Bull. Natl. Mus. Nat. Sci., Ser. A, 45(1), pp. 31–38, February 22, 2019

A Case of Congenital Occipitoatlantoaxial Malformation in a Wild Japanese Serow (*Capricornis crispus*)

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(Received 14 November 2018; accepted 26 December 2018)

Abstract The occipitoatlantoaxial malformations (OAAMs) are a rare skeletal defect with a neurological disorder caused by a malformation of the occipital bone of the skull and the first two cervical vertebrae. This malformation has been reported in various domestic mammals, while few reports in wild species. Here, we reported the OAAM in a wild adult Japanese serow (*Capricornis crispus*, NSMT-M 47824). In this specimen, the occipital part of the skull displayed an asymmetrical morphological deformation: a hemivertebra was fused to the right side of the occipital condyle. The dorsal and ventral arches of the atlas were unfused, resulting in the division to two hemivertebrae. The right hemivertebra of the atlas articulated with the C2 via the articular process like C2–C3 junction. Despite the atlanto-occipital and atlanto-axial joints showed a serious deformation, our skeletal observation indicated that dorsoventral and rotational movements of the head were allowed at each joint. The OAAM is known as a malformation involving serious neurological diseases like a tetraparesis, while it is inferred that the specimen had not troubled a severe functional problem at the head-neck joint.

Key words: hemivertebra, Japanese serow, Occipitoatlantoaxial malformation.

Introduction

The atlanto-occipital and atlanto-axial joints have become highly specialized through the evolution of mammals (Liem *et al.*, 2001). The atlanto-occipital joint allows the dorsoventral movement of the head, and the atlanto-axial joint allows the lateral rotation. These joints play an important role in moving the head freely independently of the trunk, and also provide the areas for the origin and insertion of the axial musculature and tendons that stabilize the cranial region (Campione and Reisz, 2011).

Congenital anomalies of the atlanto-occipital and atlanto-axial region may disturb the mobility of these joints and provide neurological disorders (McRae and Barnum, 1953; Al-Motabagani and Surendra, 2006). Due to its clinical significance, previous studies have described these anomalies of the craniovertebral region in humans (Jayanthi *et al.*, 2003; Fernandes and Costa, 2007; Saini *et al.*, 2009; Natsis *et al.*, 2016; Electricwala *et al.*, 2017) as well as various domestic mammals (Leipold *et al.*, 1972; Mayhew *et al.*, 1978; Watson *et al.*, 1985; Schmidt *et al.*, 1993).

The atlanto-occipital assimilation is known as one of common skeletal abnormalities at craniovertebral region caused by the partial or complete fusion of the atlas to the occipital bone (Ranade *et al.*, 2007; Saheb *et al.*, 2010). In domestic mammals, it is regarded as a rare anomaly termed the occipitoatlantoaxial malformations (OAAMs) (Leipold *et al.*, 1972; Mayhew *et al.*, 1978; Watson *et al.*, 1985; Schmidt *et al.*, 1993). The OAAMs are characterized by a malformation of the occipital bone of the skull and the first two cervical vertebrae.

Previous studies reported that the OAAM sometimes caused the paraparesis and tetraparesis by a compression of the upper portion of the

cervical spinal cord as well as a decline in the mobility of the head and neck joint (Watson and Mayhew, 1986). There have been many descriptions about the OAAM in the domestic mammals; however, few descriptions in a wild species.

Here, we report a case of the congenital craniovertebral malformation resembling the OAAM, in a wild adult Japanese serow (*Capricornis crispus*). The Japanese serow is a wild goat-like bovid that is endemic to Japan and distributes primarily in northern and central Honshu. We described the morphological characteristics of the malformation in the specimen, and also discussed the clinical aspect and the developmental background of it.

Materials and Methods

We used the skeletal specimen of a wild Japanese serow (collection number, NSMT-M 47824; field number, N94015) stored in the National Museum of Nature and Science (Tokyo, Japan). The carcass of this individual was retrieved at Sanada-machi (Nagano prefecture) on 11 December 1994 by Japan Wildlife Research Center as a part of the Specified Wildlife Conservation and Management Plan. This individual was a mature female during pregnancy. The age was estimated at 5.5 years old based on its horn.

The specimen was a partial skeleton: the skull and three cervical vertebral elements. The two cervical elements were hemivertebrae which comprised the atlas. The rest one was second cervical vertebra (C2) and was cut off at the posterior part of the vertebra. The skeleton more caudal than C2 was not stored.

We conducted exhaustive macroscopic observation in order to describe the morphological anomalies of the skull and vertebral elements in the specimen.

Results

In this specimen, there were several abnormalities in the segmentation and morphology around the atlanto-occipital and atlanto-axial joints. The skull, with exception of the occipital region, exhibited no morphological anomalies.

Three hemivertebrae were observed at the atlanto-occipital part. The first hemivertebra (C0) was fused to the right side of the occipital condyle and squamous part of the occipital bone. Apart from a small interstice at the ventral side, this element was completely fused to the skull (Fig. 1). This union exhibited five articular surfaces: three posterior articular surfaces for C1R, and medial and posterior articular surfaces for C1 L.

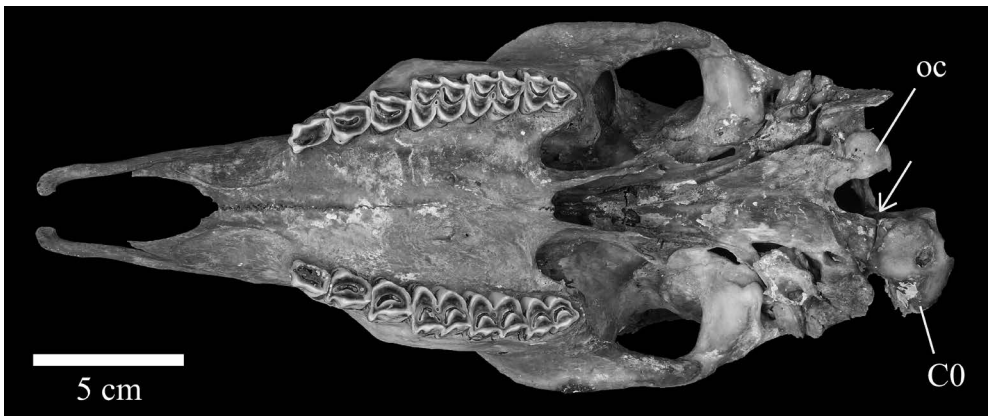

Fig. 1. Ventral view of the skull of this specimen (NSMT-M 47824). The right occipital condyle united with a hemivertebra (C0). Arrow indicates an interstice between the skull and hemivertebra. Abbreviation: oc, occipital condyle.

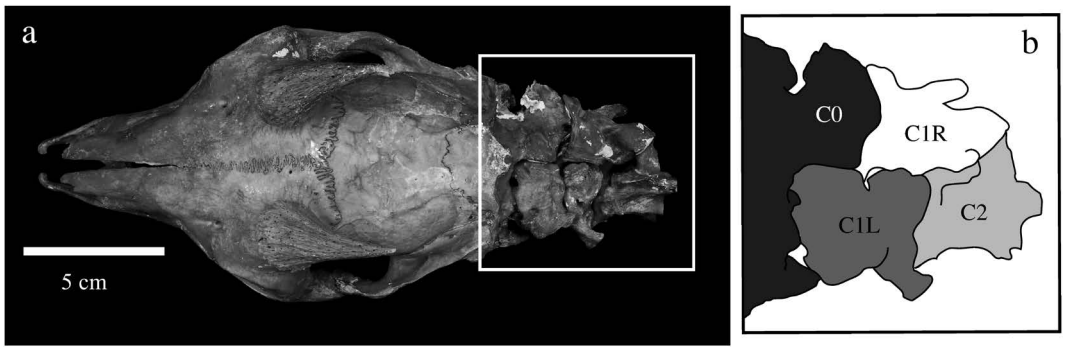

Fig. 2. Morphological characteristics of the occipitoatlantoaxial malformation in this specimen. a, a dorsal view of the articulated skeleton; b, a schematic diagram of the skeleton enclosed a box in 2a.

C1 was not merged at the sagittal plane and then composed of two unfused lateral hemivertebrae (Figs. 2, 3a). The left hemivertebra (C1 L) exhibited a clear interstice at the dorsal area (Fig. 3c). The C1 L possessed the cranial articular facet and connected with the left occipital condyle, and also contacted with the C0 by the medial articular surface. The posterior region of C1 L had the caudal articular facet and the facet for the dens of C2, and it articulated with C2 (Fig. 3a, b). There was a dent with a rough surface at the ventral region of C1 L (Fig. 3b).

C1R exhibited a significantly different morphology from the C1 L. It possessed a dorsal protrusion similar to a neural spine (Fig. 3d). The anterior part of C1R was connected with C0 by the three articular surfaces (Fig. 3e). At the posterior part, there were the caudal articular facet and the facet for the dens of C2 (Fig. 3a). Moreover, the C1R possessed the posterior articular process like a more caudal cervical vertebra (Fig. 3d). The ventral part of C1R engaged with the dent of C1 L (Fig. 3a).

C2 displayed an asymmetric morphology: it possessed the anterior articular process at the right side but did not at left side (Fig. 4a). The neural spine was tilted to the right side. The cranial articular surface of C2 separated into the right and left sides (Fig. 4a). On the other hand, in the normal specimen, the right and left cranial articular surfaces were connected below the dens (Fig. 4c). The cranial articular surface was smaller in the OAAM specimen than in the normal individuals (Fig. 4a, c). Since the posterior part of the vertebra was cut off, the presence of the malformation at the area could not be confirmed.

The joint between the skull and C1R-C1 L complex allowed the dorsoventral movement of the head (Fig. 3). According to the osteological observation, it seemed that a slight rotational movement was possible at the joint between C2 and C1R-C1 L complex (Fig. 4).

Discussion

We described a Japanese serow (NSMT-M 47824) showing serious malformations at the atlanto-occipital and atlanto-axial region (Figs. 1–2). The specimen did not have any obvious morphological anomalies on the skull with exception of the occipital area. Since there are various nerves and vessels in this region, a traumatic injury around here would lead a serious damage to nervous and circular system. Thus, at least, it is hard to explain that the malformations were occurred by a traumatic injury after this individual had grown up. Besides, the C2 possessed the right anterior articular process with a smooth articular surface like a more caudal cervical vertebra. This could not be explained as a result of traumatic skeletal deformation. Based on these evidences, we regarded the craniovertebral malformations in this specimen as a congen-

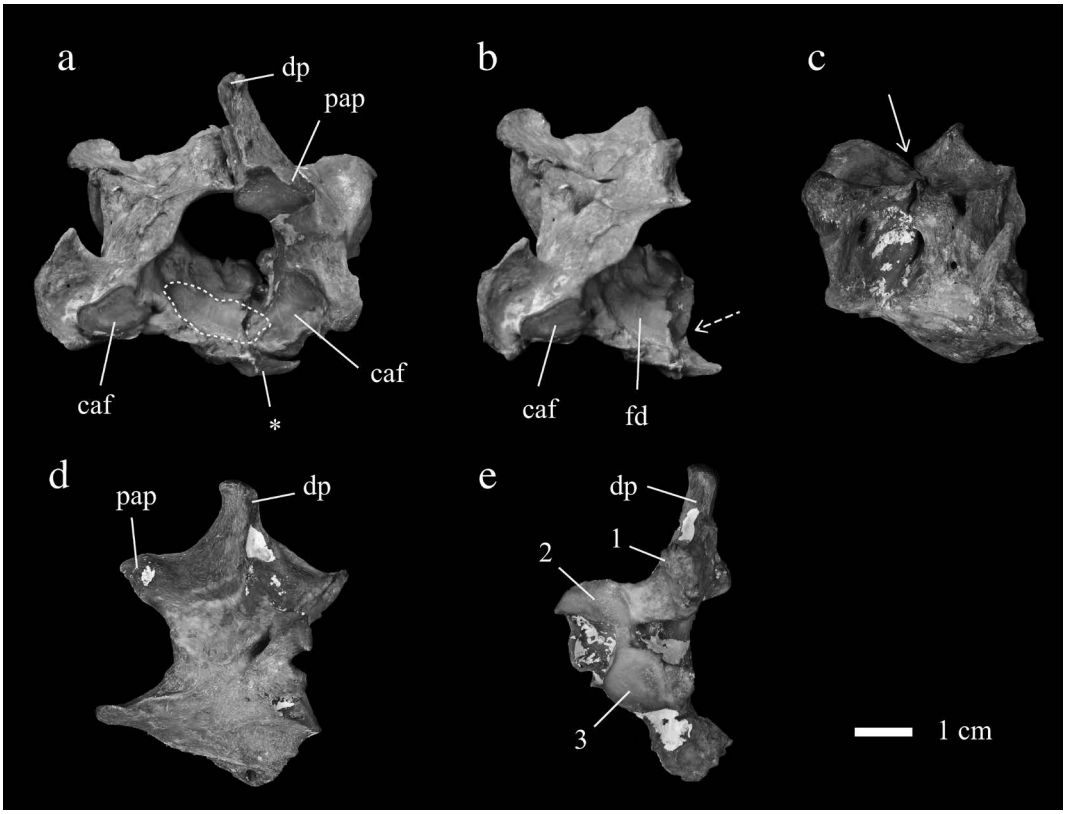

Fig. 3. Morphological characteristics of C1R and C1 L. a, the dorso-caudal view of the C1R–C1 L complex; b, the dorso-caudal view of the C1 L; c, the left lateral view of the C1 L; d, the right lateral view of the C1R; e, the cranial view of the C1R. The region contoured by the dashed line shows the facet for dens. Asterisk indicates that the ventral part of the C1 L covered the ventral protrusion of the C1R. Dashed arrow points the dent of ventral part of C1 L, and arrow indicates an interstice at the dorsal area of C1 L. The numeric characters in (e) means the three articular surfaces connecting to the C0. Abbreviations: caf, caudal articular facet; dp, dorsal protrusion; fd, facet for dens; pap, posterior articular process.

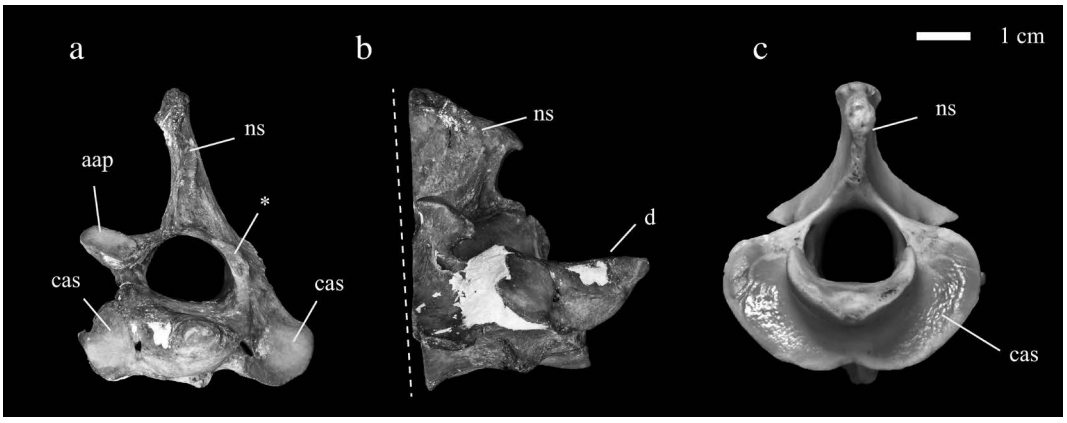

Fig. 4. Morphological characteristics of C2. a, the cranial view of the C2; b, the right lateral view of the C2; c, the cranial view of the C2 in a normal specimen (NSMT-M 37272). Asterisk indicates the articular surface contacting with C1 L. Dashed line shows a cutting position. Abbreviations: aap, anterior articular process; cas, cranial articular surface; d, dens; ns, neural spine.

ital anomaly rather than an acquired traumatic anomaly.

This specimen exhibited the asymmetrical fusion between the occipital condyle and cervical element. The atlanto-occipital fusion is regarded as a representative anomaly of the OAAM in domestic mammals (Schmidt *et al.*, 1993; Vieira *et al.*, 2017, Jayson *et al.*, 2018). Although the OAAM is a rare malformation, it has been reported in various domestic mammals: cattle (Leipold *et al.*, 1972; White *et al.*, 1978), horses (de Lahunta *et al.*, 1989; Vieira *et al.*, 2017), sheep (Schmidt *et al.*, 1993; Kerkmann *et al.*, 2009), goat (Seva *et al.*, 2008), camel (Sakamoto *et al.*, 2004), dog (Galban *et al.*, 2010), and cat (Watson *et al.*, 1985; Jaggy *et al.*, 1991; Uno *et al.*, 2005). This is a first description of the OAAM in the Japanese serow, and a precious case of the OAAM in a wild individual.

This individual was the only specimen showing the OAAM in 14102 skeletal specimens of Japanese serow donated from the Japan Wildlife Research Center and stored in the National Museum of Nature and Science (Tokyo), at this time. Hence, the OAAM is considered as a quite rare malformation in Japanese serow as well as other previously reported domestic mammals (e.g., Vieira *et al.*, 2017; Jayson *et al.*, 2018).

The previous genetic studies using mice revealed that the inactivation of *Hox*d-3 gene expression arose the craniovertebral malformations (Condie and Capecchi, 1993; Pang and Thompson, 2011). The study showed that the inactivation of the gene caused the atlanto-occipital assimilation and a deformed C2 displaying morphological characteristics of the atlas (Condie and Capecchi, 1993). In the examined specimen, the morphological characteristics of the right side of the craniovertebral region were shifted. In addition to the unilateral atlanto-occipital assimilation, the hemivertebra C1R possessed the dorsal protrusion similar to the typical morphology of more caudal cervical vertebrae. Moreover, the C1R was articulated with the right side of the C2 by the articular process like the normal C2–C3 joint. This suggests that the malformations of the specimen were produced by the failure expression pattern of *Hox*d-3 gene at the right side.

In this specimen, the position of the each craniovertebral joint at the right side was shifted by a half of a vertebra, in comparison with left sides (Fig. 2b). In mammals, during the development, the vertebrae are formed by the recombination of neighboring sclerotome halves from adjacent segments (Ward *et al.*, 2017). This process, termed as a resegmentation of sclerotome, causes a half segment shift in the boundaries of the adult vertebrae relative to embryonic sclerotomes (Fleming *et al.*, 2015). Hence, it is inferred that the asymmetrical boundaries were caused by the failure in the resegmentation of the sclerotome in the right side. The failure of the resegmentation at atlanto-occipital region would prevent to separate between an anterior sclerotome half forming the occipital condyle and posterior sclerotome half forming the atlas. This could lead to forming the C0 which fused to the occipital condyle.

The atlas of this specimen comprised of two hemivertebrae might have been caused by an incomplete fusion of the dorsal and ventral arches of the atlas. This malformation is a rare congenital disorder termed bipartite atlas (Senoglu *et al.*, 2007). It is assumed that the malformation would have been caused by a failure fusion of right and left ossification centers involved with the failure of the resegmentation (Hummel and Groot, 2013).

The atlanto-occipital region was significantly deformed in this specimen, nevertheless the joint between the skull and the C1R–C1 L complex permitted the dorsoventral movement of the skull (Fig. 5). Additionally, in this case, the dens was present despite the previous studies often reported the absent of it accompanied with the OAAM (Watson *et al.*, 1986). Our skeletal observation showed that a slight lateral rotation could occur at the joint between C2 and C1R–C1 L complex (Fig. 6).

However, the neural spine of the C2 contacted with the dorsal protrusion of the C1R when the C2 maximally rotated to the right side, and it was

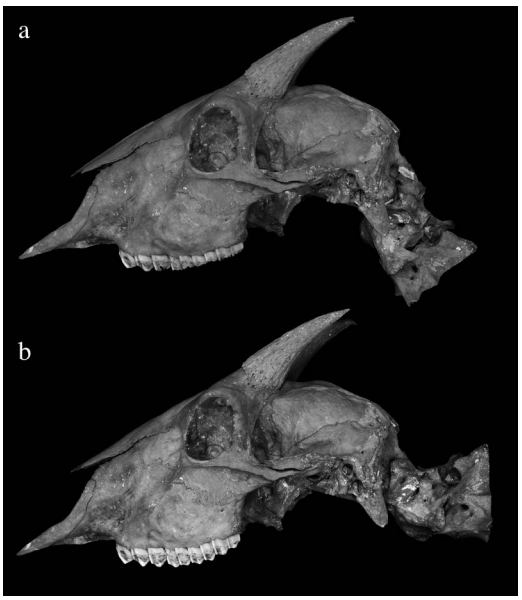

Fig. 5. Left lateral view of the skull and cervical vertebrae in maximally extended pose (a) and maximally flexed pose (b). Note that the joint between the occipital condyle and C1R–C1 L complex possesses a mobility.

separated from the dorsal protrusion of the C1R by about 1 cm when the C2 maximally rotated to the left side. This drastic change of the distance between the neural spine of C2 and the dorsal protrusion of C1R might be restricted by the soft tissue like a muscle and ligament connecting around these spines. It suggests a possibility that the rotational movement of the joint between C2 and C1R–C1 L complex had been fairly small when this individual was alive.

Given the estimated age and the fact of the pregnancy in this specimen as well as the above morphological observation, it is suggested that the specimen had not troubled a severe functional problem at the head-neck joint and had not suffered serious neurological diseases like a tetraparesis.

Acknowledgements

We are grateful to T. Miura (Japan Wildlife Research Center) for donating 14102 skeletal specimens of the Japanese serow, including the individual with the OAAM.

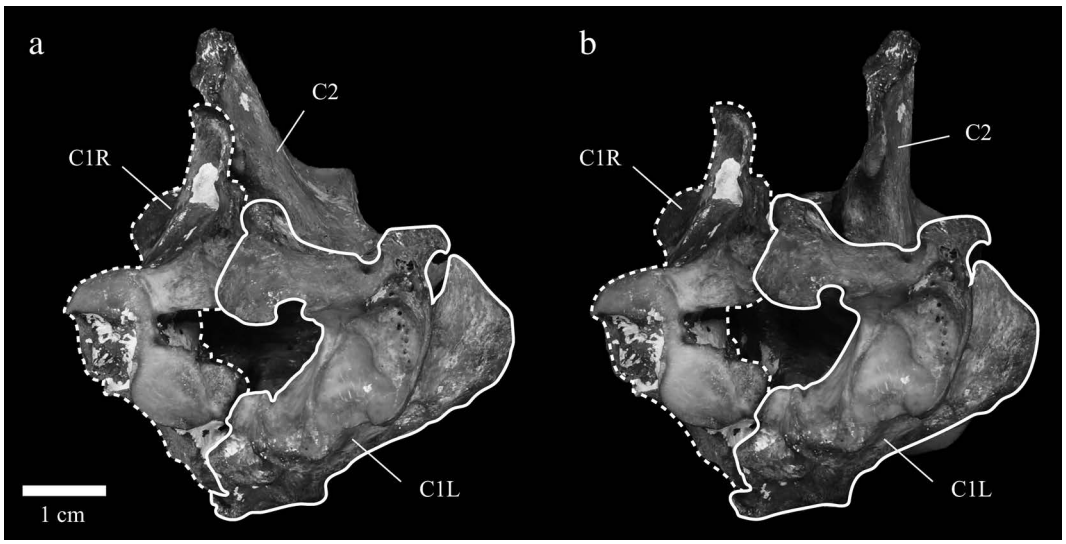

Fig. 6. Cranial view of the C1R–C1 L complex and C2 in maximally right rotated pose (a) and maximally left rotated pose. Solid line shows the outline of C1 L, and dashed line shows that of C1R. Our skeletal observation indicates that the joint between C1R–C1 L complex and C2 possesses a slight mobility. Note that the neural spine of C2 contacts with the dorsal protrusion of C1R in (a), and it was separated from the dorsal protrusion of C1R in (b).

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