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Posterior arch defect of the atlas associated to absence of costal element of foramen transversarium from 16th century Sardinia (Italy) --Manuscript Draft--

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Historical Perspective
Congenital disease; spondyloschisis; open foramen transversarium; Modern Age; Sardinia; paleopathology.
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Response

Posterior arch defect of the atlas associated to absence of costal element of foramen transversarium from 16th century Sardinia (Italy)

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Abstract

Study Design. A paleopathological case of posterior arch defect of the atlas associated to absence of costal element of the foramen transversarium.

Objective. In living patients as well as in post-mortem analysis it should be difficult to distinguish between a congenital and an acquired anomaly. Any anomaly in the anatomy of atlas should be taken into consideration by clinicians, surgeons, radiologists and anatomists in order to avoid misinterpretations and clinical complications.

Summary of Background Data. Posterior arch defect has a current occurrence of approximately 4%. Posterior arch schisis is attributed to the defective or absent development of the cartilaginous preformation of the arch rather than a disturbance of the ossification. Absence of costal element of the foramen transversarium has an incidence of ranging from 2% to 10% and is attributed to a developmental defect or to variations in the course of the vertebral artery.

Methods. The skeleton of a male aged 20-30 years, brought to light in the plague cemetery of 16th

century Alghero (Sardinia), showed anomalies of the atlas, consisting in failure of the midline

fusion of the two hemiarches with a small gap and an open anterior foramen trasversarium on the

left side. A macroscopic, radiological and stereomicroscopic study was carried out.

Results. The study allowed to rule out a traumatic origin of the defects and to diagnose

an association of two congenital anomalies.

Conclusions. Osteoarchaeological cases provides with a valuable opportunity to examine and describe variations in the anatomy of the atlas.

Article type: Historical Perspective

Key words: Congenital disease, spondyloschisis, open foramen transversarium, Modern Age, Sardinia, paleopathology

Key points:

- In living patients as well as in post-mortem analysis it should be difficult to distinguish between a congenital and acquired anomaly in the anatomy of the atlas.
- A case of posterior arch defect associated to absence of the costal element of the foramen
 transversarium was found in a skeleton from 16th century Alghero (Sardinia)
- Macroscopic, radiological and stereomicroscopic study was carried out, providing with a valuable opportunity to examine and describe anomalies of the atlas.

A case of posterior arch defect of the atlas associated to absence of costal element of the foramen transversarium was detected in a skeleton from 16th century Sardinia. Osteoarchaeological cases provides with a valuable opportunity to examine and describe anomalies of the atlas.

1 Introduction

2 Congenital defects of atlas are rare conditions and include several anatomical variations,

3 which should be considered in the clinical practice in order to avoid complications.¹

Posterior spondyloschisis of atlas is an incidental finding, usually identified in investigations after trauma.² Currarino *et al.*³ proposed a classification of posterior arch defects in five types (A–E), depending on the extent of absence of the posterior arch and the presence or absence of the posterior tubercle. Type A consists in a failure of midline fusion of the two hemiarches, with a small gap remaining; in Type B there is unilateral cleft ranging from a small defect to the complete absence of one hemiarch; Type C is the bilateral clefts of the lateral aspects with preservation of the most dorsal part of the arch; Type D is described as the absence of the posterior arch with a persistent posterior tubercle; finally, Type E is the most severe condition, consisting in absence of the entire arch including the tubercle. In living patients as well as in post-mortem analysis it should be difficult to distinguish between a posterior arch defect and an atlas fracture.4,5 The costal elements of the foramen transversarium sometimes remain deficient, resulting in an anterior aperture of the foramen. This variation might affect the trajectory of the vertebral artery.¹ Therefore, anomalies in the anatomy of atlas are of paramount importance to clinicians, surgeons, radiologists and anatomists in order to avoid misinterpretations and clinical

20 complications.

Osteoarchaeological cases provides with a valuable opportunity to examine and describe
anomalies of the atlas.

Materials and methods

Archaeological excavations carried out in the courtyard of the former College of the Jesuits in
San Michele - Lo Quarter in Alghero (Sardinia) brought to light the skeletal remains of 200
individuals.⁶ Both archaeological dating and burial modalities strongly suggest that the
skeletal remains belong to the victims of the plague that hit Alghero in 1582-1583.
A skeleton (code number: 2219) exhumed from trench 4, containing 18 individuals, showed
anomalies of the atlas. Anthropological study was performed according to standard methods.⁷
Macroscopic analysis was followed by radiological and stereomicroscopical study.

Results

The skeletal remains belong to a male aged 20-30 years old, 161 cm tall.

35 The first cervical vertebra shows a congenital defect of the posterior arch. A failure of the

36 midline fusion of the two hemiarches with a small gap is evident (fig. 1). The vertebra shows

a post-mortal breakage in correspondence of the left articular facet and therefore it is difficult

to exactly measure the gap. However, the two parts of the posterior arch are almost

39 completely developed and therefore only a small defect of a few millimetres is detectable.

40 In addition, absence of the costal element of the left foramen trasversarium is observed,

41 resulting in an open anterior foramen trasversarium.

At X-ray the edges of the posterior arch appear regular and smooth with an intact cortical wall

(fig. 2). Stereomicroscope magnification (7x) confirmed an intact cortical surface (fig. 3).

Discussion

46 Macroscopic, radiological and stereomicroscopic study allowed to rule out a traumatic origin 47 of the posterior arch defect observed in the skeleton from Alghero, as a fracture should have 48 showed irregular edges, while congenital clefts are smooth with an intact cortical wall as in 49 this case.

According to the classification of Currarino *et al.*³ this is a Type A arch defect. In a clinical study carried out on 1104 patients, 3.35 % showed congenital defect of the posterior arch and among them, 2.6% were represented by type A defect.⁸ Another clinical study demonstrated similar results, as on 1069 patients, 3.8 % showed atlas arch defect, and among them, 3.2 % were represented by type A defect.⁹ The pathogenesis of atlas defects are not yet fully understood. Posterior arch defects are attributed to the defective or absent development of the cartilaginous preformation of the arch rather than a disturbance of the ossification.¹⁰ Congenital posterior arch defects are generally asymptomatic and are considered benign anatomical variations; however, in asymptomatic individuals, they may become dangerous in the context of trauma;¹¹ in these cases it is important to differentiate between a bony injury and a congenital anomaly to rightly evaluate the differences in treatment.² The other defect observed in the atlas of the skeleton from Alghero affects the left foramen transversarium. Foramina transversaria of the cervical vertebrae permit the passage of the vertebral artery and consist of anterior and posterior parts; anterior portion is homologue of the rib in thoracic region, and therefore named as costal process or element.¹² Scanty literature is available about absence of the costal element of the foramen transversarium and its significance; recent studies carried out on collections of atlas demonstrated an incidence ranging from $2\%^{12}$ to $10\%^{13}$; in unilateral defects there is a prevalence for the right side. This variation is attributed to a developmental defect or to variations in the course of the vertebral artery.¹² Neurosurgeons and radiologists should be aware of variations of foramen transversarium as it may expose the vertebral vessels, which are at risk of being damaged. Association of posterior arch defect and absence of costal element of the foramen transversarium has never been described, making the present case worth mentioning.

75	The small number of published archaeological cases of congenital anomalies of atlas makes
76	any report important. The case presented in this study should help in performing a correct
77	study of dry atlas.
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1 Figures Legends

- 2 Figure 1 Atlas of skeleton 2219 from Alghero: superior view (a); inferior view (b)
- 3 Figure 2 AP projection of the atlas
- 4 Figure 3 The two ends of posterior arch at the stereomicroscope (7x)

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