

Congenital fusion of cervical vertebrae and its clinical significance - A case report

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Abstract

Skeletal abnormalities of cervical region or in cranio-cervical region are of interest to the anatomists, orthopaedicians, neurologists, neurosurgeons and even orthodontists. These abnormalities may result in severe neck pain, decreased neck mobility, muscular weakness of both upper limbs, sensory deficits on arms and hands etc. In condition of fusion of the cervical vertebrae, two vertebrae appear not only structurally as one but also function as one.

Keywords: Cranio-cervical, Fusion, Cervical Vertebrae.

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CASE REPORT

A healthy 14-year-girl complained of torticollis. Her medical and family histories were unremarkable. Physical examination reveals Pain on palpation over vertebral spines and neck muscles. There is limited neck extension and rotation. Neurological examination revealed normal cranial nerve function. The muscle strength was grade 4 over limbs without muscle atrophy. Nerve associated anomalies were noted.



Figure 1: Shows absence of C1 vertebrae and fusion of second and third cervical vertebrae



Figure 2: Shows fusion of C2 and C3 vertebrae in open mouth view

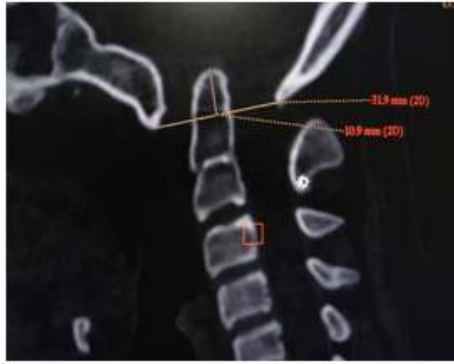


Figure 3: Superior migration of odontoid process 3.2cm above the lower margin of foramen magnum



Figure 4: Shows presence of Foramen transversarium



Figure 5: Shows Absence of C1 and fusion of axis and third cervical vertebral body and lamina



Figure 6: Shows Indentation of cervico-medullary junction

Fig -1 shows absence of C1 vertebrae and fusion of second and third cervical vertebrae. Fig – 2 shows fusion of C2 and C3 vertebrae in open mouth view. Fig – 1 Superior migration of odontoid process 3.2cm above the lower margin of foramen magnum. Fig – 4 shows presence of Foramen transversarium. Fig – 5 shows Absence of C1 and fusion of axis and third cervical vertebral body and lamina. Fig - 6 shows Indentation of cervico-medullary junction.

DISCUSSION

The spinal column is the central structure in the vertebrate body which provides stability, posture and initiates all the movements. Formation of spine during embryonic life is a highly complex and regulated process. If disrupted, can lead to a variety of congenital anomalies including block vertebra, hemivertebra etc.¹ For many years anomalies of cervical region were of interest mainly to anatomists. But these are also of great importance to orthopaedicians, neurologists, neurosurgeons, anaesthesists, and even orthodontists. In condition of fusion of the cervical vertebrae (FCV), two vertebrae appear not only structurally as one but also function as one. This fusion may be congenital (CFCV) or acquired. This anomaly

may be asymptomatic; however, it may also appear with manifestations of serious clinical features such as myelopathy or may be associated with syndromes such as Klippel-feil limitation of the neck movement or the muscular weakness, atrophy and neurological sensory loss.² Congenital anomalies are common at craniovertebral or cervical region.³ Among these congenital anomalies the important ones are the fused cervical vertebrae (FCV). Congenital fusion of axis with the third cervical vertebrae limits the movements between these bones and because of this the third vertebrae was given the name as “*vertebrae critica*”.⁴ Severe neck pain and sudden unexpected death may occur due to these abnormalities.⁵ Cervical vertebrae are seven in number. C3-6 are typical whereas C1 (atlas), C2 (axis) and C7 (cervicae prominens) are atypical. C2 vertebrae is different from other by the presence of Dens (odontoid process), which projects cranially from the superior surface of the body. The axis acts as an axle for rotation of atlas and head around the dens.⁶ The fusion may be congenital due to failure of segmentation of sclerotomes at certain levels or may be acquired due to a number of other causes like tuberculosis, juvenile rheumatoid arthritis or trauma etc. Congenital anomalies like Klippel

feil syndrome, fetal alcohol syndrome are associated with vertebral fusion. 75% of vertebral fusions occur in cervical region.⁷ Congenital anterior fusion of vertebrae is usually asymptomatic. Awareness of this anomaly is important for correct diagnosis.⁸ During development of vertebrae re-segmentation is very important. Inappropriate vertebral fusion results in anomalous vertebral synostosis or spinal fusion.⁹ The etiology of fusion may be congenital, acquired or surgical. Congenital Cervical vertebral fusion leads to decrease in length of spine, prominent trapezei, webbed neck, lowered hair line, signs of peripheral nerve compression. Congenital fusion of vertebrae is most commonly seen in the cervical region. The incidence of CFCV of C2-C3 is around 0.4% to 0.7% with no sex predilection.¹⁰ According to the frequency of block vertebrae, the order is C2-C3, C5-C6, L4-L5 and thoracic spine (any segment).¹¹ Fused cervical vertebrae (FCV) have clinical and embryological importance. In FCV, the fusion may be either congenital or acquired. Congenital FCV is one of primary malformations of chorda dorsalis believed to be due to defects which take place during the development of the occipital and cervical somites. Cause of this anomaly is often a combination of environment and genetics which occurs during the 3rd week postconception. Although radiologic appearance of FCV has a characteristic feature, its precise diagnosis is complex, particularly, among young cases. It is because ossification of the vertebral body is not complete till adolescence and cartilage which has not ossified, may appear like a normal disc area.² Acquired FCV is generally associated with diseases like tuberculosis, other infections, juvenile rheumatoid arthritis and trauma.¹¹ All these abnormalities may lead to shortening of spine in the cervical region, trapezei are unduly prominent laterally and give a webbed appearance, limited neck motion, osseous malformation (scoliosis, kyphosis, torticollis). Sign and symptoms of peripheral nerve irritation and compression appears such as pain, burning sensations, cramps, hypoaesthesia/ anaesthesia, weakness/paralysis, fibrillations and reduced deep reflexes.⁵ These signs and symptoms are similar with that of Klippel-feil syndrome (congenital fusion of cervical vertebrae, brevicollis). This is a clinical condition of congenitally fused and deformed cervical vertebrae that results in restricted neck motion and neurologic phenomena. Based on clinic-radiological features, Kleippel – feil syndrome is classified into:¹¹

Type 1: Fusion of cervical and upper thoracic vertebrae with synostosis accounting for 40% cases.

Type 2: Isolated cervical spine fusion accounting for 47% cases.

Type 3: Cervical vertebrae with lower thoracic or upper vertebral fusion accounting for 13% cases.

The presence of block vertebrae results in more biomechanical stress in the adjoining segments leading to more degenerative changes. The other changes are rupture of ligaments (mainly transverse ligaments), tear of intervertebral disc resulting in herniation of nucleus pulposus resulting in compression of spinal cord, fracture of odontoid process and spondylosis.¹⁰ The radiologic appearance of congenital anomalies may be due to defects in fusion or normal segmentation, occipitalization of the atlas, odontoid and atlas malformations, spina bifida and abnormal ossifications. In about 70% of occipitalizations instability at the C1-C2 articulation is associated with fusion of C2-C3.¹⁰ In vivo, it is easy to diagnose the existence of the FCV by plain x-ray, but important point is to be able to distinguish between a pathologic condition and whether it is CFCV or acquired.¹² If these anomalies are diagnosed early, they will help us in finding the change due to an injury, ageing or progression of a degenerative process and also motivates the patient to change their lifestyles to lead a normal life. These anomalies like block vertebrae cannot be treated surgically as they carry a higher mortality and morbidity. Cervical vertebrae fusions are sometimes done following traumatic, degenerative and inflammatory dislocations of upper and lower cervical vertebrae.¹³ The most important differential diagnosis in CFCV is decreased AP diameter of the vertebra, and individual measurements of the two vertebrae's bodies height is equal to the two fused vertebrae's height including the intervertebral disc. There are calcifications and atrophic appearance in intervertebral disc on x-ray or MRI.¹⁴

Embryological Significance

Somites are formed from para-axial mesoderm that lie on each side of neural tube. The somites are divided into three parts: Ventromedial sclerotome; Intermediate myotome; and Lateral dermatome. The vertebral column is formed from the sclerotome of the somites. Normal segmentation of the sclerotomes is important for the development of a normal vertebral column. But in certain cases due to decreased local blood supply during the third to eight week i.e. embryonic period results in abnormal segmentation and formation of congenitally fused vertebrae or block vertebrae. Vertebral fusion anomalies are likely to be associated with disturbance of Pax-1 gene expression in the developing vertebral column.¹⁵

Clinical Significance

While doing endotracheal intubation, extension of the neck is done. So in persons with block vertebrae in cervical region we have to take care to prevent hyperextension as it can precipitate disc prolapsed. If cisternal puncture or lumbar puncture is to be done, we should look for possibility of block vertebrae in cervical and lumbar regions respectively.

CONCLUSION

We insist that clinical heterogeneity and radiographic abnormalities found in cervical vertebrae may simulate acute pathology and thus require comprehensive evaluation and definitive diagnostic and appropriate management.

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