A global study shows that population history is a better predictor of the shape of the human os coxae than climate

LIA BETTI[1,2], NOREEN VON CRAMON-TAUBADEL[2], ANDREA MANICA[3] AND STEPHEN J. LYCETT[2]
[1]Department of Archaeology and Anthropology, University of Cambridge, UK.
[2]School of Anthropology and Conservation, University of Kent, UK.
[3]Department of Zoology, University of Cambridge, UK.

INTRODUCTION

Human populations living in high-latitude regions tend to have wider pelves than populations from tropical regions. This pattern has been explained as thermoregulatory adaptation, whereby a wider pelvic girdle would reduce the surface-to-volume ratio of the body and help body heat retention in cold environments, while a narrow pelvis would facilitate heat dissipation in hot tropical regions (e.g., Ruff, 1991, 1994; Holliday and Hilton, 2010). There is, however, **a serious theoretical problem in testing the effect of hypothetical selective factors without taking into account the neutral pattern of variation due to population history**. Neutral processes such as genetic drift and gene flow have been shown to explain a high proportion of cranial variation in modern populations. (e.g., Roseman 2004, Relethford 2004, Betti et al 2009, 2010). Similarly, modern human pelvic shape diversity was shown to reflect neutral population history, carrying the signature of the serial founder events that accompanied the expansion of our species out of Africa (Betti et al., 2012; 2013). When examining pelvic differences between modern populations, therefore, we might expect that **shared population history and gene flow would have generated a pattern of genetic and morphological similarity between populations that needs to be explicitly taken into account** before testing the effect of any proposed selective factor. In other words, the underlying neutral pattern of variation due to population history should represent the **null hypothesis** against which to test the effect of additional, non-neutral processes (Roseman and Weaver, 2007).

We compiled a large global dataset of 3D landmark configurations of the human os coxae, in order to gain a representative view of hip bone variation in our species.

The aims of this study can be summarized as follows:

1) **To identify the effect of neutral population history** in shaping human pelvic variation.

2) **To evaluate the effect of climatic adaptation** whilst accounting for the underlying neutral pattern of population history.

MATERIALS AND METHODS

The shape and size of the os coxae was recorded with **27 3D landmarks** (Fig. 1) in 1,494 individuals, 922 males and 572 females, representing **30 male and 23 female populations (**Fig. 2). Male and female configurations were subjected to separate generalized Procrustes superimposition, and new morphometric variables were obtained by performing a Principal Component Analysis (PCA) on the covariance matrix of the Procrustes coordinates (as projected on the tangent space). Regional climate was recorded as average minimum temperature of the coldest month, average maximum temperature of the hottest month and average annual precipitation.

Estimating the signature of neutral population history

As **genetic similarity** between geographically proximate human populations is in part due to reciprocal exchange of migrants, it **declines exponentially with increasing geographic distance** in what is often defined as **an "Isolation-By-Distance" (IBD) process**. Between-population morphometric distance (Q) was calculated following Relethford and Blangero (1990), assuming total heritability, and normalized using an arcsine square root transformation. Pair-wise population geographic distances (D) were calculated as the shortest route on landmasses, avoiding areas with a mean altitude higher than 2,000 m over sea level (Manica et al., 2005). Following Betti et al. (2010), **the fit to the IBD model was tested using an explicit non-linear model derived from Malécot's (1969) coefficient of kinship** :

$$q_{ij}^2 = A'(1 - e^{-bd_{ij}})$$

where q_{ij} is the pair-wise phenotypic distance between populations i and j, A' is the value at which phenotypic distance reaches the asymptote and stops increasing when geographic distance (d_{ij}) increases, and b is the rate of increase in phenotypic distance. The parameters of the non-linear model were estimated by nonlinear least square optimization, using the function *nls* in R.

Testing for the effect of climate on the size of the os coxae

Average centroid size for each population was regressed on the three climatic variables, using a generalized least square model. As geographically closer populations are expected to be more similar to each other due to shared population history and gene flow, **spatial autocorrelation was taken into account** in the regression model.

Testing for the effect of climate on shape differences

The correlation between the matrices of between-populations **climatic distance** (Tmin.d, Tmax.d and Prec.d) **and morphometric distance (Q) was tested by Mantel test.** In order to control for population history, **we repeated the analysis by first fitting the non-linear relationship between Q and D, and then testing for a linear association with the climatic matrices** (Betti et al., 2010).

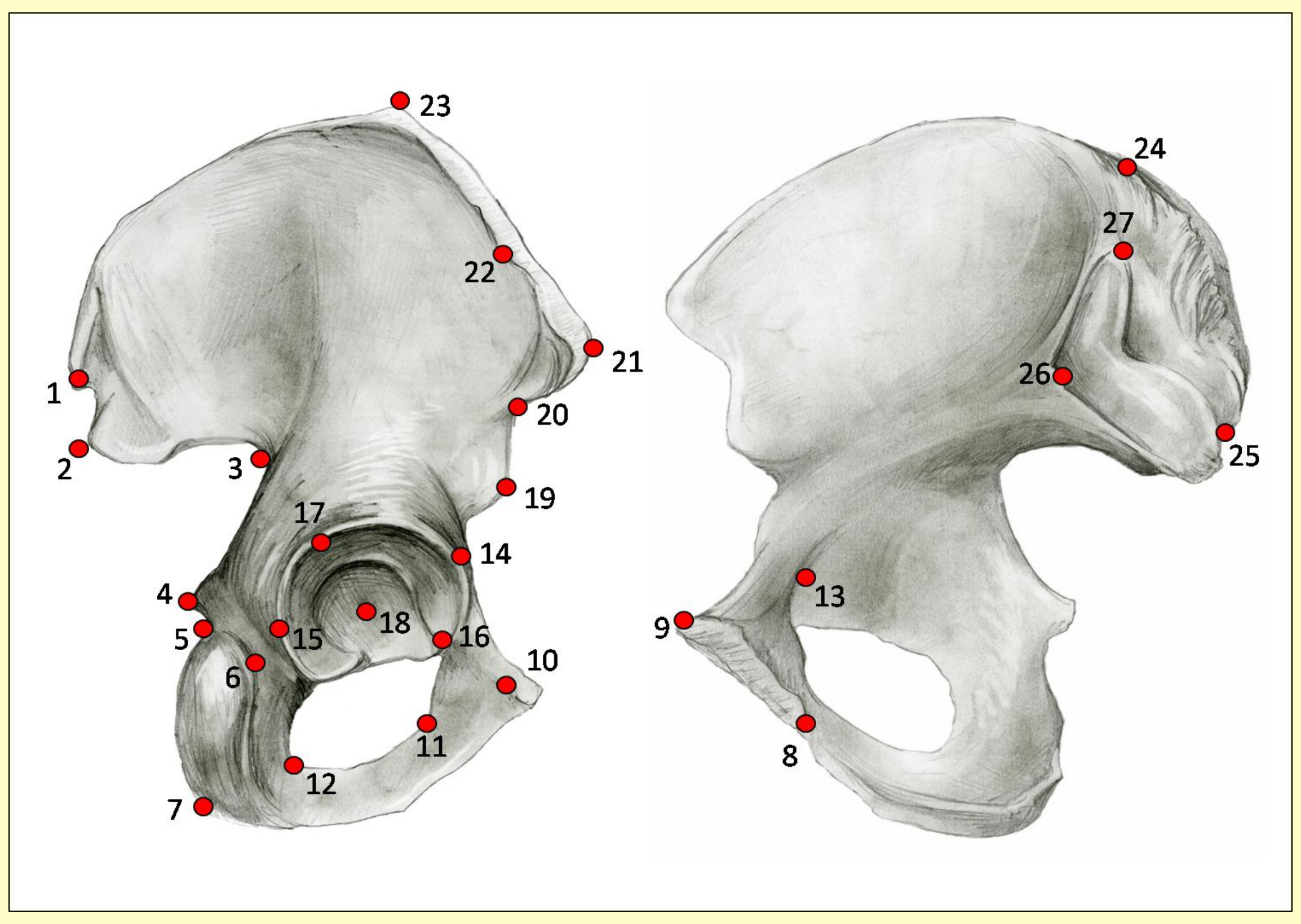

Figure 1. Landmarks of the os coxae used in this study.

Figure 2. Distribution of the population samples (circles = both sexes, triangles = males only, inverted triangles = females only).

RESULTS

The results show a **significant fit of coxal shape variation to the demographic history signature** (IBD), for both sexes (r^2 = 0.344, p<0.001 in males; r^2 = 0.300, p<0.001 in females) (Fig. 3). When accounting for spatial autocorrelation, **only minimum temperature and precipitation show a significant correlation with coxal size in males, and no significant correlation is found in females** (Table 1). In males, larger os coxae are associated with cold temperature and dry conditions, while smaller size is associated with hot and humid climates. **Differences in shape among populations correlate with differences in temperature** in males and females. **The correlation remains significant once corrected for spatial autocorrelation** in the form of a non-linear model of the demographic history signature, **but it is substantially reduced** (Table 2).

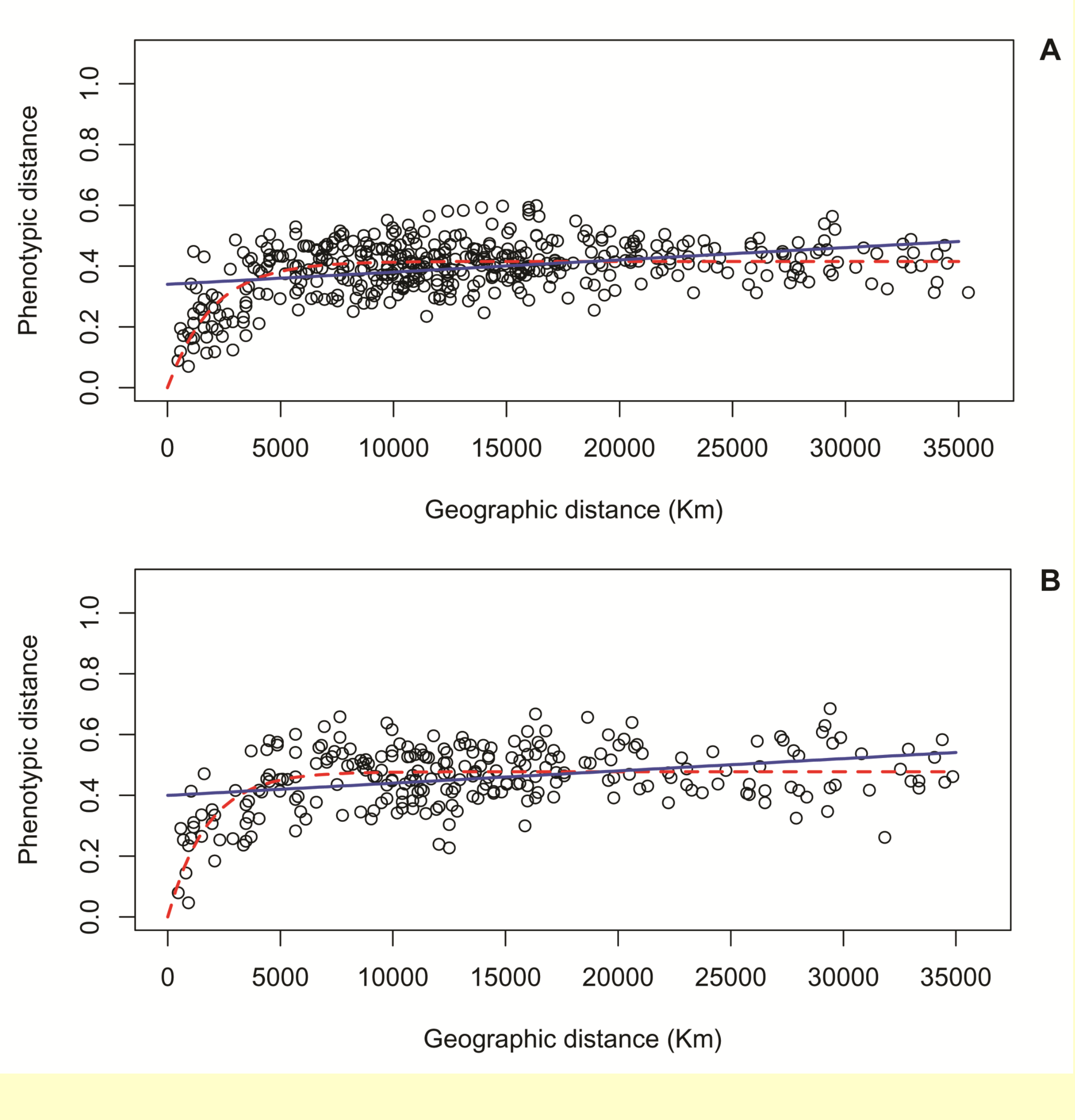

Figure 3. Plots of between-population phenotypic distance and geographic distance for males (A) and females (B). Each circle represents a pair of populations. The fit with a linear and a non-linear model of the neutral signature of past population history are represented by a full blue line and a dashed red line, respectively.

Table 1. Results of the correlation between average population coxal size and each climatic variable. *The new p-values take into account spatial autocorrelation.

Model	MALES			FEMALES		
	R^2	P-value	New p-value*	R^2	P-value	New p-value*
size VS Tmin	0.426	< 0.001	**0.006**	0.323	0.005	0.111
size VS Tmax	0.219	0.009	0.614	0.129	0.093	0.971
size VS Prec	0.252	0.005	**< 0.001**	0.102	0.138	0.250

Table 2. Results of the Mantel tests between population coxal shape differences (Q) and climatic distances, and after correcting for the IBD signature.

Model (males)	R^2	P-value	Model (females)	R^2	P-value
Q VS IBD	0.344	**<0.001**	Q VS IBD	0.300	**<0.001**
Q VS Tmin.d	0.183	**< 0.001**	Q VS Tmin.d	0.048	**0.033**
Q VS Tmax.d	0.089	**<0.001**	Q VS Tmax.d	0.054	**0.006**
Q VS Prec.d	0.022	0.084	Q VS Prec.d	0.002	0.337
Q VS Tmin.d / IBD	0.096	**< 0.001**	Q VS Tmin.d / IBD	0.014	0.215
Q VS Tmax.d / IBD	0.037	**0.006**	Q VS Tmax.d / IBD	0.026	**0.033**
Q VS Prec.d / IBD	0.008	0.328	Q VS Prec.d / IBD	0.003	0.656

DISCUSSION

The results of this study confirm the importance of neutral processes in shaping os coxae variation. **At least 30% of coxal shape differences between globally distributed populations can be accounted for by neutral processes** related to past dispersal events and recurrent gene flow (i.e. population history).

The presence of a signal of neutral population history does not necessarily rule out a role of diversifying selection or plasticity in shaping global variation in the size and shape of the os coxae. Indeed, the analyses confirm that **populations living in hotter and more humid environments tend to have smaller pelvic bones** than populations from cold and dry regions. The analyses also reveal **a significant (albeit weak) effect of temperature and precipitation on the shape of the os coxae** even after correcting for neutral processes, partially explaining variation between populations living in different environments.

On the other hand, **the effects of climate appear substantially reduced once neutral processes are accounted for**, revealing a tendency to overestimate the signal when population history is not explicitly included in the model. von Cramon-Taubadel (2009) and Betti et al. (2010) found a similar decrease in the correlation of climate with cranial variation once the neutral pattern was taken into account. The overestimation of the effect of selection appears to be a recurrent risk in studies of human phenotypic variation, confirming the importance of explicitly modeling the underlying neutral variation structure together with the hypothetical selective factors.

Neutral and selective processes are compatible and not mutually exclusive, and both contribute to modern pelvic variation. However, **neutral processes appear to have left a much stronger signature on global patterns of population diversity than climatic adaptation**.

The results have also **important implications for the interpretation of pelvic differences between hominin species.** Differences in cranial shape between modern humans and Neanderthals, in some cases interpreted in the light of climatic adaptation (Franciscus, 2003; Holton and Franciscus, 2008) have been recently shown to be compatible with neutral phenotypic evolution in the two lineages (Roseman and Weaver, 2007; Weaver et al., 2007, 2008). In a similar way, differences in pelvic shape between Neanderthals and modern humans are often attributed to the adaptation of the former to colder climates (Ruff, 1994; Holliday, 1997; Weaver and Hublin, 2009). In the light of these new results, suggesting a substantial effect of neutral evolutionary processes on the human pelvis, and a limited effect of climatic selection, it may be necessary to re-evaluate previous adaptive interpretations of hominin pelvic shape differences.

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