Occipitoatlantal Anteroposterior Subluxation Associated with Condylar Hypoplasia and Congenital Atlantoaxial Fusion: Clinical Correspondence

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Nontraumatic occipitoatlantal (OC1) anteroposterior subluxation is a very rare type of instability. The condition has been caused by occipital condylar hypoplasia in one case⁵, occipital condylar enlargement in one case⁶, upper cervical spine congenital fusion in two cases⁷, and unknown reasons in one case⁸. Myelopathy caused by nontraumatic OC1 anteroposterior subluxation is an extremely rare pathology. The coexistence of occipital condylar hypoplasia and congenital atlantoaxial fusion is one of the most uncommon malformations among occipitocervical anomalies⁹⁻¹⁰. This report describes a very rare case of nontraumatic OC1 anteroposterior subluxation presenting as myelopathy that was caused by coexisting condylar hypoplasia and congenital atlantoaxial fusion.

A 70-year-old male patient complained of clumsiness in the bilateral hands, severe gait imbalance, dullness of the left upper limb, and numbness of the right lower limb. The symptoms were progressive, and the patient became unable to walk or use chopsticks. He had no history of comorbidity or trauma attributable to neurological disorders. A short neck, low hairline, and limited range of motion in the neck were noted. According to these findings and related vertebral anomalies, a diagnosis of Klippel-Feil syndrome was made. Neurological examination revealed hyperreflexia for the BTR, BRTR, and TTR, with bilateral positive Babinski reflexes.

Cervical radiographs showed OC1 translation and canal stenosis at C3-C7. OC1 translation was evaluated based on a report by Tredwell et al.⁶ (Fig. 1). Computed tomography (CT) indicated congenital fusion of the C1-C2 (Cave’s Group 2)⁹ (Fig. 2A-2E) and flat surfaces of the bilateral OC1 joints in both sagittal and coronal planes⁷ (Fig. 2D). Magnetic resonance imaging (MRI) of a neutral position showed myelomalacia of the cervicomedullary junction (Fig. 2F, 2G). C3-C6 spinal cord compression was also observed (Fig. 2F). MRI during flexion revealed a bowstring deformity of the cervicomedullary junction (Fig. 2H, 2I).

The patient underwent O-C3 posterior fusion and C3-C7 laminoplasty (Fig. 3). As the O/C1 joint had subluxation both anteriorly and posteriorly, we realized that it was necessary to fix it in the middle position, which was selected such that the foramen magnum and the cervical spinal canal would be aligned normally under lateral fluoroscopy. In addition, rod bending was carefully performed for in-situ fixation. The patient’s myelopathy was rapidly alleviated after surgery, and he became ambulatory and able to use chopsticks at postoperative 4 weeks. The dullness of the left upper limb and numbness of the right lower limb also improved.

Congenital atlantoaxial fusion is one of the rarest forms of cervical anomaly⁵⁻⁸. Occipital condylar hypoplasia is a remnant of proatlas malformation and likely to be caused by Pax-1 gene aberrations⁶. The occipital condyles are biomechanically important for OC1 stability; the articular surface...
Figure 1.  Lateral radiographs during flexion (a, A), in a neutral position (b, B), and during extension (c, C). Six millimeters of anterior translation in flexion (a) and 5 mm of posterior translation in extension (c) at the OC1 level were observed.

Figure 2.  Sagittal CT demonstrated a flat surface of the bilateral OC1 joints and congenital atlantoaxial fusion (A: right, B: left). Axial-view CT at the C2 pedicle level revealed a high-riding vertebral artery (VA) on the left side (C). Coronal-view CT of the craniovertebral junction disclosed an abnormal OC1 joint axis angle (D). 3D CTA revealed no vascular anomalies apart from the high-riding VA on the left side (E). MRI of a neutral position demonstrated myelomalacia without OC1 compression and spinal cord compression at C3–C7, with a high intensity change at C5–C6 (F). Asymmetric deformity of the spinal cord was recognized at the OC1 level (G). By contrast, a bowstring deformity of the cervico-medullary junction was detected during flexion (H and I).

of the atlas is cup-shaped to accommodate the arcuate occipital condyle. This characteristic structure of the OC1 joints limits translation and axial rotation\(^1\). Previous in vivo and in vitro biomechanical studies have described almost no forward-backward translation\(^9\). A cadaveric biomechanical investigation by Vishteh et al. demonstrated that if 50% of the unilateral occipital condyles were resected, the ranges of motion in flexion extension, side bending, and axial rotation would increase by 15.3%, 40.8%, and 28.1%, respectively\(^10\). OC1 instability in the absence of these diseases or high-energy trauma is very rare (Table 1). Wiese et al.\(^3\) encountered two cases of OC1 instability resulting from congenital
Figure 3. Posterior O-C3 instrumentation using occipital screws (A), a unilateral C2 pedicle screw (B), and bilateral C3 pedicle screws (C) followed by local bone grafting. Additional C3–C7 laminoplasty was performed. Postoperative MRI demonstrated sufficient spinal cord decompression (D). Functional radiographs at postoperative 2 years disclosed no screw loosening or motion at the O-C3 level, indicating successful bony fusion (E and F).

Table 1. Literature Review of Atraumatic Occipitocervical Horizontal Translation.

| Author                  | Year | Age (years) | Sex | Cause                                      | Symptom(s)                                        | Treatment                                |
|-------------------------|------|-------------|-----|--------------------------------------------|---------------------------------------------------|------------------------------------------|
| Wiesel SW et al.3)      | 1979 | 23          | F   | Congenital fusion of C1-2                  | Neck pain, intermittent weakness in arms and legs | O-C2 posterior arthrodesis with wiring   |
| Wiesel SW et al.3)      | 1979 | 17          | F   | Congenital fusion of C2-3                  | Neck pain, dizziness                              | O-C2 posterior arthrodesis               |
| Georgopoulos G et al.2) | 1987 | 14          | M   | Congenital enlargement of the occipital condyles | Nausea, projectile vomiting                      | O-C1 posterior arthrodesis               |
| Georgopoulos G et al.2) | 1987 | 6           | F   | ND                                         | Neck pain, vertigo                                | O-C1 posterior arthrodesis               |
| Abumi K et al.1)        | 1998 | 28          | M   | Tropism of the occipitoatlantal articulations | Neck pain, vertigo, nausea, tightness in the chest | O-C1 posterior arthrodesis with wiring   |
| Present report          | 2018 | 70          | M   | Occipitocondylar hypoplasia and congenital fusion of C1-2 | Myelopathy                                       | O-C3 posterior arthrodesis with a rod and screws |

F: female, M: male, ND: not determined

The patient had two possible risks for C1-2 fusion: O-C1 joint flattening and O-C1 joint stability. Nonetheless, the symptoms did not appear until the age of 70 years, likely since the cervical spine below C2 could distribute the load. The O-C1 level is physiologically wide in the spinal canal; thus, only mild instability may not cause clinical myelopathy. With age, there may have been a decrease in range of motion of the middle and lower cervical spine due to spondylotic changes. The burden on O-C1 increased, which could have led to the appearance of spinal cord symptoms. In future cases, testing the scapulohumeral reflex is advisable to more clearly identify the responsible lesion.

The current case revealed several important findings. First, congenital atlantoaxial fusion and occipital condylar upper cervical fusion. Georgopoulos et al. reported on a patient with congenital hypertrophy of the occipital condyles that indicated excessive OC1 movement2. Abumi et al. described a case of OC1 instability with tropism of the OC1 articulations9. Importantly, these cases did not exhibit spinal cord impairment, indicating that nontraumatic anteroposterior subluxation alone at the OC1 level rarely caused myelopathy.
Hypoplasia can coexist. Second, such a combination results in myelopathy. Both congenital and surgical atlantoaxial fusion biomechanically increases stress on the OC1 joints. The configuration of these joints should accordingly be assessed prior to atlantoaxial fixation to avoid postoperative OC1 instability.

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