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Case Report

X-Ray Anatomical Features of Klippel-Feil Syndrome

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Received: October 03, 2019; **Published:** December 31, 2019

Abstract

A man born in 1990 was submitted to the University clinic with complaints of weakness in his legs and uncertainty when walking. A CT of the brain and cervical spine was performed. Convincing data on the presence of pathological changes in brain tissue density during the study was not obtained. The study of the cervical spine on multiplanar reconstructions of CT images in the axial, sagittal and frontal projections revealed a number of anomalies, which were subjected to careful study and description.

Keywords: *Klippel-Feil Syndrome; Malformation; Cervical Vertebrae*

Introduction

Klippel-Feil syndrome (KFS) is a genetically determined abnormality of the structure of the cervical spine, consisting in a decrease in the number of cervical vertebrae due to synostosis. A typical and constant feature of this syndrome is pronounced shortening of the neck [1,2].

Case Report

A man born in 1990 was submitted to the University clinic with complaints of weakness in his legs and uncertainty when walking. A CT of the brain and cervical spine was performed. CT of the cervical spine had revealed the following anatomical changes: dens cleavage of C2 vertebra, complete fusion of bodies, arcs and articular processes of C2 - C3 vertebrae. The body of the C1 vertebra is fragmented and there are diastases of anterior and posterior arcs (5.4 mm and 6.72 mm respectively), lateral masses are displaced laterally, the left one was underdeveloped. Cervical lordosis is straightened. The patient was diagnosed with Klippel-Feil syndrome with anatomical changes of the structure of the cervical spine belonging to type II.

In the axial projection of the CT image, the odontoid process of the C2 vertebra is located in the cavity of the posterior cranial fossa. Vertical size (height) of the odontoid process is 18.6 mm, the transverse size of the tooth is 8.8 mm at the top and is increased up to 17.5 mm at the base. In the sagittal projection of the CT image, the odontoid process is above the foramen magnum, protruding into the cavity of the posterior cranial fossa up to 7.11 mm. The longitudinal size of the foramen magnum is 37.9 mm, the transverse - 30,1 mm.

The C2 vertebra's odontoid process connects to the body with the help of a joint, the articular cavity of which is 1.7 mm. The bodies of the C2 and C3 vertebrae fused together, the height of this fusion is 26.6 mm. The anteroposterior size of the spliced bodies of the C2 and C3 vertebrae was measured at three points on the sagittal section of the CT image: at the upper edge of the body - 13.42 mm, in the

middle -14.19 mm and at the lower edge -18.71 mm. The foramen of the transverse processes of the fused vertebrae had the following dimensions: left - 5,89*6,23 mm, right - 5.28 x 5,63 mm.

The structure of C4 - C7 vertebrae was not changed. The height of the C4 - C7 vertebral bodies increased evenly from 9.7 mm (C4) to 11.7 mm (C7). Intervertebral disc height decreased evenly from 6.9 mm (between C3 - C4) to 4.8 mm (between C6 - C7).

The first rib on both sides was in its infancy.

Discussion

In 1912, Maurice Klippel and Andre Feil independently provided the first descriptions of KFS. They described patients who had a short, webbed neck; decreased range of motion in the cervical spine; and a low hairline.

Mutations of the GDF6, GDF3 and MEOX1 gene are associated with KFS [3-5]. The prevalence of KFS is unknown due to the lack of studies to determine its prevalence [6,7]. It is estimated to occur 1 in 40,000 to 42,000 newborns worldwide [8]. In addition, females seem to be affected slightly more often than males [9-11].

Feil subsequently classified the syndrome into three categories:

1. Type I - fusion of C2 and C3 with occipitalization of the atlas. Flexion and extension is concentrated within the C1 and C2 vertebrae. As with aging, the odontoid process can become hypermobile, narrowing the space where the spinal cord and brain stem travel (spinal stenosis).
2. Type II - long fusion below C2 with an abnormal occipital-cervical junction. Flexion, extension, and rotation are all concentrated in the area of an abnormal odontoid process or poorly developed ring of C1, which cannot withstand the effects of aging.
3. Type III - a single open interspace between two fused segments. Cervical spine motion is concentrated at single open articulation. This hypermobility may lead to instability or degenerative osteoarthritis. This pattern can be recognized as the cervical spine is often seen to be at an angle or hinge at this open segment [12].

In most of cases, the syndrome is sporadic, there is evidence of its genetic heterogeneity, for example, type II is inherited autosomal dominant, and type III is autosomal recessive [8,13].

Congenital mergers can occur at any level of the cervical spine, although 75% occur in the first three cervical vertebrae [9,13]. The most common fusion occurs between C2 and C3 [4,10]. KFS is often associated with skeletal malformations such as scoliosis, facial asymmetry, cleft palate, rib anomalies, polydactyly [14]. In 45% of cases, hypoplasia and dystopia of the kidneys are observed, in 25% - deafness, 15% - heart defects.

The heterogeneity of KFS has made it difficult to outline the diagnosis as well as the prognosis for this disease. However, the diagnosis of the syndrome is based on a triad of clinical symptoms: shortening of the neck, observed from birth, low hairline and limited mobility of the head.

Conclusion

The described x-ray anatomical picture of the KFS type II includes complete fusion of C2 - C3 vertebrae, cleavage of the arches of the C1 vertebra and non-fusion of the body and odontoid process of C2, underdevelopment of the first ribs that expands the morphometric, topographic and x-ray anatomical characteristics of the cervical spine.

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Volume 3 Issue 1 January 2020