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Congenital Atlanto-Occipital Fusion and its Effect on the Myodural Bridge: A Case Report Utilizing the P45 Plastination Technique

Fusión Atlanto-Occipital Congénita y su Efecto sobre el Puente Miodural: Reporte de un Caso Utilizando la Técnica de Plastinación P45

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SUMMARY: The atlanto-occipital joint is composed of the superior fossa of the lateral masses of the atlas (C1) and the occipital condyles. Congenital Atlanto-occipital fusion (AOF) involves the osseous union of the base of the occiput (C0) and the atlas (C1). AOF or atlas occipitalization/assimilation represents a craniovertebral junction malformation (CVJM) which can be accompanied by other cranial or spinal malformations. AOF may be asymptomatic or patients may experience symptoms from neural compression as well as limited neck movement. The myodural bridge (MDB) complex is a dense fibrous structure that connects the suboccipital muscular and its related facia to the cervical spinal dura mater, passing through both the posterior atlanto-occipital and atlanto-axial interspaces. It is not known if atlas occipitilization can induce structural changes in the MDB complex and its associated suboccipital musculature. The suboccipital region of a cadaveric head and neck specimen from an 87-year-old Chinese male having a congenital AOF malformation with resultant changes to the MDB complex was observed. After being treated with the P45 plastination method, multiple slices obtained from the cadaveric head and neck specimen were examined with special attention paid to the suboccipital region and the CVJM. Congenital atlanto-occipital fusion malformations are defined as partial or complete fusion of the base of the occiput (C0) with the atlas (C1). In the present case of CVJM, unilateral fusion of the left occipital condyle with the left lateral mass of C1 was observed, as well as posterior central fusion of the posterior margin of the foramen magnum with the posterior arch of C1. Also noted was a unilateral variation of the course of the vertebral artery due to the narrowed posterior atlanto-occipital interspace. Surprisingly, complete agenesis of the rectus capitis posterior minor (RCPmi) and the obliques capitis superior (OCS) muscles was also observed in the plastinated slices. Interestingly, the MDB, which normally originates in part from the RCPmi muscle, was observed to originate from a superior bifurcation within an aspect of the nuchal ligament. Therefore, the observed changes involving the MDB complex appear to be an effective compensation to the suboccipital malformations.

KEY WORDS: Craniovertebral junction malformations; Congenital atlanto-occipital fusion; Myodural bridge; P45 plastination.

INTRODUCTION

Congenital atlanto-occipital fusion is a form of craniovertebral junction malformation (CVJM). Previous studies have described four major types of anomalies of the occipito-cervical junction: of the occipital bone; III, atlanto-axial dislocation or atlanto-occipital fusion; IV, failure of fusion of ossification Patients with congenital atlanto-occipital fusion may present with compensatory hyperactivity of the atlanto-axial joint, tonsillar herniation, syringomyelia (Hirose *et al*., 1998; Iwata *et al*., 1998), and limited neck movement. According to the specific location of the fused areas, the morphological classification of AOF is divided into three categories: a fused anterior arch of C1 to the occiput (C0); a fused lateral mass of C1 to the occiput (C0); and a fused posterior arch of C1 to the occiput (C0)

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(Goel & Shah, 2010; Tubbs *et al*., 2011). The present paper reports a case of fusion of both lateral masses of the atlas (C1) to the occiput (C0) as well as fusion of the posterior arch of atlas C1 with the occiput (C0). To our knowledge, this unusual type of atlanto-occipital fusion has not been described previously in the literature. Based on our observations, a narrowing was noted within the posterior atlanto-occipital interspace (PAOI). Moreover, a dense fibrous band of tissue, presumably the MDB, was identified passing through the narrowed PAOI. Observations from the present case report offers new insight into the potential association between AO bone fusion and suboccipital muscle agenesis.

MATERIAL AND METHOD

The formalin fixed adult head and neck specimens from an 87-year-old Chinese male were obtained from the Body and Organs Donation Center of Dalian Medical University for the present study. The P45 plastinated slices were prepared at the Dalian Hoffen Preservation Technique Institution. Median and paramedian sagittal slices of the P45 plastinated specimens were examined with special attention paid to the suboccipital region and the CVJM and recorded by photography. The slice thickness was 3 mm. The procedures involved in the P45 sheet plastination process, included slicing, bleaching, dehydration, forced impregnation, and curing.

Slicing. Embalmed head and neck specimen was frozen at -70 ºC for 2 weeks. Then, a set of 3-mm-thick sagittal slices was prepared with a high-speed band saw.

Bleaching. All slices were rinsed with running water for 6-8 h and immersed in 5 % hydrogen peroxide overnight for bleaching.

Dehydration. After bleaching, the slices were dehydrated with 100% acetone by the freeze substitution method.

Casting and forced impregnation. After dehydration, the casting mold was prepared. The slices were lifted from the acetone bath and placed between two glass plates. The molds were then filled with polyester (Hoffen polyester P45, Dalian Hoffen Bio-Technique Co. Ltd., Dalian, P. R. China). The filled mold was then placed upright into a vacuum chamber at room temperature for impregnation. The large bubbles on the surface of the slices were removed manually with a 1-mm stainless steel wire. The absolute pressure was slowly decreased to 20, 10, 5, and 0 mm Hg, according to the bubble releasing. The pressure was maintained at 0 mm Hg until bubbling ceased. The impregnation step lasted for more than eight hours.

Curing. After vacuum release, the top of the mold was clamped with large fold back clamps, and the sheet was ready for curing. The sheets were placed in an upright position and cured in a heated water bath at 40? for 3 days. After curing, the slices were taken out from flat chamber and were covered with adhesive plastic wrap for protection. A small band saw wasused to cut and trim the plastic along the outside edges of the slices at a distance of approximately 1 mm from the tissue. To cut out the sharp edges of the slices, a wool sander was used. The sheets were then ready for use.

CASE REPORT

The present authors report the case of an 87-year-old Chinese man with congenital atlanto-occipital fusion. The human head and neck specimen, exhibiting a bony union between the occiput and atlas (C1), was observed via a series of slices using the P45 plastination technique.

It was observed that the posterior arch of atlas (C1) was fused to the base of the occiput with the resultant narrowing of the normally present posterior atlanto-occipital interspace. Additionally, the posterior margin of foramen magnum and the posterior arch of C1 were also fused together via a patch of compact bone at the posterior center of the atlanto-occipital interspace (Fig. 1A,B,H).

The lateral aspects of the atlanto-occipital joints were also observed. The left lateral mass of C1 was observed to be fused to the left occipital condyle via not only the compact bone superficially, but also the deep spongy bone (Fig. 1E,H). Unlike the left AOJ, the right atlanto-occipital joint remained separate as is normally the case (Fig. 1F,G,H). However, the left lateral part of the posterior atlanto-occipital interspace was observed to be much narrower than the right lateral part, and not large enough to allow the normal passage of the left vertebral artery. As a result, the left vertebral artery passed under the posterior arch of C1 (Fig. 1F) and then entered the vertebral canal along the lateral margin of the vertebral foramen of C1 (Fig. 1G).

In the sagittal slices, the rectus capitis posterior major (RCPma) and the obliquus capitis inferior (OCI) muscles were present and distinct (Fig. 1E,F,G). However, there was no evidence of either the rectus capitis posterior minor (RCPmi) or the obliquus capitis superior (OCS) muscles in the suboccipital region. Interestingly, fibrous bundles (Fig. 1C,D,F,G), of the MDB complex entered the narrowed posterior atlanto-occipital interspace and then inserted onto the cervical spinal dura mater (Fig. 1C,D).

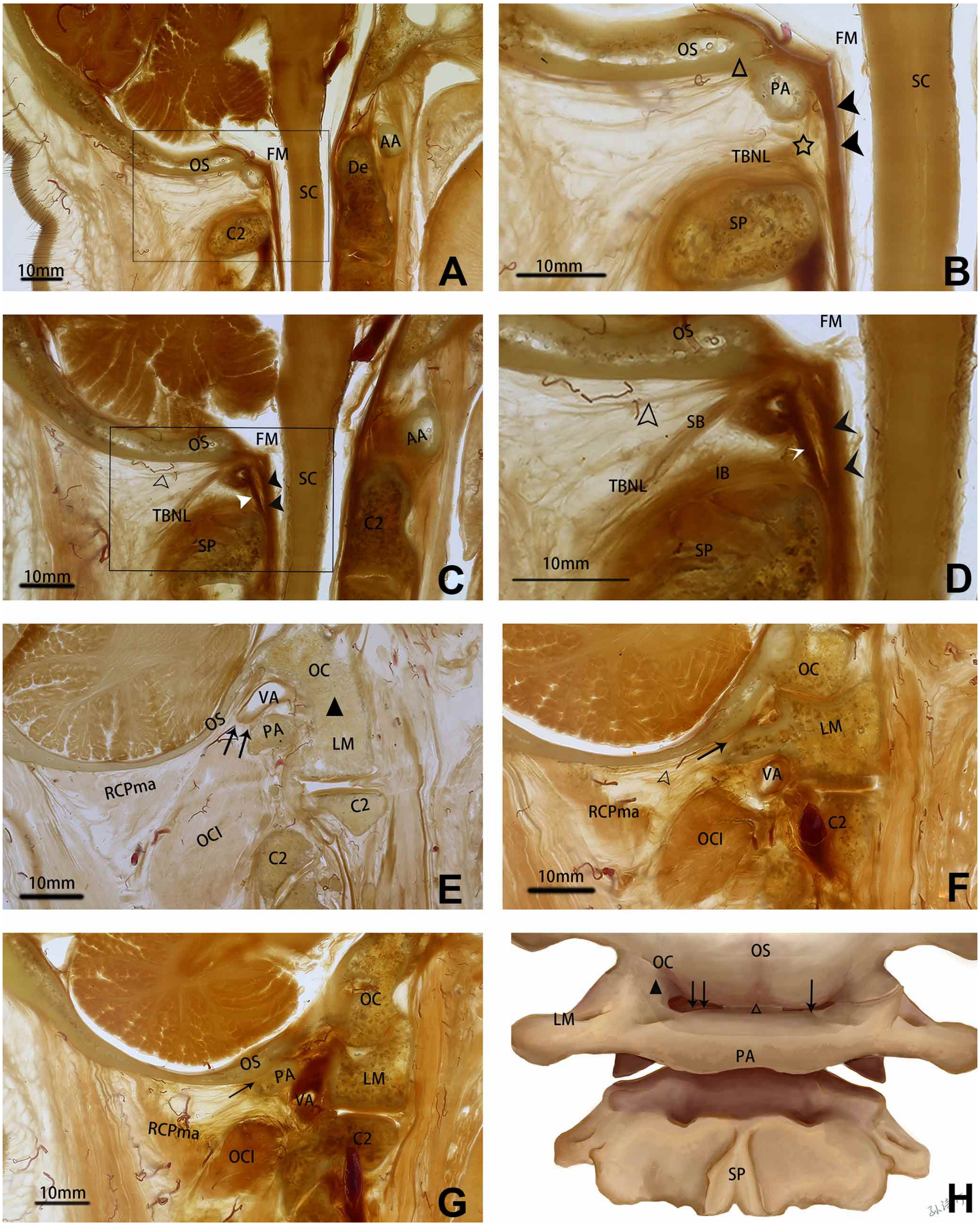

Fig. 1. (A-G) Sagittal P45 plastinated sections of the craniovertebral junction. (h) Image of C0, C1, and C2. Figure A, Median sagittal section; Figure B, Magnification of the box in Figure A; Figure C, Parasagittal section on the left; Figure D, Magnification of the box in Figure C; Figure E, Sagittal section through the left lateral mass of C1; Figure F, Sagittal section through the right Atlanto-occipital joint; Figure G, Parasagittal section on the right; Figure H, Diagram of combined fusions of the occiput and the atlas. OS, occipital squamous; FM, foramen magnum; SC, spinal cord; AA, the anterior arch of atlas; De, dens of axis; PA, the posterior arch of atlas; TBNL, To Be Named Ligament (an aspect of the nuchal ligament); SP, spinous process of axis; SB the superior bifurcation of TBNL; IB, the inferior bifurcation of TBNL; OC, occipital condyle; LM, the lateral mass of atlas; VA, vertebral artery; OCI, obliquus capitis inferior muscle; RCPma, rectus capitis posterior major muscle; Black arrowheads, spinal dura mater; Hollow star: the posterior atlanto-axial interspace; Hollow triangle, posterior center fusion of the posterior margin of foramen magnum and the posterior arch of C1; White arrowhead, the vertebradural ligament ; Solid triangle, fusion of the lateral mass of C1 and the occipital condyle on the left; Double black arrows: the narrowed posterior atlanto-occipital interspace on the right; Black arrow: the slit (narrowed space) of the posterior atlanto-occipital interspace on the left; Hollow arrowheads, fibrous bundles of the MDB complex entering the narrowed posterior atlanto-occipital interspace.

The "To Be Named Ligament (TBNL)" was originally described by anatomists Sui and Yu in 2014, and represents a dense fibrous structure within the nuchal ligament that arises from the posterior margin of the nuchal ligament and extends forward and upward passing through the atlanto-axial interspace (Zheng *et al*., 2014). The dense TBNL, observed in the present specimen, bifurcated and extended into the posterior atlanto-occipital and the atlanto-axial interspaces, respectively (Fig. 1c,d). The superior bifurcation of the TBNL ran superiorly and anteriorly, and then entered the posterior atlanto-occipital interspace to be inserted onto the spinal dura mater. Furthermore, within the posterior atlanto-axial interspace, the vertebrodural ligament (VDL) was observed to also consist of denser fibers. The VDL originated from the posterior arch of C1 as well as the inferior bifurcation of the TBNL, and then the VDL ran downwards to eventually fuse with the posterior wall of the spinal dura mater (Fig. 1C,D). Consequently, the posterior wall of the upper cervical spinal dura was observed to be unusually thick (Fig. 1C,D).

DISCUSSION AND CONCLUSIONS

The present authors report on a case of congenital fusion of the atlas with the occipital bone, which also presents with the unilateral course variation of the vertebral artery, as a consequence of the narrowing of the posterior atlanto-occipital interspace. Agenesis of both the RCPmi and the OCS muscles was also noted. Gholve *et al*. (2007) previously suggested that in the morphological classification of the fusion of the atlas and occiput, the prevalence of spinal canal invasion is highest in when the lateral masses of the atlas fuse with the occipital condyles. Other studies have reported that symptoms of tonsillar herniation and spinal cord cavitation were observed in patients with the fusion of the atlas to the occiput (Li *et al*., 2012). However, in the present case, spinal canal invasion, vertebral artery compression, posterior fossa stenosis, and other abnormalities were not detected. In fact, atlanto-occipital joint fusion is many times detected during physical examination without clinical symptoms however with movement of the neck being slightly limited during hyperflexion or hyperextension (Hall *et al*., 2015).

Recently, there has been numerous reports suggesting that the suboccipital muscles as well as the nuchal ligament serve as an origination of the myodural bridge (MDB) complex, which inserts onto the upper cervical spinal dura mater, passing through both the posterior atlanto-occipital and the atlanto-axial interspaces (Kahkeshani & Ward, 2012; Yuan *et al*., 2016; McElroy *et al*., 2019; Zheng *et al*., 2020). In the present case, fibrous bundles of the MDB complex were still observed, despite the aplasia of the RCPmi and OCS muscles that normally serve as an origin of the MDB complex. In spite of the agenesis of the RCPmi and OCS muscles, the MDB was still present, and was observed passing through the narrowed posterior atlanto-occipital interspace. In the present case, the MDB complex was supported by the superior bifurcation of the TBNL as well the VDL. Furthermore, the MDB complex still passed through the posterior atlanto-axial interspace. Even though the atlas and occiput were fused and the suboccipital muscles that usually subserve the myodural bridge complex (MDBC) were absent, the body apparently compensated for this unique congenital anomaly and the MDB complex survived. Therefore, this suggests that the survival of the MDB complex indicates that it plays a significant physiological role in the human body.

In this case report, the observed atlanto-occipital fusion malformation which is reminisced of Klippel-Feil syndrome (KFS), in which there are two or more cervical vertebrae fusion deformities. KFS also presents with a reduction in the number of cervical vertebrae, a shortened neck, limited neck movement, and sometimes abnormal occipital development. The Pax1 gene, alone or in conjunction with other genetic or environmental factors, may play a role in the pathogenesis of KFS (McGaughran *et al*., 2003). Pax genes are a highly conserved family of developmental control genes that encode transcription factors (Chi & Epstein, 2002). Pax1 gene may provide vital clues to the pathogenesis of congenital atlanto-occipital fusion as described in the present report.

In summary, this case report adds to the current body of knowledge concerning craniovertebral junction malformations, our present findings contribute new insights into the important physiological role of the myodural bridge complex.

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RESUMEN: La articulación atlanto-occipital está compuesta por las caras articulares superiores de las masas laterales del atlas (C1) y los cóndilos occipitales. La fusión atlanto-occipital congénita (FAO) implica la unión ósea de la base del occipucio (C0) y el atlas (C1). La FAO u occipitalización/asimilación del atlas representa una malformación de la unión craneovertebral (MUCV) que puede presentar otras malformaciones craneales o espinales. La FAO puede ser asintomática o los pacientes pueden experimentar síntomas de compresión neural así como movimiento limitado del cuello. El complejo del puente miodural (PMD) es una estructura fibrosa densa que conecta el músculo suboccipital y su fascia relacionada con la duramadre espinal cervical, pasando a través de los espacios intermedios atlanto-occipital posterior y atlanto-axial. No se sabe si la occipitilización del atlas puede inducir cambios estructurales en el complejo PMD y en la musculatura suboccipital. Se observó en la región suboccipital de un espécimen cadavérico, cabeza y cuello de un varón chino de 87 años con una malformación congénita de FAO con los cambios resultantes en el complejo PMD. Se examinaron múltiples cortes obtenidos de la muestra de cabeza y cuello después de ser tratados con el método de plastinación P45, con especial atención a la región suboccipital y la MUCV. Las malformaciones congénitas por fusión atlanto-occipital se definen como la fusión parcial o completa de la base del occipucio (C0) con el atlas (C1). En el presente caso de MUCV se observó la fusión unilateral del cóndilo occipital izquierdo con la masa lateral izquierda de C1, así como fusión posterior central del margen posterior del foramen magnum con el arco posterior de C1. También se observó una variación unilateral del curso de la arteria vertebral por el estrechamiento del espacio interatlanto-occipital posterior. Se observó además agenesia completa de los músculos *Rectus capitis posterior minor* (RCPmi) y *oblicuos capitis superior* (OCS) en los cortes plastinados. Curiosamente, se observó que el MDB, que normalmente se origina en parte del músculo RCPmi, se origina en una bifurcación superior dentro de un aspecto del ligamento nucal. Por lo tanto, los cambios observados en el complejo PMD parecen ser una compensación de las malformaciones suboccipitales.

PALABRAS CLAVE: Malformaciones de la unión craneovertebral; Fusión atlanto-occipital congénita; Puente miodural; Plastinación P45.

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