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Transverse basilar cleft: two more probable familial cases in an archaeological context
 Running title: Transverse basilar cleft: probable familial cases

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Abstract

Transverse basilar cleft (TBC) is a very rare congenital anomaly representing a coronal fissure completely or partially intersecting the basioccipital at the level of the pharyngeal tubercle. It is usually asymptomatic, but can sometimes be part of a syndrome. Population frequencies of this anomaly are no more than 1%, with few exceptions. Two skulls with TBC were found in the Ayrydash 1 cemetery of Altai Mountains nomads, Russia, attributed to the Hunno-Sarmatian period (2nd c. BC – 5th c. AD). In this paper, we test the possibility of a familial relationship between the individuals who shared by this condition, using the Alt and Vach method for non-spatial analysis of skeletal kinship based on cranial and dental non-metric traits. Results of the study show that the frequency of TBC is significantly higher in the Ayrydash 1 sample than in the rest of the skeletal population from the Hunno-Sarmatian period ($p < 0.05$). This suggests that the individuals affected by TBC were most likely genetic relatives. The observed clustering of TBC and some other rare anomalies within local groups of Altai Mountains nomads appears to be due to parental consanguinity in their community.

Key words: Altai, cranial congenital anomalies, Hunno-Sarmatian period, kinship analysis, proatlas, transverse basilar cleft.

Introduction

The craniovertebral junction (CVJ) is a phylogenetically and ontogenetically unstable region of the axial skeleton, in which numerous anatomical variants, anomalies and malformations can occur (Schmorl, Junghanns, 1971). The most common of these are manifestations of the proatlas and assimilation of the atlas into the basiocciput. In embryogenesis, the basiocciput is formed from amalgamation of the first four sclerotomes, three of which have merged at an earlier stage. The fourth develops as a transitional vertebra (Müller & O'Rahilly, 2003) called the proatlas, because in lower vertebrates, including reptiles and dinosaurs, it remains as an independent element (Hayek, 1924; Jenkins, 1971). The proatlas forms the anterior margin of the foramen magnum and the occipital condyles. Its failure to integrate with other occipital sclerotomes can result in such anomalies as precondylar tubercle, third occipital condyle, hypochondral arch, paracondylar process, hypoglossal canal bridging, or transverse basilar cleft (Prescher, 1997). The latter represents a coronal fissure partially or completely intersecting the basioccipital at the level of the pharyngeal tubercle (Figure 1). This anomaly is very rare in clinical and skeletal samples. Clinically, transverse basilar cleft (TBC) is usually asymptomatic (Lombardi, 1961; Schmidt et al., 1978; Johnson & Israel, 1979), however,

it sometimes appears as a part of syndromes (List, 1941; Johnson & Israel, 1979; Woon et al., 1980; Whitehead et al., 2015; Mahdi & Whitehead, 2017).

In archaeological contexts, rare congenital anomalies can be useful for identifying closely related individuals within a site (Alt & Vach, 1992; Case et al., 2017). As a familial trait, TBC has been described in two full-term fetuses from a tomb in medieval Apulia, Italy (Anderson, 2000). In this paper, we test whether two adult individuals showing this anomaly from a single burial site in Altai, Russia, attributed to the Hunno-Sarmatian period, could be genetic relatives.

Materials and Methods

In total, more than 650 skulls of adults and children from burial sites in the Altai Mountains have been examined. The remains belong to nomads of the Pazyryk Culture from the Scythian period (5th-3rd c. BC) and the Bulan-Koba Culture from the Hunno-Sarmatian period (2nd c. BC – 5th c. AD).

Transverse basilar cleft was visually scored as present, absent, or, when the basioccipital was missing/severely damaged, as unscorable. The trait was recorded as present at any degree of expression. Frequency of this anomaly was calculated as the ratio of the number of skulls with the defect, to the total number of scorable ones.

Two cases of TBC were found among skulls from the Bulan-Koba burial site of Ayrydash 1 dated to the second half of the 3rd – the end of the 4th c. AD (Seregin & Matrenin, 2014; Figure 2). This cemetery has been completely excavated. The graves were clustered in groups of several rows orientated from north-west to south-east. The majority of burials were single inhumations (Soenov, 2003). Graves #38 and #108, in which the individuals affected by TBC were buried, were located in different sectors of the cemetery. There were no local distinctions in the burial rite within the site. Both skulls with TBC were damaged, the postcranial skeletons were unavailable for the study. Sex and age of the individuals were determined using standard osteological methods (Alekseev & Debec, 1964; Buikstra & Ubelaker, 1994). Sexual dimorphic cranial features, including the supraorbital ridge/glabella, supraorbital margins, slope of forehead, inion protuberance, nuchal crest, mastoid processes, mental eminence, mandibular ramuses, were estimated, taking into account their overall variability within the general population of Altai from the Hunno-Sarmatian period.

The Alt and Vach (1992) method for non-spatial analysis of skeletal kinship based on cranial and dental non-metric traits was applied, by comparing frequency of TBC in Ayrydash 1 (n=59) to that from a reference sample. The reference sample consisted of 293 skulls from other Bulan-Koba burial sites. Statistical testing for frequency differences between these samples was conducted using Fisher's Exact Test.

Results

Both individuals died at a mature age. Skull from grave #38 was determined with confidence as female. Skull from grave #108 exhibits controversial combination of sexual dimorphic features, in which, however, feminine ones prevail.

In both skulls, the basioccipital is divided by coronal lateral fissures into two portions, anterior and posterior, which are connected only by a median bridge. In one of them, the bridge was broken post-mortem, and the portion of the basioccipital between the coronal fissures and sphenobasilar symphysis was lost [Figure 3(A)-3(B)].

The frequencies of TBC are 3.4% (2/59) and 0% (0/293) in Ayrydash 1 and the reference sample, respectively. The difference between them is statistically significant at the 5% level ($p=0.028$). In the general Bulan-Koba population, including individuals from all cemeteries studied, the frequency of TBC is 0.6% (2/352). In the Pazyryk population this congenital trait has not been found (0/214).

The statistically significant difference in the frequency of TBC between Ayrydash 1 and the Bulan-Koba reference sample indicates that random clustering of these anomalies in one

cemetery is unlikely. As such, the two individuals with TBC buried in Ayrydash 1 apparently were genetic relatives.

Discussion

As TBC is a very rare and usually asymptomatic anomaly, its etiology and pathogenesis are not known.

The first evidence that there appears to be a genetic component in this condition was archaeologically derived from medieval Apulia, Italy, **where familial burial practices are known to be common**. It was found that two full-term fetuses affected by identical TBC were buried together in the same small tomb (Anderson, 2000).

The phenotypic spectrum of TBC varies from a little gap in one of the lateral sides to complete coronal separation of the basioccipital into two parts. On the inferior surface of the basioccipital, there are sometimes transverse or inclined bilateral ridges on either side of the pharyngeal tubercle, called the *crista sinostolica* by Mingazzini (1891), which appear to mark the fusion of the proatlas with the remainder of the basioccipital [ibid.; Figure (3C)]. Analysis of the literature data shows that the most common manifestation of TBC is an incomplete bilateral form, which appears twice as often as a unilateral form (39:17). The left side is affected more frequently in the unilateral forms (13:3), and possibly more severely in the incomplete bilateral forms. A complete TBC occurs in about 10% of all TBC cases.

Transverse basilar cleft is often associated with other anomalies of the skull including assimilation of the atlas into the basiocciput (Morselli, 1890; List, 1941; Lombardi, 1961), premature synostoses of the cranial sutures (Schultz, 1929; Limson, 1932; Kruffy, 1967; Woon et al., 1980), choanal atresia, and occurrence of a third condyle (Barth, 1896; Schultz, 1929; Johnson & Israel, 1979). None of the listed features, however, are present in the skulls from Ayrydash 1. Transverse basilar cleft can also be a part of severe syndromes such as Cornelia de Lange (Whitehead et al., 2015), CHARGE (Mahdi & Whitehead, 2017), Arnold-Chiari (List, 1941), as well as some others (Le Double, 1903; Johnson & Israel, 1979; Woon et al., 1980). In syndromic cases of TBC, infants have reduced chances of survival into adulthood. From a clinical practice perspective (Woon et al., 1980), above two full-term fetuses with TBC buried in a familial tomb in medieval Italy (Anderson, 2000) are likely to be twins affected by a syndromic disease. **Pathological traits, which would be indicative of a syndrome, have not been found in the skulls from Ayrydash 1.**

Although more than 70 cases of TBC have been reported in the anatomical, clinical and bioarchaeological literature, there are very few population-based studies (Table 1). In the majority of the recorded archaeological groups, the frequency of this anomaly is less than 1%. Only in the Jomon population, Japan, does it reach 6%. Transverse basilar cleft cases of different degrees of expression have been found in skeletal samples from six local sites of the Middle to Final Jomon period (Kawakubo et al., 2014). Results from studies of dental, craniometric and molecular data suggest that the origin of Jomonese people traces back to the indigenous inhabitants of Southeast Asia (Hanihara, 1993). In this regard, it should be noted that the so-called Deep Skull from Niah Cave in Sarawak, Malaysia, one of the earliest anatomically modern human fossils in Southeast Asia, displays TBC as well (Brothwell, 1960). Increase of frequency of this anomaly within the Jomon groups, therefore, may be explained by random genetic drift in a geographically isolated and/or sparsely populated region.

Judging by the number and size of burial sites, the Altai Mountains valleys were densely populated during the Scythian and Hunno-Sarmatian periods (Seregin & Matrenin, 2014). Groups of the Pazyryk and Bulan-Koba Cultures are similar craniometrically, which suggests biological continuity between them. This is especially evident for females (Chikisheva & Pozdnyakov 2000). However, there was aggregation of rare traits within some local burial sites of the Bulan-Koba Culture. Besides two cases of TBC in Ayrydash 1, three cases of premature cranial synostosis and three cases of internal frontal hyperostosis have been found in the Bulan-

Koba 4 and Ust'-Edigan cemeteries, respectively (S. Tur, unpublished data), which may be interpreted as evidence of parental consanguinity and inbreeding. Even today, consanguineous marriages are widely practiced in several large communities. The effect of consanguinity on the prevalence of minor and major congenital anomalies, as well as recessive and multifactorial diseases, has been documented in many clinical studies (e.g. Shawky et al., 2013). Although the two individuals with TBC buried in Ayrydash 1 were probably relatives, their graves were located in different segments of the cemetery. Since at least one of these individuals was determined as a female, this may indicate that Altai Mountains nomads during the Hunno-Sarmatian period practiced patrilocality, in which it was common to bury married women near their husband's family graves.

Conclusions

Transverse basilar cleft is a very rare congenital anomaly characterized by a coronal fissure completely or partially intersecting the basioccipital at the level of the pharyngeal tubercle. Population frequencies of TBC are usually no more than 1%, with few exceptions. Only two skulls display this anomaly in a large skeletal sample from burial sites of the Bulan-Koba Culture, Altai Mountains, attributed to the Hunno-Sarmatian period. Both findings originate from the Ayrydash 1 cemetery. The statistically significant difference in the frequency of TBC between Ayrydash 1 and the Bulan-Koba reference sample indicates that random clustering of this trait in the cemetery is rather unlikely and suggests instead a familial relationship between the individuals sharing this condition. Parental consanguinity and inbreeding could result in increased frequency of this rare anomaly in the local group of the nomads.

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List of figure legends

Figure 1. Location map and plan of the Ayrydash cemetery. Marked are graves #38 and #108 (modified from Soenov, 2003). This figure is available in color at wileyonlinelibrary.com/journal/oa.

Figure 2. Schematic drawings, illustrating different degrees of TBC: A – a mild bilateral incomplete symmetric form, B – a bilateral incomplete asymmetric form, C – a complete cleft (modified from Lucy, 1890).

Figure 3. Transverse basilar cleft in the skulls from the Ayrydash 1 cemetery: A – from grave #38; B – from grave #108; C – normal variant of the basioccipital, 1 – *crista synostotica* (after Mingazzini, 1891), 2 – *crista muscularis*. This figure is available in color at wileyonlinelibrary.com/journal/oa.

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Table 1. Frequencies of TBC in anatomical and archaeological samples

Location	Sample	Occurrence of TBC		References
		n	% (N)	
Italy	Anat	1 adult	0.5 (200)	Morselli, 1890
France, Lion	Anat	2 adults	1.3 (150)	Lucy, 1890
New Caledonia	Anat	1 adult	5.0 (20)	Lucy, 1890
Norway, medieval	Arch	1 adult	0.4 (243)	Barth, 1896
Egypt, Third dynasty	Arch	1 child	0.1 (1100*)	Schmidt, 1912
Hungary	Anat	2 adults	0.4 (500*)	Davida, 1913
Baltimore, USA	Anat	2 fetuses	1.8 (163**)	Limson, 1932
		1 infant		
South India	Anat	1 adult	0.5 (202)	Ray et al., 2014
Japan, recent	Anat	1 adult	0.3 (349*)	Kawakubo et al., 2014
Japan, Jomon period	Arch	3 adults,	6.0 (83*)	Kawakubo et al., 2014
		2 subadults		
Altai Mountains, Hunno-Sarmatian period	Arch	2 adults	0.6 (352*)	Present paper
Altai Mountains, Scythian period	Arch	no	0.0 (214*)	Present paper

n – number of individuals with TBC; N – number of observations; % = 100 x (n/N)

* samples consist of adults and subadults;

** samples consist of fetuses and infants only; other samples consist of adults only;

Anat – anatomical; arch – archaeological.