Familial os odontoideum

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A familial asymptomatic os odontoideum with a Klippel-Feil type II fusion of C-2 and C-3 is reported. The pattern of inheritance within this family is consistent with that of autosomal dominance. The index case, a 16-year-old boy, was studied with plain cervical spine x-ray films, lateral cervical tomography in flexion and extension, fluoroscopic evaluation of the subluxation, and magnetic resonance (MR) imaging of the spine in flexion and extension. In spite of the subluxation noted on flexion and extension, there was no evidence of cord compression on MR imaging. The etiology and management of this condition are discussed.

KEY WORDS  • familial disorder  • os odontoideum  • Klippel-Feil deformity  • cervical spine

Os odontoideum was first described by Giacomini in 1886 from a postmortem examination in which a joint was found separating the odontoid process from the body of the axis. The origin of os odontoideum has been ascribed to both acquired and congenital conditions. It has been argued that the radiological features per se do not distinguish between acquired or congenital etiologies. Proponents of an acquired origin have cited in support of their thesis the location of the separation and longitudinal studies of patients who have been shown to develop the separation. The evidence in favor of a congenital etiology includes association with congenital anomalies and a pathological study. The treatment of os odontoideum has also been controversial. While it is generally accepted that symptomatic patients should undergo surgical intervention, the case for prophylactic fusion is less clear. A report of a familial os odontoideum is presented and the etiology and management of the index case is discussed.

Case Report

This 16-year-old schoolboy was involved as the driver in a motor-vehicle accident in March, 1988. He reported striking the left side of his head but sustained no loss of consciousness or neurological symptoms. On examination at a nearby hospital, it was reported that he had a small scalp hematoma in the left posterior parietal region. After the accident, he complained of mild headache and neck pain but no neurological deficit was found. Cervical spine x-ray films in the emergency room disclosed an os odontoideum and he was transferred to the Mayo Clinic for recommendations regarding treatment.

Examination. On arrival at the Mayo Clinic, it was confirmed that he had no neurological deficit or history of neurological symptoms. The patient's father and grandmother could recall no trauma that he might have suffered as a young child that could account for an ununited odontoid fracture. A review of the plain-film radiographs of the patient revealed that in addition to the os odontoideum he also had a Klippel-Feil type II fusion of C-2 and C-3 (Fig. 1). Flexion and extension tomograms and fluoroscopy demonstrated that C-1 moved relative to C-2 by 4 mm anteriorly on flexion and 6 mm posteriorly on extension. To ascertain whether there was any cord compression during head and neck movement, the patient underwent magnetic resonance (MR) imaging in flexion and extension. No compression of the upper cervical cord was demonstrated (Fig. 2). An examination of skull x-ray films taken of the patient at the age of 7 and 11 years after minor trauma revealed that the os odontoideum...
Familial os odontoideum and Klippel-Feil fusion had been present at least by 7 years of age.

Familial Associated Disorders. The patient's paternal grandmother (64 years) had previously had cervical x-ray films taken at the Mayo Clinic which demonstrated an os odontoideum and Klippel-Feil fusion of C-2 and C-3. The x-ray films were obtained after minor trauma and there was no history of neurological abnormality. Both the patient's father (39 years) and his maternal grandmother denied having symptoms suggestive of spinal cord compression at any time in the past. However, in view of the radiological abnormalities of the patient and his grandmother, it was decided to request the entire family to undergo cervical x-ray films and flexion and extension tomography if abnormalities were detected.

Investigation of the family demonstrated that the father and the paternal grandmother had both an os odontoideum and a Klippel-Feil type II fusion of C-2 and C-3. In flexion and extension, the relative movement of C-1 on C-2 of the father was 50% less than that of his son and in the maternal grandmother was almost negligible. All other descendants of the maternal grandmother and her sibling were evaluated by plain spine radiography and reviewed at the Mayo Clinic (Fig. 3). No other family member was found to have this abnormality.

Course. In view of the family history of asymptomatic os odontoideum, the lack of neurological symptoms in the patient, and the absence of MR imaging evidence of compression of neural structures, it was decided that prophylactic surgical fusion at this time was not indicated. Within 1 week of discharge, the patient's neck pain resolved.

Discussion

This report is of a familial os odontoideum and Klippel-Feil type II fusion of C-2 and C-3. The mode of inheritance of this condition in this family fits well with the autosomal dominant pattern reported to occur in the minority of cases of Klippel-Feil syndrome, but which has not been previously described for os odontoideum. A congenital origin for os odontoideum in this case would seem reasonable in view of the associated Klippel-Feil anomaly and the apparent Mendelian inheritance of the condition. This is supported by a pathological study by Sherk and Dawoud of a patient with both conditions in whom ligamentous attachment to the os odontoideum suggested its congenital origin.

Fielding, et al., attest that, because the epiphyseal plate of the odontoid process lies below the anatomical base of the dens (which appears to be the site of separation in os odontoideum), the abnormality represents an acquired lesion such as an ununited fracture. The predilection for loss of bone tissue at this location is related to the blood supply to the proximal odontoid process, which is vulnerable to trauma. Fielding, et al., supported their thesis with nine cases in which it was alleged that cervical x-ray films taken at an earlier age demonstrated a normal odontoid process. This has also been observed by others. However, nine of 35 patients with os odontoideum reported by Fielding, et al., had no history of previous trauma and five of these nine patients had associated congenital abnormalities, including one with a Klippel-Feil syndrome.
One might argue that the congenital conditions that have been associated with os odontoideum (Morqio’s disease, epiphyseal dysplasias, Down’s syndrome, and Klippel-Feil syndrome\(^5,8,25,27\)) may in some way predispose the patient to developing an acquired os odontoideum.\(^8\) However, the pathological description of os odontoideum previously reported by Sherk and Da-woud\(^29\) and the association of a Klippel-Feil type II fusion with os odontoideum in three cases in the same family (as reported here) in an otherwise uncommon association support evidence for the existence of a congenital etiology for the os odontoideum in this case. Therefore, it is suggested that both congenital and acquired etiologies for os odontoideum are possible.

The management of asymptomatic os odontoideum is controversial. While anecdotal reports of death and irreversible cord injury from trivial insults have been documented for many years,\(^21\) the true natural history is unknown. Spiers and Braakman\(^30\) failed to demonstrate a difference in outcome in asymptomatic patients when comparing those undergoing conservative treatment with those undergoing posterior cervical fusion. They found that none of 16 patients who presented without “cord signs” and who were treated conservatively developed neurological deficits in 98 patient years of follow-up evaluation. They also presented evidence that the critical factor in the genesis of “cord signs” in this condition is not the amount of subluxation of the atlantoaxial joint \(\text{per se}\) but rather the sagittal diameter of the canal. They found that, in 11 patients without evidence of myelopathy, the minimum distance between the posterior border of the body of C-2 and the posterior atlantal arch on flexion was 15.2 ± 3 mm compared with 11.9 ± 3.2 mm in those with transient or permanent “cord signs.” In the patient described here, the MR image demonstrated no cord compression by either bone or ligament in flexion or in extension, despite the significant excursion of C-1 on C-2.

While the natural history for this condition is unknown in general and for the index case presented here, it would appear that within this family it has been benign. If it is assumed that the os odontoideum was congenital in all three family members, then the family has experienced 119 patient years without neurological sequelae from this condition. Although the subluxation of C-1 on C-2 is greater for the index case than for either of his affected relatives at the present time, this may be due to the reduction in ligamentous laxity with age.\(^3,15,14\) Therefore, the other two family members could conceivably have had a similar degree of movement as the index case at a comparable age.

The risks of surgical management are short- and long-term. Although both posterior and anterior decompression may be suggested in certain situations,\(^13,18-20,26,27\) the most widely accepted therapeutic intervention is stabilization by posterior wiring and concomitant bone graft fusion.\(^4,5,8,15,22,24,33,35\) This approach has proved to be a procedure associated with low morbidity and mortality rates; however, unexpected death has been reported\(^2,4,33\) and persistent postoperative myelopathic deficits have been noted in patients who were previously normal.\(^20\) Therefore, it is unlikely that the procedure can be carried out with no risk of serious complications.

Of perhaps even greater significance for morbidity are the long-term consequences of fusing such an important motion segment at such a young age. There is good evidence that fusion at one interspace accelerates the degenerative changes in adjacent motion segments.\(^5,17\) Based on the asymptomatic nature of the familial os odontoideum in the present case, a conservative approach will be used. Periodic neurological reevaluation and repeat cervical spine x-ray studies will be performed. In view of the morbidity associated with surgery, an attempt to prevent the theoretically increased risk of cervical cord injury from a possible future flexion/extension injury seems injudicious in this patient unless there is an increase in his C1–2 excursion or symptoms of cord compression are documented.

### References

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