due to the involvement of other parts of the thoracic or lumbar spine. Other patients develop compensatory scoliosis in the thoracic spine to compensate for their cervical or cervicothoracic scoliosis. In addition to the fusion anomalies in the cervical spine, cervical spinal stenosis can occur. As seen in our specimen, rudimentary disc spaces may be observed (Fig. 1) [13]. Sprengel's deformity (a small elevated dysmorphic scapula) occurs in 20––30% of patients. An omovertebral bone, which is an osteocartilaginous connection between the scapula and spine, may tether the scapula to the vertebral column, thus limiting function. An omovertebral bone may ossify with age, thus further limiting the ROM.

Fusion between adjacent vertebral segments can also be formed iatrogenically. The surgical technique commonly used in the cervical region (anterior cervical fusion with discectomy) most often utilises metal hardware (such as plates) for a secure construction between unstable vertebral segments. Posterior fusion between adjacent cervical vertebrae can also be created with techniques such as sublaminar wiring and/or removal of the cartilage from the zygapophyseal joints. No such intervention was seen in our specimen.

Although KFA is defined by its skeletal components, other systems commonly affected in patients with KFA include the renal, auditory, and cardiovascular systems. Renal anomalies are common in individuals with KFA. Hensinger et al. [6] reviewed 50 patients and found renal anomalies in 34%. These included a double collecting system, renal ectopia, hydronephrosis and a horseshoe kidney. Hearing loss is common with KFA. The hearing loss can be sensorineural, conductive or mixed. Cardiovascular anomalies occur in 14–29% of cases. The most common cardiovascular defect is an interventricular septal defect. Other less common anomalies are congenital limb deficiencies and craniosynostosis.

No proven theory exists to explain the development of congenital fusion of the cervical vertebrae. However, similar changes have been produced in animals with teratogenic agents [13]. Some have speculated that bilateral failure in the development of the fissure of von Ebner halts development of the spine prior to the stage of resegmentation, with the resultant vertebral bodies appearing to be fused with the primitive disc formation at appropriate levels, since normal somite segmentation has been completed [13]. C5–C6 vertebral fusion may be recessively inherited [4]. Lubs et al. [9] found 2 of 11 siblings with this type of fusion and a third with narrowing of the cervical intervertebral spaces. Ohashi et al. [11] suggested that one of the genes responsible for KFA may be located at 5q11.2 or 17q23. Of course, it is always possible in the case of such translocations that there is the creation of a fusion gene that is responsible for the abnormality of development. More recently Smith and Tuan [17], using in situ hybridisation, have observed that HuP48 (developmental gene of the PAX family) is expressed in a segmented pattern in the developing spine, specifically in the cells of the intervertebral discs of seven to eight-week-old foetuses. Further, these authors found that at this early stage in development, HuP48 expression appeared to be restricted to the vertebral column. These authors concluded that the pattern of expression of HuP48 indicates that it may play a role in establishing the segmented pattern of the vertebral column, perhaps by helping to establish and/or maintain a border between two different populations of cells, those of the prevertebrae and those of the intervertebral discs.

The treatment for KFA ranges from simple administration of oral anti-inflammatory agents for arthritic pain to surgical stabilisation of degenerative and hypermobile cervical vertebrae. Although the literature is conflicting, some have advocated cardiac, renal, and auditory tests to evaluate for the possible dysfunction of these systems [13].

Our male specimen also exhibited rudimentary cervical ribs, which have been rarely reported in patients with KFA [8, 12, 19–21]. A cervical rib arises from the transverse process of the seventh cervical vertebra (or rarely the sixth and very rarely the fifth). The rib reflects hyperplasia of the transverse process secondary centre of ossification. It is more common in females, occurring in 11% of the general population, and is slightly more common on the right side. Occasionally it is just a fibrous band or a complete rib with articulation with the manubrium or the first rib. The transverse processes of the cervical vertebrae slope inferiorly from the neural arch, whereas they slope superiorly in the thoracic region. This anatomical difference helps to differentiate between a cervical rib

and a rudimentary first rib on radiographs. Cervical ribs occur either unilaterally or bilaterally. They vary greatly in size and shape, and the clinical symptoms that may be exhibited have very little relationship to the size and shape of the rib. A fibrous band of varying thickness and width is anatomically bound to the cervical rib in a plane parallel to the scalenus muscles. Compression of the adjacent neurovascular bundle can cause symptoms. Van Kerckhoven and Fabry [19] evaluated 18 patients with KFA and found that one patient had a concomitant cervical rib unilaterally. Likewise, Yiannikas and Walsh [21] studied 11 patients with cervical ribs and identified one patient with KFA.

An increased rate of cervical rib formation has been found in animals treated with Vigabatrin [1], butanol [10], boric acid [3] and retinoic acid [15]. Interestingly, Charite et al. [2] have shown in an animal model that when Hoxb-8 was ectopically expressed, cervical vertebrae can be made to take on characteristics of thoracic vertebrae such as rib formation.

Following our review of the literature and case report it appears that there is an increased incidence of the presence of cervical ribs in KFA. This knowledge may alert the clinician to observe for both these entities in patients that present with either of these diagnoses.

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