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Does the distribution of Wormian bone frequencies across different world regions reflect genetic affinity between populations?

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Abstract

Background Wormian, or sutural bones, are additional, irregularly shaped bone fragments that can occur within cranial sutures. These bones may arise due to various factors, including mechanical pressure on skull bones during early ontogenetic stages, such as during artificial cranial deformations, or due to genetic and environmental influences. This study investigates the potential genetic basis of sutural bones by comparing their frequencies across diverse global regions. It analyzed 33 craniological series, encompassing 2059 crania, to assess the frequency of sutural bones in the coronal, squamous, lambdoid, and occipitomastoid sutures among skeletal populations from regions including Aboriginal Australia, Melanesia, Southeast Asia, Siberia, Europe, and Native America. Biological distances between populations were calculated using Smith's mean measure of divergence (MMD), with results visualized through multidimensional scaling.

Results The analysis identified distinct clusters of Caucasian and Siberian populations. Siberian aboriginal populations are compactly grouped, consistent with mtDNA data indicating genetic roots dating back to the Neolithic inhabitants of the Lake Baikal region. Further, differentiation within these populations is linked to the founder effect and gene flow. Notably, genetically related groups like the Inuit and Chukchi of Chukotka differ from other Siberian groups. In contrast, southern Siberian populations, such as the Buryats and Mongols, are closely positioned, aligning with genetic data. The differentiation between Southeast Asian and African regions was subtler, with their clusters largely overlapping. Yet, genetic links between populations were observed in some cases. Thus, Australians, Melanesians, and Papua New Guineans were located close to each other on the multidimensional scaling map, as were two African populations.

Conclusions The findings tentatively suggest a potential genetic component in the expression of Wormian bones, although this hypothesis requires further empirical support, particularly through genetic studies. While genetic factors may influence the expression of Wormian bones, environmental conditions and pathological processes also play significant roles. It can be suggested that Wormian bones could potentially serve as an additional tool in kinship analysis within burials; however, their utility significantly depends on the extent of their genetic influence. If future genetic studies confirm a substantial genetic component and its dominance over environmental factors, the use of these bones in anthropological and forensic analyses would receive additional validation.

Keywords Sutural bones, Cranial nonmetric traits, Cranial morphology, Cranial suture variability, Skeletal populations

Background

Sutural or Wormian bones (*ossa Wormiana*), named after Danish physician Ole Worm (1588–1654), who first described these bones in the lambdoid suture as *ossicula lambdoideum*, are additional irregularly shaped

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bone fragments that may develop within cranial sutures. It is established that the primary ossification centers of all cranial vault bones form in the connective tissue around the 8th week of embryonic development. Cranial bones expand by apposition, where bone tissue is radially deposited from the central point of ossification (Standring 2016). Ossification then extends from the cranial bones into the intervening connective tissue layers. However, small secondary ossification centers may emerge at the bone margins, which typically fuse directly with the adjacent bone edge. If fusion does not occur in the vicinity of a future suture, small sutural bones persist within it (Parker 1905; Murlimanju et al. 2011, Patel et al., 2015; Sreekanth & Samala 2016).

According to various hypotheses, the appearance of Wormian bones may be caused by mechanical pressure on the skull bones at early stages of ontogeny, such as during artificial cranial deformations (Sanchez-Lara et al. 2007; O'Loughlin 2004; El-Najjar & Dawson 1977), as well as by genetic and environmental factors (Ghosh et al. 2017; Barberini et al. 2008). Moreover, Wormian bones are thought to occur more frequently in conditions with fewer cranial ossification centers, hypotonia, or reduced movement, which can result in deformational brachycephaly (Sanchez-Lara et al. 2007). Some authors suggest that the presence of sutural bones may indicate developmental instability associated with congenital diseases (Di Ieva et al. 2013; Vishali et al. 2012). The formation of Wormian bones may also be influenced by environmental changes in dural strain within open sutures and fontanelles (Sanchez-Lara et al. 2007). These bones are found in various conditions such as Down syndrome, kinky hair syndrome, Menke's syndrome, otopalatodigital syndrome, osteogenesis imperfecta, cretinism, cleidocranial dysostosis, primary acro-osteolysis, rickets, and hypothyroidism (Atoni et al. 2021). However, it has been proposed that the structural appearance of Wormian bones is influenced by genetic factors, while mechanical stress and various pathological conditions affect their number (Mao et al. 2003; Sanchez-Lara et al. 2007). Therefore, Wormian bones, like other nonmetric cranial traits, are considered epigenetic threshold traits, with phenotypic expression linked to genetic predisposition (Mao 2003; Goto et al. 2004; Sanchez-Lara et al. 2007; Barberini et al. 2008). Individual genes or groups of genes are believed to influence the formation of Wormian bones (Kague et al. 2016; Zimmerman et al. 2019). Comprehensive reviews of research studies on the presence of Wormian bones in populations worldwide were provided by Bellary et al. (2013) and Bisiecka and Romero-Reveron (2023).

Wormian bones can manifest in virtually all cranial sutures, yet their prevalence across different locations varies significantly. According to various authors (Marti et al., 2013; Ortadeveci, 2023; Singh 2024), lambdoid suture is the most frequent site for sutural bones. Wormian bones of the occipitomastoid, coronal, and squamosal sutures are less common, while those of the sagittal suture are very rare. Figure 1 illustrates the location of Wormian bones in the coronal, squamosal, lambdoid, sagittal, and occipitomastoid sutures. In the coronal suture, small, individual bones are typically observed (Fig. 1a). In the squamosal suture, Wormian bones appear to emerge from beneath the edge of the temporal scale (Fig. 1b). In the lambdoid suture, the sizes and positions of Wormian bones vary significantly, ranging from small, singular bones to clusters extending along the entire length of the suture (Fig. 1c, d). In very rare cases, a sutural bone (*os bregmale*) is found in the sagittal suture between the parietal bones (Fig. 1e). In this study, *os bregmale* was encountered only once, on the skull of an Inuit individual. Small, typically elongated sutural bones may be situated along the occipitomastoid suture (Fig. 1f).

Studying the nature of Wormian bones is crucial for estimating ancestry, identifying congenital conditions, distinguishing between trauma and normal anatomical variations, and conducting kinship analysis in anthropological and forensic contexts.

The objective of this study was to reveal the possible genetic background of sutural bones by comparing their frequencies across various global regions and examining the correspondence of frequency distribution patterns with genetic affinity between populations.

It is important to note that additional ossification centers also form in the cranial fontanels, leading to the emergence of fontanel bones (Standring 2016). However, this study focused exclusively on bones located within the cranial sutures.

Methods

A comparative analysis was conducted on 2059 adult crania from nineteenth- to twentieth-century burials of Aboriginal Australians, Melanesians, Southeast Asians, Africans, Siberians, Europeans, and Native Americans. The craniological series were examined by the author at the Museum of Anthropology of Moscow State University, the Museum of Anthropology and Ethnography in Saint Petersburg, and the University of Cambridge. The ethnic affiliation of the craniological series was established by the institutions housing the collections, based on the geographical locations of the skull finds, associated cultural artifacts, and historical records of the populations at those sites during the relevant periods. Due to the well-preserved condition of the crania, scoring of the traits was conducted using the individual count method, where bilateral traits were counted only once

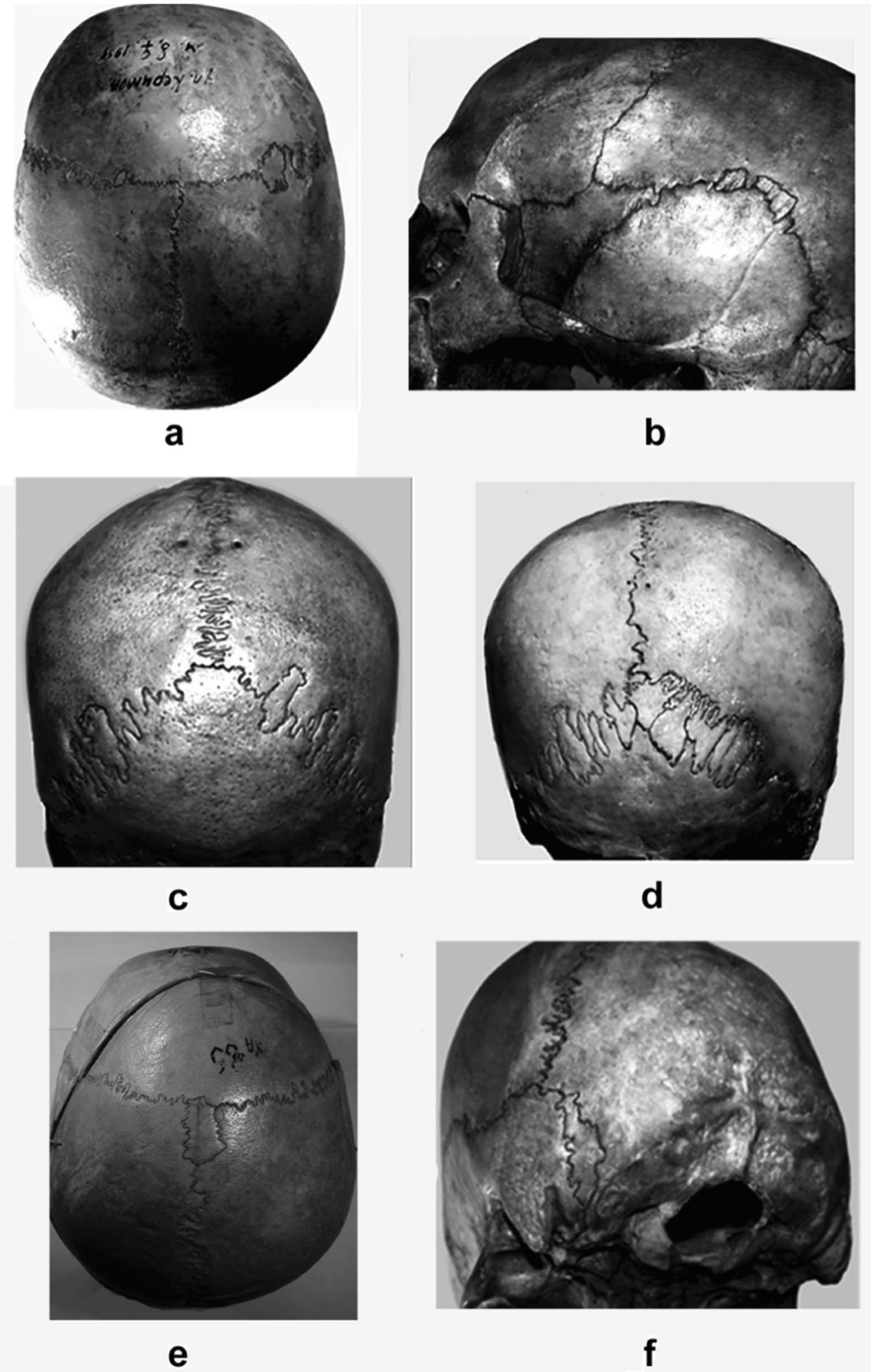

Fig. 1 Various types of Wormian bones encountered in this study. **a** Ossicle in coronal suture. **b** Ossicles in squamosal suture. **c**, **d** Ossicles in lambdoid suture. **e** Ossicle in sagittal suture (*os bregmale*). **f** Ossicle in occipitomastoid suture

per cranium, regardless of their bilateral appearance (Brasili, Zaccagni, Gualdi-Russo, 1999). Crania exhibiting pathological features, including trauma, disease-induced deformities, artificial deformation, sutural agenesis, premature cranial synostosis, or other abnormalities that could confound study results, were excluded from the analysis. The analysis also excluded the few children's and elderly skulls, leaving two age groups: 13–39, and 40–59 +. The age and sex composition of the craniological series is presented in Table S1 in supplementary materials.

Despite data indicating a lack of sexual dimorphism in Wormian bones (Goyal, 2019; Natsis, 2019), the chi-square test and Fisher's exact test were used to examine correlations of individual traits with sex and age. Correlations were calculated for the most representative series, including Inuit, Chukchi, Mongols, Khanty, Armenians, and Australians (Tables S2, S3). Since significant correlations were absent, the data from different sex and age groups were consolidated for subsequent analysis.

Biological divergence between sample pairs was assessed using the modified Smith's mean measure of divergence (MMD), which is particularly effective for quantifying differences in nonmetric cranial traits across populations, allowing to assess the degree of biological divergence by calculating the average pairwise differences among groups, providing a robust measure that is less sensitive to sample size variations than other metrics. This method was repeatably and successfully used for inter-sample comparison of nonmetric trait frequencies (e.g., Irish 2010; Hanihara et al. 2003; Hallgrimsson et al., 2004; Sutter and Mertz 2004; Ossenberg et al. 2006; Nikita et al. 2012; Movsesian et al. 2014, 2017; 2020; Weiss 2018). The full description and discussion of the mean measure of divergence (MMD) can be found in the studies by Sjøvold (1977) and Irish (2010). Irish validated the effectiveness of MMD by comparing it to the Mahalanobis D^2 statistic, establishing that both are equally effective for analyzing nonmetric traits. In this study, MMD calculations employed the Freeman and Tukey angular transformation for small samples or extreme trait frequencies (less than 0.05 or greater than 0.95) as outlined by Green and Suchey (1976) and Sjøvold (1977). These are considered significant at the 0.025 level if they exceed twice the standard deviation. Multidimensional scaling (MDS) was applied to the MMD matrix due to its capacity to visually represent the distances or dissimilarities among the studied groups in a low-dimensional space. MDS was chosen over other statistical methods like principal component analysis (PCA) because it directly utilizes the dissimilarity matrix generated from MMD, facilitating a more intuitive understanding of group relationships in terms of overall divergence. As noted by Irish, MMD distances are well-suited for MDS procedures, despite MDS typically being based on Euclidean distances. For instance, when MDS was conducted using three dimensions, the obtained stress value was 0.058, and Spearman coefficient was 0.972, indicating a good representation of the MMD distances (Irish 2010). Similar findings were also reported by Nikita et al. (2012). The calculations were performed using an R script (package "AnthropMMD").

Results

The frequencies of Wormian bones in the studied populations are presented in Table 1.

There are noticeable differences in the distribution of trait frequencies both within and between regions. For example, Siberian populations almost completely lack sutural bones in the coronal suture; there is little variation in the frequency of sutural bones in the squamosal suture between regions, though some variability exists within regions; the frequency of Wormian bones in the lambdoid suture is the lowest in the Siberian region. Sutural bones in the occipitomastoid suture are most frequently found among Native Americans and populations of Southeast Asia. The matrix of distances between populations (MMD), standard deviations, and significance levels are displayed in Tables S4 and S5.

Figure 2 presents the results of multidimensional scaling of MMD distances.

It should be noted that the results of the multidimensional scaling (MDS) indicate stress values close to zero and a high Spearman coefficient. In the MDS graph, the stress value measures the accuracy with which the distances in the reduced-dimensional space reflect those in the original, higher-dimensional space. A lower stress value indicates a more accurate representation, demonstrating that the two-dimensional representation effectively preserves the true distances between data points. Additionally, the Spearman coefficient, which evaluates the rank correlation between two datasets, confirms the extent to which the relationships among variables are maintained when dimensionality is reduced. A Spearman coefficient near + 1 suggests a strong positive correlation, affirming that the rank order of the original distances is well preserved in the scaled representation, which indicates a strong correlation between the graph distances and the actual differences between populations in the frequencies of Wormian bones.

Discussion

The etiology of sutural bones remains somewhat unclear. However, it has been repeatedly noted that Wormian bones are often found in healthy individuals, and their presence is usually not associated with pathological conditions (Natsis et al. 2019; Andrade, Kalthur, 2018; Johal et al. 2017). Only

Table 1 Frequencies of Wormian bones in the studied populations

Groups	*N*	Coronal suture	Squamosal suture	Lambdoid suture	Occipitomastoid suture
Siberia					
Inuit (Chukotka)	102	0	0	0.063	0.049
Chukchi (Chukotka)	154	0	0	0.084	0.1
Aleuts	56	0	0.02	0.192	0.073
Negidals	33	0	0	0.096	0
Ulchi	50	0	0	0.120	0.07
Tungus	29	0	0	0.103	0.074
Telengits	90	0	0.011	0.144	0.111
Mongols	100	0.011	0.02	0.126	0.02
Buryats	40	0	0.025	0.107	0.041
Tuvans	59	0	0.01	0.232	0.034
Bashkirs	70	0	0	0.173	0.044
Khanty	129	0	0.016	0.218	0.039
Mean		**0.001**	**0.009**	**0.138**	**0.055**
South and North America					
Peruvians	94	0.042	0.011	0.362	0.168
Pueblo Indians	35	0	0	0.235	0.176
Mean		**0.021**	**0.006**	0.299	**0.172**
Africa					
Tanzanians	56	0	0.018	0.214	0.107
Kenyans	60	0.008	0	0.317	0.067
S-E Africans	30	0.036	0	0.345	0.069
Mean		**0.015**	**0.006**	**0.292**	**0.081**
S-E Asia					
Burmese	56	0.005	0.018	0.143	0.161
Indians (Andhra Pradesh)	56	0.071	0.036	0.428	0.107
Australian aborigines	78	0.026	0.026	0.410	0.102
Melanesians	65	0.011	0.016	0.406	0.219
Papuans	68	0.011	0.011	0.336	0.118
Malays	21	0	0	0.190	0.19
Mean		**0.021**	**0.018**	**0.319**	**0.150**
Europe					
Armenians	129	0.025	0.015	0.341	0.062
Shapsugs	42	0.048	0.024	0.438	0.01
Abkhaz	55	0.022	0	0.280	0.02
Italians	43	0	0.023	0.169	0.01
Turks	31	0	0	0.097	0.067
French	31	0.008	0	0.161	0.008
Latvians	81	0.025	0.012	0.456	0.063
Estonians	39	0.025	0	0.417	0.028
Poles	24	0	0	0.333	0.01
English	53	0.024	0	0.238	0.01
Mean		**0.018**	**0.007**	**0.299**	**0.029**

a significant number and relatively large size of these bones might be considered indicators of certain congenital disorders, primarily osteogenesis imperfecta, characterized by an abnormally large number of sutural bones (Semler et al. 2010). Wormian bones are common and may be numerous without necessarily indicating osteogenesis imperfecta (Marti et al., 2013). Similarly, studies on Southwestern Native American skulls found no significant differences in

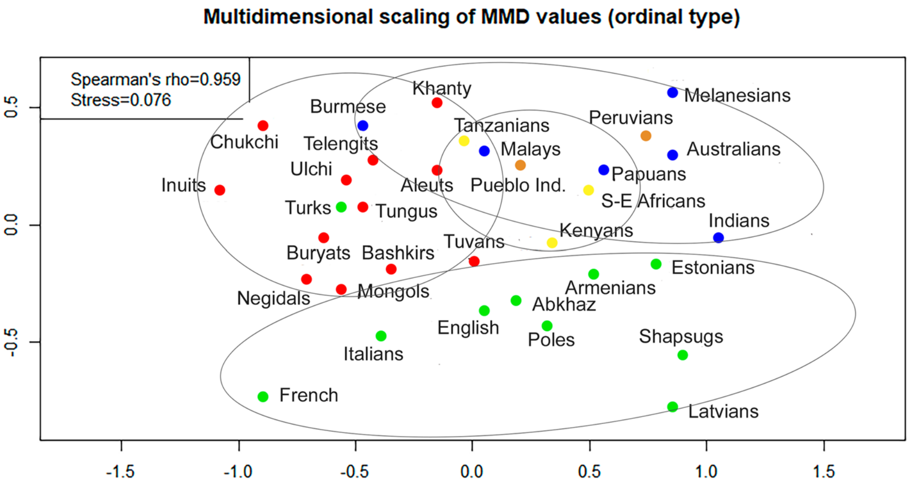

Fig. 2 Location of the studied populations on the multidimensional scaling graph. Red dots — Siberian populations, blue dots — South-East Asians, yellow dots — Africans, brown dots — American Indians, green dots — Europeans

the frequency of these bones between artificially deformed and non-deformed skulls; deformation only affected their total number on the skull (El-Najjar, Dawson, 1977).

According to Güler et al. (2024), the prevalence of Wormian bone varies in different geographical regions. Rathmann et al. (2023) systematically analyzed the utility of four cranio-dental phenotypic data types in capturing neutral genomic variation: cranial metrics, dental metrics, cranial nonmetric traits, and dental nonmetric traits, as well as a combined dataset. The meta-analysis revealed that these data types differentially capture neutral genomic variation, with the highest signals in dental nonmetric and cranial metric data, followed by cranial nonmetric and dental metric data.

Some studies have suggested that sutural bones may serve as biological markers of interpopulation differences to some extent (Pal, Routal, 1986; Gumusburun et al. 1997; Natsis et al. 2019). The results of our study align with these hypotheses. The multidimensional scaling graph (Fig. 2) reveals certain patterns that, in some cases, reflect the genetic proximity of populations. For example, although populations traditionally classified as European occupy a large part of the field, they still form a separate cluster, except for the Turks. On the left side of the European cluster are the Western European populations — English, French, and Italians — who, despite contemporary genetic diversity, remain genetically connected due to their complex history of migrations, invasions, and cultural exchanges over thousands of years. In contrast, the genetically related South Caucasus populations, specifically Armenians and Abkhazians (Teuchezh et al. 2013), are grouped together at a distance from the Western European section of the cluster.

Aboriginal populations of Siberia are more compactly situated. This is consistent with mtDNA data, which indicates that the populations of Siberia have common genetic roots tracing back to the Neolithic inhabitants of the Lake Baikal region, and their further differentiation is attributed to the founder effect and gene flow (Gill et al. 2023). Additionally, among the Siberian populations, distinct-related groups can be identified. For instance, genetically related Inuit and Chukchi of Chukotka (Agdjoyan et al. 2021) are distinct from other Siberian groups, while the populations of southern Siberia such as the Buryats and Mongols are located close to each other, which is also consistent with genetic data (Karafet et al 2018). Similarly, genetically related groups from eastern Siberia, such as the Ulchi and Tungus (Agdjoyan et al. 2019), are closely positioned.

The differences between the regions of Southeast Asia and Africa are not as sharply defined, and the clusters formed by these populations significantly overlap. Nonetheless, the arrangement of populations from these regions is not as random as it might seem at first glance: Australians are found close to Melanesians and Papuans, as indicated by mtDNA data (van Holst Pellekaan, Frommer, Sved, Boetcher, 1998), while Southeastern Africans are close to Kenyans. Thus, the frequency of Wormian bones varies significantly across different ethnic groups, suggesting that genetic predispositions may play some role in their development. However, nongenetic factors such as environmental influences, including nutritional deficiencies and exposure to toxins, as well as lifestyle choices and conditions that affect bone development like osteogenesis imperfecta and rickets, also significantly contribute to the formation of Wormian bones.

Additionally, mechanical stresses during childbirth or from traditional cranial deformation practices can further influence their occurrence.

Conclusions

Despite the approximate nature of the biological connections inferred from the frequency of sutural bones, as identified in this study, the spatial arrangement of the examined populations is non-random and exhibits discernible patterns. The findings tentatively suggest a potential genetic component in the expression of Wormian bones, although this hypothesis requires further empirical support, particularly through genetic studies. It can be suggested that Wormian bones could potentially serve as an additional tool in kinship analysis within burials; however, their utility significantly depends on the extent of their genetic influence. If future genetic studies confirm a substantial genetic component and its dominance over environmental factors, the use of these bones in anthropological and forensic analyses would receive additional validation.

Limitations

The scope of this study is limited by the availability of craniological series, which affects the diversity of the analysis. Key regions such as Africa, North America, and South America are notably underrepresented. Furthermore, merging data from Australia and Melanesia with Southeast Asia might obscure significant regional variations pertinent to Wormian bones. This limited geographic coverage restricts the generalizability of our findings. To address these limitations, future research should aim to include a wider array of population samples from underrepresented regions to enhance the diversity and relevance of the findings. Expanding the geographic scope will facilitate more comprehensive genetic analyses, providing clearer insights into the global variability of Wormian bone frequencies. Additionally, to explore genetic influences on Wormian bones more thoroughly, conducting genetic studies on well-documented family burials spanning multiple generations is recommended. This method permits direct observation of inheritance patterns and assessment of genetic predispositions, offering deeper insights into the hereditary aspects of these traits. Such studies could enhance the robustness and applicability of the results in anthropological and forensic contexts.

Abbreviations

MMD	Mean measure of divergence
MDS	Multidimensional scaling

Supplementary Information

The online version contains supplementary material available at https://doi.org/10.1186/s41935-024-00405-1.

Supplementary Material 1: Supplementary tables: Table S1. Distribution of craniological series by sex and age. Table S2. Results of Fisher's Exact Test and Chi-Square Test for Association Between Sex and Presence of Trait. Table S3. Results of Fisher's Exact Test and Chi-Square Test for Association Between Age and Presence of Trait. Table S4. MMD values (upper triangular part) and associated SD values (lower triangular part). Table S5. MMD values (upper triangular part) and associated significant indicators *(lower triangular part).

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Author's contributions

AM collected and analyzed the data, reviewed the relevant literature, performed the photography, and wrote the main text. The author has read and approved the manuscript.

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Availability of data and materials

The datasets used and analyzed during the current study are available from the corresponding author on reasonable request.

Declarations

Ethics approval and consent to participate

The author confirms that all procedures involving human remains complied with the guidelines set by the Bioethics Commission at Moscow State University. Necessary permissions were obtained from relevant authorities and institutions prior to the commencement of the study. Moreover, the author ensured that all human remains were treated with the utmost respect and dignity, with careful consideration of cultural and historical contexts, consistent with best practices in anthropological research.

Consent for publication

Not applicable.

Competing interests

The author declares that he has no competing interests.

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