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Atlanto-occipital fusion: Case report

Fusão atlanto-occipital: Relato de caso

Fusión atlanto-occipital: Reporte de caso

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Abstract
The article discusses cranio-cervical joints, specifically the atlanto-occipital and atlantoaxial joints. The atlanto-occipital joints include the occipital condyle and the superior articular surfaces of the first cervical vertebra (C1 or Atlas), allowing flexion, extension and lateral tilt of the head. The atlantoaxial joints include C1 and C2 (Axis) vertebrae, enabling rotation. Atlanto-occipital fusion is a rare congenital condition in which C1 fuses with the occipital bone of the skull. This can lead to narrowing of the Foramen Magnum, with or without compression of central structures (spinal cord or brain stem). Although this condition can be asymptomatic, when associated with other cranio-cervical anomalies, such as Basilar Invagination and Platybasia, it can bring instability to the neck, in addition to tonsillar herniation, neural compression and cerebral ischemia. This condition is related to an embryonic malformation of the Pro Atlas, which can occur in different regions of the joint. Furthermore, the type of fusion can vary, eventually resulting in stress at the C1-C2 junction, leading to subluxations and neurovascular complications. Despite its rarity, preoperative diagnosis using computed tomography or magnetic resonance imaging is essential to identify these changes and avoid injuries during surgery. In conclusion, atlanto-occipital fusion is a rare condition that, when associated with other anomalies, can cause damage to the central nervous system, highlighting the importance of adequate diagnosis and treatment. The objective of the study is to illustrate, discuss and investigate the prevalence of atlanto-occipital fusion in human skulls, which the authors found one case among the 117 skulls analyzed.
Keywords: Atlanto-occipital function; Assimilation of the atlas; Vertebral artery variation.

Resumo
O artigo discute as articulações crânio-cervicais, especificamente as articulações atlanto-occipital e atlantoaxial. As articulações atlanto-occipital incluem o côndilo occipital e as superfícies articulares superiores da primeira vértebra cervical (C1 ou Atlas), permitindo flexão, extensão e inclinação lateral da cabeça. As articulações atlantoaxiais incluem as vértebras C1 e C2 (Eixo), permitindo a rotação. A fusão atlanto-occipital é uma condição congênita rara em que C1 se funde com o osso occipital do crânio. Isto pode levar ao estreitamento do Forame Magno, com ou sem compressão de estruturas centrais (medula espinhal ou tronco cerebral). Embora esta condição possa ser assintomática, quando associada a outras anomalias crânio-cervicais, como Invaginação Basilar e Platibasia, pode trazer instabilidade ao

pescoço, além de hérnia tonsilar, compressão neural e isquemia cerebral. Essa condição está relacionada a uma malformação embrionária do Pró Atlas, que pode ocorrer em diversas regiões da articulação. Além disso, o tipo de fusão pode variar, resultando eventualmente em estresse na junção C1-C2, levando a subluxações e complicações neurovasculares. Apesar de sua raridade, o diagnóstico pré-operatório por meio de tomografia computadorizada ou ressonância magnética é essencial para identificar essas alterações e evitar lesões durante a cirurgia. Concluindo, a fusão atlanto-occipital é uma condição rara que, quando associada a outras anomalias, pode causar danos ao sistema nervoso central, destacando a importância do diagnóstico e tratamento adequados. O objetivo do estudo é ilustrar, discutir e investigar a prevalência da fusão atlanto-occipital em crânios humanos, onde os autores encontraram um caso entre os 117 crânios analisados.
Palavras-chave: Função atlanto-occipital; Assimilação do atlas; Variação da artéria vertebral.

Resumen
El artículo analiza las articulaciones cráneo-cervicales, específicamente las articulaciones atlantooccipital y atlantoaxial. Las articulaciones atlantooccipital incluyen el cóndilo occipital y las superficies articulares superiores de la primera vértebra cervical (C1 o Atlas), lo que permite la flexión, extensión e inclinación lateral de la cabeza. Las articulaciones atlantoaxiales incluyen las vértebras C1 y C2 (Axis), lo que permite la rotación. La fusión atlantooccipital es una afección congénita poco común en la que C1 se fusiona con el hueso occipital del cráneo. Esto puede provocar un estrechamiento del agujero magno, con o sin compresión de estructuras centrales (médula espinal o tronco del encéfalo). Aunque esta condición puede ser asintomática, cuando se asocia con otras anomalías cráneo-cervicales, como la Invaginación Basilar y Platibasia, puede traer inestabilidad al cuello, además de hernia amigdalina, compresión neural e isquemia cerebral. Esta condición está relacionada con una malformación embrionaria del Pro Atlas, que puede ocurrir en diferentes regiones de la articulación. Además, el tipo de fusión puede variar, lo que eventualmente resulta en estrés en la unión C1-C2, lo que provoca subluxaciones y complicaciones neurovasculares. A pesar de su rareza, el diagnóstico preoperatorio mediante tomografía computarizada o resonancia magnética es fundamental para identificar estos cambios y evitar lesiones durante la cirugía. En conclusión, la fusión atlantooccipital es una condición poco común que, cuando se asocia con otras anomalías, puede causar daño al sistema nervioso central, resaltando la importancia de un diagnóstico y tratamiento adecuado. El objetivo del estudio es ilustrar, discutir e investigar la prevalencia de la fusión atlantooccipital en cráneos humanos, de la que los autores encontraron un caso entre los 117 cráneos analizados.
Palabras clave: Función atlantooccipital; Asimilación del atlas; Variación de la arteria vertebral.

1. Introduction

The cranio-cervical joints encompass two groups: the first is called atlanto-occipital joints, consisting of the first cervical vertebra C1 (also known as Atlas) and the skull, responsible for connecting the cervical spine to the skull. This joint is composed of the occipital condyle and the superior articular surfaces of C1, allowing flexion and extension movements of the head (approval movement), in addition to enabling lateral tilting of the head, such as slight flexion and rotation movements. This is a condylar-type synovial joint (Moore, 2019). The second group is called atlantoaxial joints, formed by 3 joints, made up of the C1 and C2 vertebrae (also known as the axis). These joints are separated into lateral atlantoaxial joints (left and right, from the lower articular surfaces of C1 to the upper articular surfaces of C2), these being flat synovial joints; and the median atlantoaxial joint (between the odontoid process of C2 and the anterior arch of C1), this being a trochoid joint (Moore, 2019). Because of this, this group is the least stable of the spine due to its greater mobility, as they allow the head to perform a rotation movement (rejection movement), in which the skull and C1 rotate as a single unit about the odontoid process of C2 (Netter, 2013).

Atlas assimilation (or atlanto-occipital fusion) is a rare congenital osseous or fibrous anomaly with an incidence ranging from 0.14% to 0.75% of the general population, affecting both sexes equally (Natsis et al., 2017; Piplani & Kullar, 2017), furthermore, this incidence may vary from 0.5% to 1.0% in caucasians (Al-Motabagani & Surendra, 2006), being the most common in the cranio-cervical junctions, which has an incidence of 0.08 to 3.63% (Kim, 2015). Furthermore, the literature shows that the condition can also occur as a result of tumors and infections in the craniocervical junction, as well as due to post-traumatic deformities (Abumi et al., 2010). However, in these cases where the condition does not have a congenital origin, the most correct term to describe it would be “synostosis” (Canelas et al., 1952). It is characterized by the complete or incomplete fusion of the Atlas vertebra with the occipital bone, causing the narrowing of the foramen magnum and, therefore, compression of the canal and spinal cord or even the brain stem (Sharma et al., 2017). In isolation, this condition is asymptomatic and may go unnoticed.

However, when associated with other cranio-cervical junctions, such as Basilar Invagination, Platybasia and variations in the vertebral artery, it can cause numerous symptoms with a mild onset in childhood, progressing with aging to more significant symptoms, which may include instability in the neck with decreased movement, tonsillar herniation, spinal cord compression and cerebral ischemia (Joaquim et al., 2021; Menezes & Dlouhy, 2020).

The objective of the study is to illustrate, discuss and investigate the prevalence of atlanto-occipital fusion in human skulls, which the authors found one case among the 117 skulls analyzed.

2. Methodology

This article aims to describe an anatomical variation found during a routine analysis of an anatomical specimen from the Anatomy Laboratory of Centro Universitário Barão de Mauá de Ribeirão Preto, São Paulo, in which the presence of fusion between the occipital bone and the atlas vertebra. This fact prompted the study to investigate the prevalence of this fact in which fusion occurs between the occipital bone and the atlas vertebra in the skulls that are available in the Anatomy Laboratories of this institution as well as the description of the possible surgical and clinical outcomes of this fact. The study was carried out preserving the ethical principles of the Declaration of Helsinki and observating protocols of use of human cadaveric tissues in anatomy research papers (Iwanaga et al., 2022) as the skulls used in this study cannot be identified (all obtained from non-identifiable corpses dissected along the years of the anatomy department, since the academic center foundation - all of them properly stored and preserved for the practical anatomy classes).

We found a total of 117 (one hundred and seventeen) human skulls (of both sexes) at the laboratory; all of them were analyzed by four medical school professors (three of the same faculty and one external - coauthors of this article). Only one skull was found with the fusion and media records has been made - outside the foramen magnum, inside the posterior fossa (through this foramen with all angles) and inside the oral and nasal cavity (searching for possible concomitant malformations, such as cleft palate - that were not found).

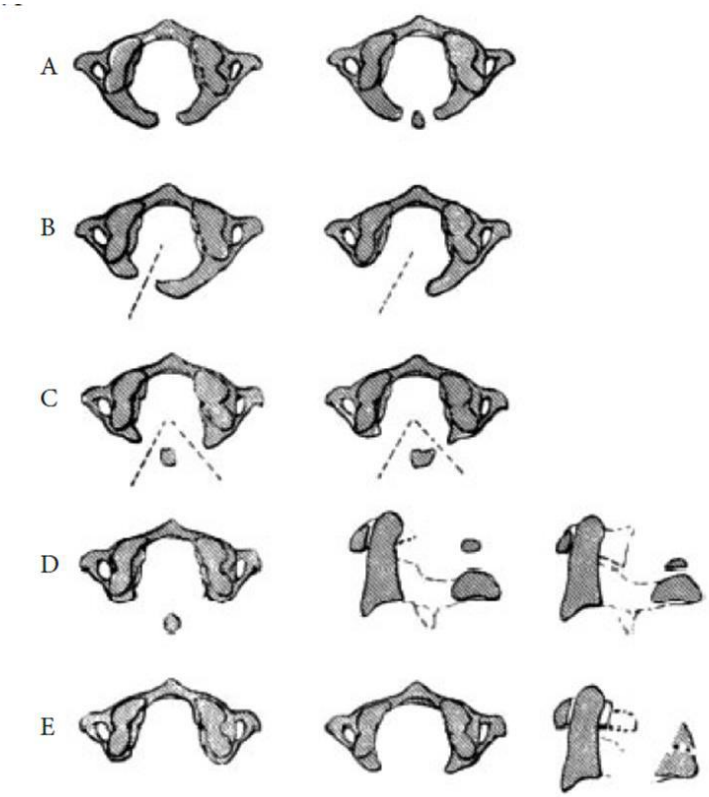
After photographic documentation and properly storage of the media (ensuring data security, only used for this academic publication), a non-systematic review of the literature was carried out in the most relevant databases (Pubmed/Medline, Scielo, Embase and Scopus) in search of similar cases similar cases and for accessing other pathologic variations, radiologic features and potential clinical consequences regarding the aforementioned fusion - which was used for composing this publication.

3. Results and Discussion

After evaluating human skulls from the anatomy laboratories at Barão de Mauá University Center, only one skull was detected with the anatomical variation of atlanto-occipital fusion. This condition is secondary to an embryonic malformation between the 4th occipital sclerotome (Pro atlas) and the 1st cervical sclerotome, which can cause narrowing of the foramen magnum, interfering with the entire development of this region, affecting vital vascular and neural structures, as well as the mobility of the cervical spine (Joaquim et al., 2021; Kim, 2015; Menezes & Dlouhy, 2020; Piplani & Kullar, 2017).

This condition may be classified into 4 types according to the region where it occurred; in region 1, the anterior arch of the atlas is in front of the lateral masses (20% of cases); in region 2, it occurs in the lateral processes (17%); in region 3, the posterior arch of the atlas is behind the lateral masses (13%); and finally, the combination of the 3 regions that represents 50% of cases. When malformation of the posterior arch occurs, it can be classified into 5 types represented in Figure 1 and Figure 2, with “A” being the mildest classification, with only a small fistula or gap in the arch; “B” the absence of one of the arch arms; “C” bilateral defect with preservation of the dorsal region of the arch; “D” absence of the posterior arch with the presence of the posterior tubercle; and “E” the complete absence of the posterior arch and tubercle (Kim, 2015; Tubbs et al., 2011).

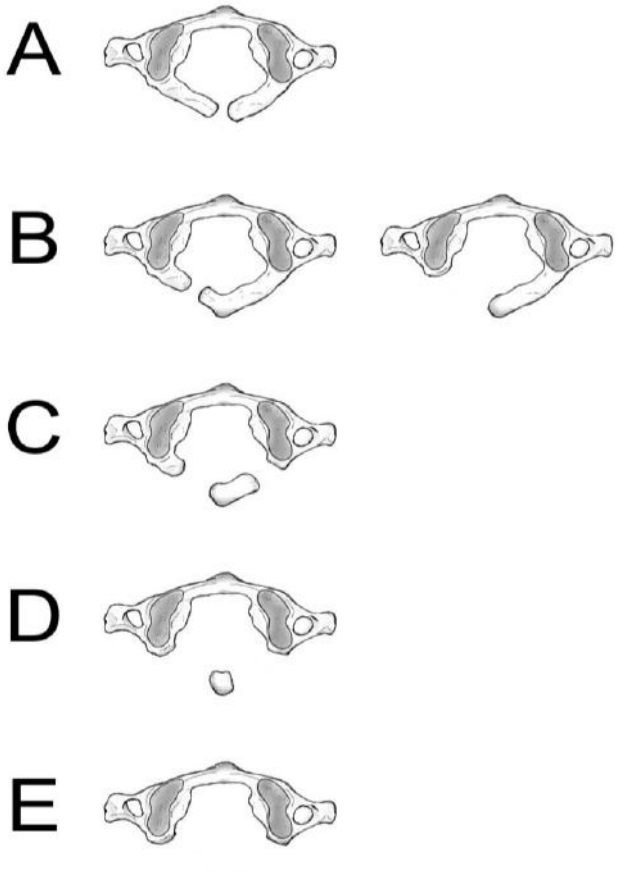
Figure 1 - Classification of types of malformation of the posterior arch of the Atlas.



Source: Kim (2015).

The diagram illustrates the various forms of atlas malformation in relation to the posterior arch. It may present variation A, characterized by a deficiency in the posterior arch of the atlas, leading to the inability of the posterior hemiarchs to fuse. Variation B displays an opening of varying size in the posterior arch, ranging from a small gap to the total absence of a hemiarch. Variation C shares similar characteristics with type B but includes the presence of the posterior tubercle with a portion of the posterior hemiarch. Variation D is distinguished by the complete absence of the posterior arches, accompanied only by the presence of the posterior tubercle. Finally, variation E implies the complete absence of the posterior arch and tubercle.

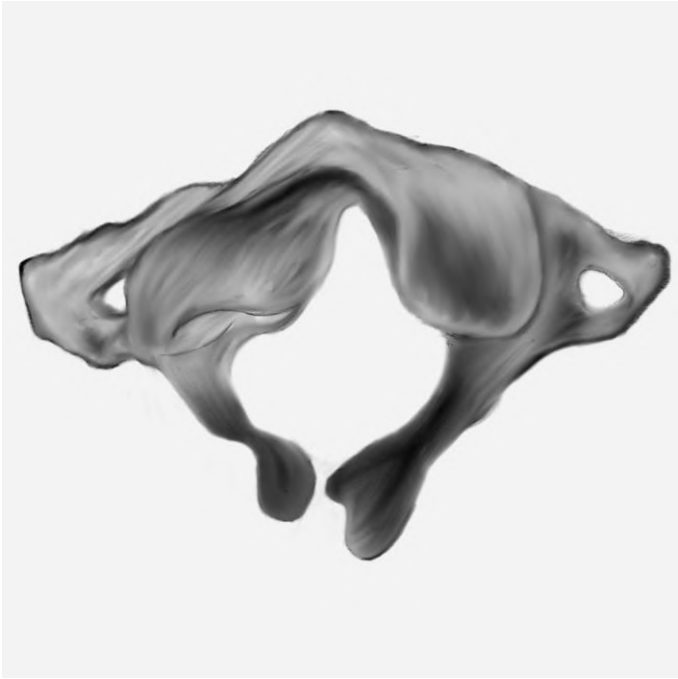
Figure 2 - Classification of types of malformation of the posterior arch of the Atlas.



Source: Authors (2023).

To analyze the type of the variation, it is necessary to observe Figures 3 and 4.

Figure 3 - Schematic drawing of the Atlas vertebra found in the reported skull.

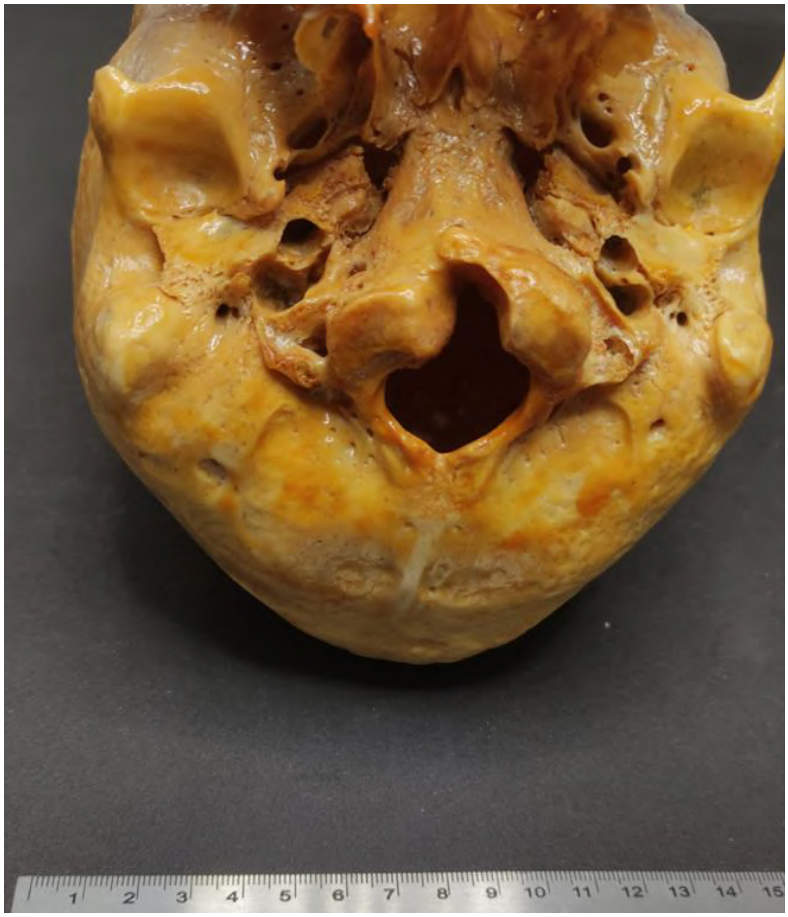


Source: Authors (2023).

The drawing presents a representation of the atlanto-occipital fusion found, exemplifying the malformation of the posterior arch.

In this condition, the laboratory skull (Figure 4) presents a combination of the atlanto-occipital fusion regions, with a complete fusion of the anterior arch and incomplete fusion of the posterior arch presenting type B, with unilateral fissure, which is a small gap in the right hemiarch and with the absence of a posterior tubercle. The right and left transverse processes are partially fused to the occipital bone, so that both present the transverse foramen. Furthermore, both articular processes of the Atlas and the occipital condyles are fused and the condyles are entering the foramen magnum.

Figure 4 - Skull with atlanto-occipital fusion found in the Anatomy laboratory of Barão de Mauá University Center.



Source: Authors (2023).

The photo shows the anatomical piece with the variation that is being described, which may highlight the atlanto-occipital fusion, as well as the malformation of the posterior arch of the atlas.

In isolation, this anatomical variation can be asymptomatic and undetected, only being discovered in radiological research in contexts of trauma or other neurological and orthopedic conditions (Joaquim et al., 2021; Kim, 2015). However, this condition can often be investigated early in the physical examination by testing hyperflexion and hyperextension of the neck, which will present a small limitation of movement (Chi et al., 2022). Greater stress on the C1 – C2 junction is reported in the literature due to the impossibility of movement between the skull and the Atlas, the first mobile segment of the spine, with consequent stretching of the transverse ligament in relation to the odontoid process and gradual loosening of this – evolving into a progressive subluxation of the atlanto-axial joint, favoring the displacement of the odontoid process and resulting in the perpetuation of synostosis through an inflammatory process. Furthermore, this condition can lead to a reduction in the foramen magnum by itself (Kim, 2015; Piplani, & Kullar, 2017; Sharma et al., 2017).

The area of the foramen magnum (Sfm) can be calculated using Radinsky's formula (1967), given by Sfm = π x Dt x Dap x 1/4, where Dt is the greatest transverse diameter and Dap is the greatest anteroposterior diameter of the foramen magnum (Pires et al, 2016). The expected Sfm is approximately 7.83 cm² (Cirpan et al, 2016) using Radinsky's formula. In the analyzed skull, the measured values for Dt were 2.8 cm and for Dap were 3.3 cm, resulting in an Sfm of 7.25 cm², considering π = 3.14 cm. However, due to atlanto-occipital fusion, there was a reduction of 1.5 cm in Dap, resulting in an actual surface area of 3.95 cm². Therefore, we estimate a 46% reduction in the foramen magnum area.

This condition can also compromise the cranio-cervical ligaments and membranes. These structures play a crucial role in providing structural support and preventing excessive or unnatural movements, which can result in serious injuries. The main ligaments associated with the atlanto-occipital junction are the Anterior and Posterior Atlanto-Occipital Ligaments, connecting the occipital bone to the atlas joint, playing a significant role in stabilizing the region and preventing excessive movements, especially in the flexion and extension of the head. Additionally, the Tectorial Membrane and the Cruciform Ligament are also fundamental for atlanto-occipital stabilization, offering additional support to the joint. Thus, compromising this cranio-cervical stability can impact the functional integrity of the joints, making it more challenging to perform coordinated movements of the neck and head (Debernardi et al., 2011; Natsis et al., 2017).

Added to this, neck joint instability itself can cause injuries to some structures of great importance, such as the vertebral artery, the first spinal nerve, the hypoglossal nerve and the spinal cord, but which in the first decades of life can be compensated by neuroplasticity. and neuroregeneration (Piplani, & Kullar, 2017). However, the chance of regeneration in cases of repeated injuries decreases with age; therefore, patients between the ages of 30 and 40 may begin to develop symptoms related to neural compression or spinal instability, making surgical intervention necessary to decompress the fusion (Joaquim et al., 2021; Kim, 2015). In these cases, this condition is of great clinical importance due to the possibility of compromising these structures, which can cause compression of the spinal cord and brainstem, producing numerous signs and symptoms, from a simple headache or muscle weakness to dysarthria, focal deficits, symptoms of low output or death (Kim, 2015; Piplani, & Kullar, 2017). The coexistence with other variations in the cranio-cervical junction can lead to a heterogeneity of symptoms, especially Arnold-Chiari Syndrome, Klippel-Feil Syndrome and variations in the vertebral artery, where there is a worse prognosis[2] (Kim, 2015). In the first case, there may be tonsillar herniation due to the existence of a connection between the back of the head and the spinal column; In the second, due to the fusion of two or more cervical vertebrae (the most common being C2 - C3) which contributes to atlantoaxial displacement, there may be stenosis of the cervical canal and basilar invagination. According to Wang et al. (2009), variations in the vertebral artery are present in most cases of atlanto-occipital fusion and can be of 4 types depending on the path of the artery through the occipitalized Atlas: in type 1 it follows below the lateral mass; in type 2 it passes posteriorly to the surface of the lateral mass; in type 3 it creates a foramen between the fused atlas and occipital bone, to reach the skull; finally, in type 4, it is absent on the side of the vertebra.

Furthermore, according to Skrzat et al. (2011), the abnormal passage of the vertebral arteries through the transverse foramina can lead to compressions, leading to a reduction in their lumen and compromising blood flow, causing symptoms such as dizziness, convulsions, mental flexibility and syncope. In more severe cases, it can cause ischemic symptoms, vertebrobasilar insufficiency and brain stem hypoxia.

4. Conclusions

Despite the low incidence of posterior atlas arch defects, we recommend that surgeons keep the possibility of their existence in mind when approaching cases of traumatic atlantoaxial subluxation and atlas fracture. Preoperative diagnosis is extremely important to detect these changes and avoid neurovascular injuries during surgery. Preoperative 3D computed tomography or magnetic resonance imaging should be carefully analyzed for better surgical planning.

Due to the low incidence and underdiagnosed cases of this condition, there is a lack of studies with high levels of evidence. Moreover, the multiple morphologic patterns of this fusion causes another theoretical research problem: difficulties on standardizing interventions and groups for controlled studies. It is interesting to point out that the presence of fusion may hinder suboccipital punctures for CSF analysis (when lumbar puncture is contraindicated) and other surgical procedures of the spinal cord (for example biopsies, neoplasm resections, laminectomies etc). This superposition of diseases and complications may be even more challenging for decision making and may take even more time to be clarified.

In the literature, several ways of managing patients with atlanto-occipital fusion are discussed, given the heterogeneity of symptoms and coexisting conditions. To treat patients who have symptomatic atlanto-occipital fusion, decompression of the foramen magnum must be performed, evaluating each case individually (Joaquim et al., 2021; Kim, 2015). Up to now, there's no indication of surgery for the asymptomatic fusions per se, but for the associated symptomatic malformations such as Chiari I, Arnold-Chiari and foramen magnum stenosis (Kagawa et al., 2006; Kassim et al., 2010).

The authors sincerely thank those who donated their bodies to science so that anatomical research could be performed. Results from such research can potentially increase mankind's overall knowledge that can then improve patient care. Therefore, these donors and their families deserve our highest gratitude (Iwanaga et al., 2022).

5. Suggestions for Further Research

Due to the diagnostic challenges mentioned above, it is possible that the fusion is more common than we thought. Several complementary exams compose the newborn evaluation (such as the Phenylketonuria PKU - test), but there is no mention, in our review, of active screening for spinal cord malformations in babies with cleft palate (a possible comorbid condition), neither studies of cost-effectiveness recommending (or not) spinal cord radiologic images at this age. More case series publications are necessary to clarify an eventual demand for new clinical protocols, maybe a multi-centric screening involving necropsy laboratories, obstetric clinics and anatomy or orthopedic university departments. A larger number of cases would encompass case-control studies (for example: Is the absence of prenatal vitamin supplementation a risk factor for the fusion? Maybe mother age, or drugs/medications during pregnancy?). The same is applicable for radiology clinics, for example searching for fusions accidentally found in CT/MRI performed for other reasons - and orthopedic clinics, on screening other malformations in patients who undergo foramen magnum stenosis surgery.

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