

# A Rare Presentation of Os Odontoideum with Multiple Vertebral Fusion in Type III Klippel-Feil Syndrome (KFS) – A Case Report

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## ABSTRACT

An abnormal atlas and axis with presence of os odontoideum and fusion of multiple vertebrae were noted in an intact skeleton, in the osteology museum of the Department of Anatomy of North Bengal Medical College, West Bengal, India. These multiple abnormalities at various levels along with increased thickness of antero-posterior arch of atlas pointed towards the congenital nature of the anomalies, possibly due to Klippel-Feil syndrome (KFS). These unusual findings denote a developmental background of the manifestations. The cervical instability, resultant neurodeficit and impairment of quality of life of the affected individuals, which are inherent in such cases, reveal their clinical importance.

**Keywords:** Congenital, Klippel-Feil Syndrome, Multiple vertebral fusion, Os odontoideum

## CASE REPORT

During a study of the intact skeletons preserved in the osteology museum of the Department of Anatomy of North Bengal Medical College, West Bengal, an interesting observation was made in an intact skeleton. A careful assessment of the different bones of the said skeleton helped us to come to the conclusion that it possibly belonged to an adult male of apparent bone age of 60 yrs. The following unusual findings were noted:

### Atlas (C 1 Vertebra)

A mass resembling a detached odontoid process was found to be fused to the anterior arch of the atlas, occupying the position of the missing body of the vertebra [Table/Fig-1a]. The antero-posterior dimension of the anterior arch of the atlas was measured to be 13.6mm [Table/Fig-1b].

### Axis (C 2 Vertebra)

The odontoid process was absent. The surface of the superior aspect of the body of C2 was flattened, relatively smooth and resembled the bodies of other vertebrae. The surface of the inferior articular facet of the left side was rough, enlarged with ragged margins [Table/Fig-2].

### C3 To C7 Vertebrae

The most distinctive feature was the complete fusion of the fifth and sixth cervical vertebrae. The bodies and zygapophyseal joints, with the exception of the laminae and the spine, were found to be fused, with features of degenerative changes [Table/Fig-3].

The cervical vertebrae thus showed multiple variations from the normal [Table/Fig-4].

### Thoracic And Lumbar Vertebrae

Another valuable finding was the complete fusion of the bodies of 12<sup>th</sup> thoracic with the 1<sup>st</sup> lumbar vertebra. The zygapophyseal joints were found to be fused leaving the spine and laminae separate [Table/Fig-5].

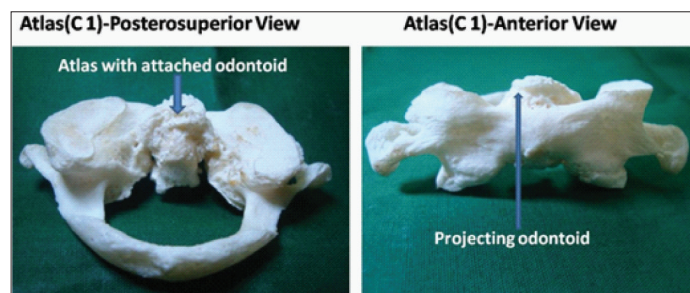
## DISCUSSION

The vertebral column develops from the para-axial mesoderm, which gets organized into block-like somites [1]. Each somite differentiates

into a myotome, a dermatome, and a bone forming sclerotome that condenses around the neural tube [Table/Fig-6] [2]. The sclerotome portion of each somite undergoes a process called resegmentation so that each vertebra is formed by the union of the caudal half of one somite and cranial half of the next somite [Table/Fig-6] [1]. During the third to eighth week of embryonic development, failure of normal segmentation of the cervical somites may result in fusion of the cervical vertebrae [3].

Among the seven cervical vertebrae, the first one known as atlas, is identified by the absence of its body and looks like a ring of bone. The second one, known as axis (as it allows the rotation of the head along a vertical axis), is identified by the presence of an elongated process called the dens (odontoid process), at the upper surface of the body of the vertebra [4]. Os odontoideum is defined as an ossicle with smooth, circumferential cortical margin representing odontoid process but without any bony continuity with body of axis vertebrae [5]. During development of foetus, in the cranio-vertebral (occipito-cervical) region, the centra formed from the sclerotomes 5(X), 6(Y) and 7(Z) fuse together to form the odontoid process which fuses with upper surface of the body of axis [4]. Muller and O'Rahilly, classified the odontoid process on the basis of failure of fusion of XYZ components amongst themselves as :-

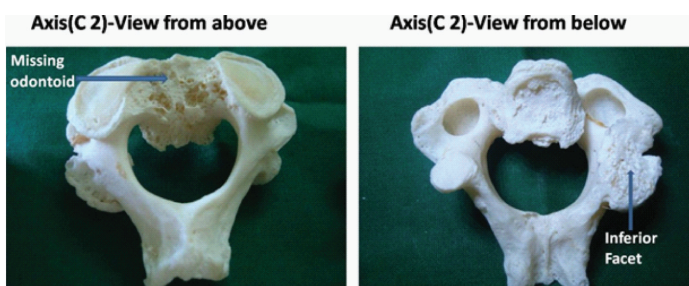
1. Ossiculum terminale, where X fails to fuse with YZ complex.
2. Os odontoideum, where XY complex fails to fuse with Z. This may occur due to presence of transitory intervertebral disc or as a result of increased movements at the time of ossification of the dens [4]. Os odontoideum is of two types- orthotopic which is



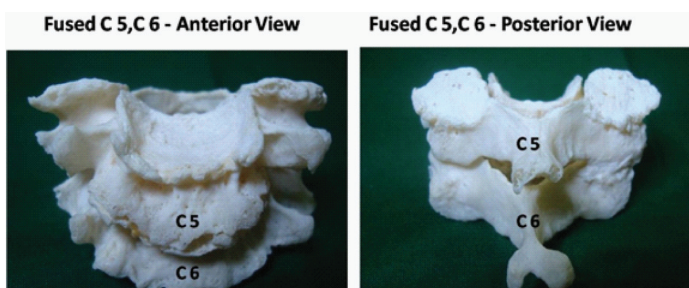
**[Table/Fig-1a]:** Photograph of abnormal atlas vertebra(C 1) with odontoid process attached to the anterior arch of atlas



**[Table/Fig-1b]:** Photograph showing the measurement of the anterior arch of the atlas



**[Table/Fig-2]:** Photograph of abnormal axis vertebra(C 2) with missing odontoid process (Left) and rough, ragged and enlarged left inferior articular facet of the axis vertebra compared to the right one(Right)

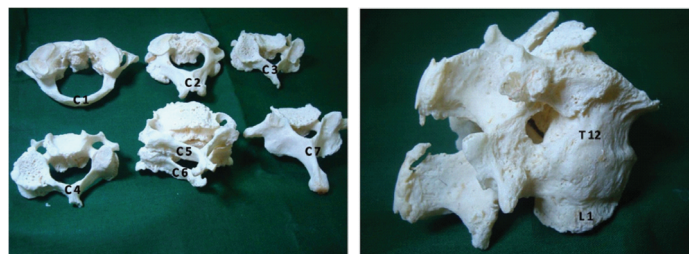


**[Table/Fig-3]:** Photograph showing complete fusion of the fifth(C 5) and the sixth(C 6) cervical vertebra distinctly visible from both anterior aspect (Left) and the posterior aspect(Right)

attached with anterior arch of atlas(as in our case thus mimicking body of atlas),and dystopic which are fused to basion [3]. This leads to either fusion of odontoid process to the anterior arch of atlas, resulting in an abnormal axis with missing dens or subluxation of os odontoideum anterior to the arch of atlas (C1) [6].

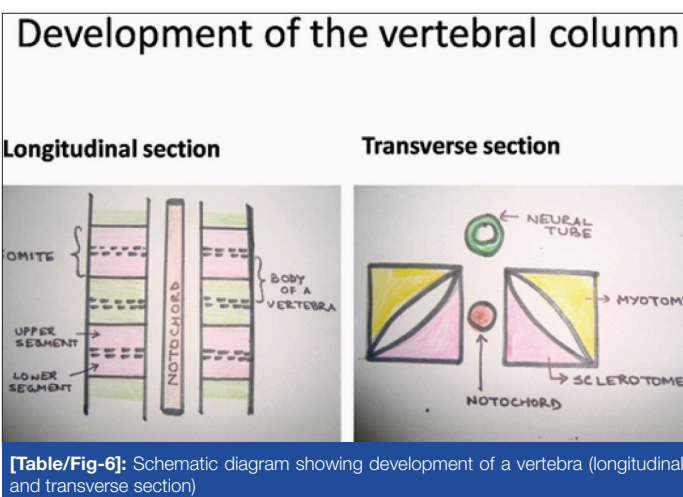
Os odontoideum has a multifactorial aetiology- post-traumatic and congenital [7,8]. Klippel-Feil syndrome (KFS), skeletal dysplasias, Morquio syndrome, Downs syndrome etc are amongst the congenital causes of os odontoideum [3,9-11]. Even if congenital in nature, the anomaly often comes to notice in adulthood after a trivial trauma, increasing the dilemma in the clinician's mind as regard to the aetiological nature of the defect [3]. Presence of multiple anomalies and antero-posterior thickness of anterior arch of atlas more than 7.9 mm have been found to be strong predictors of congenital aetiologies [12]. Presence of fusion of cervical and/or other vertebrae along with os odontoideum strongly points towards KFS [3].

KFS is a rare congenital condition where fusion of the cervical vertebrae are common. The vertebral affection may be in the form of involvement of the two segments, a congenital block vertebra or the entire cervical spine can be affected [13,14]. The classical presentation is an individual with a triad of short neck, low posterior hairline and restricted movement of the neck [3,13]. The incidence of KFS varies from 1 in 42,400 births to 3 in 700 [3]. According to Feil's classification, KFS can be of three varieties [3]. Of these, Type 3 is the most uncommon variety, being found in 13% of KFS. In Type 3 KFS, in addition to fusion of cervical vertebrae, there



**[Table/Fig-4]:** Photograph showing all the seven cervical vertebra in one frame clearly demonstrating the array of aberrations from the normal

**[Table/Fig-5]:** Photograph showing complete fusion between the twelfth thoracic(T 12) and the first lumbar(L 1) vertebra



**[Table/Fig-6]:** Schematic diagram showing development of a vertebra (longitudinal and transverse section)

may be fusion of the lower thoracic or lumbar vertebrae leading to clinical manifestations of scoliosis and kyphosis. Congenital os odontoideum is an important associated finding in Type 3 KFS [3]. Genetic abnormality in KFS is not well understood. Some studies have linked mutations in PAX-1 gene and GDF6 gene with KFS [13,15]. Presence of C5-C6 fusion has been associated with autosomal recessive transmission [15]. Radiograph of the whole spine should be done in cases of KFS for determination of the extent of vertebral anomalies. Evaluation of other systems is also essential as associated cardiac, pulmonary, renal, auditory and extraspinal skeletal anomalies like Sprengel's deformity are sometimes noted in KFS [3,13].

In our case, antero-posterior thickness of anterior arch of atlas was found to be 13.6 mm. In addition, there was presence of orthotopic os odontoideum, complete fusion of fifth and sixth cervical vertebrae along with fusion between twelfth thoracic (T12) and first lumbar (L1) vertebra. All these findings strongly favoured a congenital aetiology especially Type 3 KFS.

It can be surmised that any sort of fusion of the atlas and axis or other cervical, thoracic or lumbar vertebrae may lead to restriction of mobility, pain or serious neurodeficit resulting from nerve compression [16]. Diagnosis of Os odontoideum requires at least a routine cervical spine radiograph including open-mouth odontoid view. Computed Tomography (CT) scan with reconstruction view and Magnetic Resonance Imaging (MRI) show better anatomical details [17]. Management may be conservative, surgery in the form of posterior atlanto-axial or other fusion procedures or combination of both. Progressive symptoms, neurodeficit and/or increasing atlanto-axial instability are the main indications of surgery [17].

## CONCLUSION

Os odontoideum is a rare vertebral anomaly and KFS is an important congenital condition associated with it. Early ante-mortem diagnosis requires high index of suspicion and is crucial for an early multidisciplinary management including surgery and rehabilitation for a better outcome and quality of life.

## REFERENCES

- [1] Sadler TW, editor. Langman's Medical Embryology. 11<sup>th</sup> ed. Philadelphia : Lippincott Williams and Wilkins. 2010.
- [2] Cochard LR, editor. Netter's Atlas of Human Embryology. 1<sup>st</sup> ed. Philadelphia: ICON Learning Systems. 2002.
- [3] William C, Warner Jr. Pediatric Cervical Spine. In: Canale ST, Beaty JH, editors. Campbell's Operative Orthopaedics. Philadelphia: Elsevier Mosby. 2013. pp. 1653-91.
- [4] Collins P, Healy JC, Newell RLM. Development of the back. In: Standring S, Borley NR, Collins P, Crossman AR, Gatzoulis MA, Healy JC, et al, editors. Gray's Anatomy- The Anatomical Basis of Clinical Practice. China: Churchill Livingstone Elsevier. 2008: pp. 763-73.
- [5] Spierings EL, Braakman R. The management of os odontoideum: analysis of 37 cases. *J Bone Joint Surg Br*. 1982;64:422-28.
- [6] Feilding JW, Hensinger RN, Hawkins RJ. Os odontoideum. *J Bone Joint Surg*. 1980; 62:376-83.
- [7] Sankar WN, Wills BP, Dormans JP, Drummond DS. Os odontoideum revisited: the case for a multifactorial etiology. *Spine*. 2006;31:979-84.
- [8] Zygorakis CC, Cahill KS, Proctor MR. Delayed development of os odontoideum after traumatic cervical injury: support for a vascular etiology. *J Neurosurg Pediatr*. 2011;7:201-04.
- [9] McHugh BJ, Grant RA, Zupon AB, DiLuna ML. Congenital os odontoideum arising from the secondary ossification center without prior fracture. *J Neurosurg Spine*. 2012;17:594-7.
- [10] Morgan MK, Onofrio BM, Bender CE. Familial os odontoideum. *Case report. J Neurosurg*. 1989;70:636-69.
- [11] Al Kaissi A, Chehida FB, Ghachem MB, Grill F, Klaushofer K. Atlanto-axial segmentation defects and os odontoideum in two male siblings with opsismodysplasia. *Skeletal Radiol*. 2009;38:293-96.
- [12] Holt RG, Helms CA, Munk PL, Gillespy T. Hypertrophy of C1 anterior arch: useful sign to distinguish os odontoideum from acute dens fracture. *Radiology*. 1989;173: 207-09.
- [13] Spiegel DA, Dormans JP. The Neck. In: Kliegman LM, Stanton BF, Schor NF, St. Geme JW, Behrman RE, editors. Nelson's Textbook of Pediatrics. 19th ed. Philadelphia: Elsevier. 2011. pp. 2377-82.
- [14] Samartzis D, Kalluri P, Herman J, Lubicky JP, Shen FH. The extent of fusion within the congenital Klippel-Feil segment. *Spine*. 2008; 33: 1637-42.
- [15] Mc Gaughran JM, Oates A, Donnai D, Read AP, Tassabehji M. Mutations in PAX 1 may be associated with Klippel-Feil syndrome. *Eur J Hum Genet*. 2003;11:468-74.
- [16] Sugita S, Chikuda H, Ohya J, Taniguchi Y, Takeshita K, Haga N, et al. Cervical canal stenosis caused by progressive fusion and enlargement of cervical vertebrae with features of Proteus syndrome and Klippel-Feil syndrome. *Skeletal Radiol*. 2013;42:1743-46.
- [17] Rozzelle CJ, Aarabi B, Dhall SS, Gelb DE, Hurlbert RJ, Ryken TC, et al. Os odontoideum. In: Guidelines for the management of acute cervical spine and spinal cord injuries. *Neurosurgery* 2013;72:159-69.

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