MODERN HUMAN PHENOTYPIC VARIATION

EXPLORING PATTERNS OF DIFFERENTIATION WITHIN AND BETWEEN CONTINENTS

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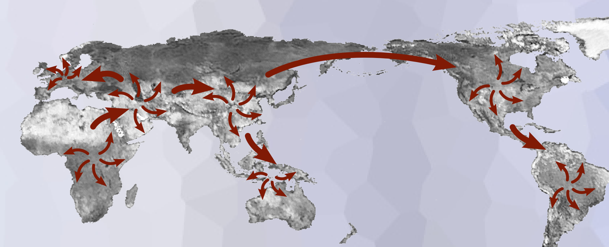
1. INTRODUCTION

Current consensus holds that modern human cranial phenotypic variation is the result of isolation-by-distance and neutral evolutionary processes (e.g., Relethford 2004, Manica et al. 2007, Betti et al. 2009), with natural selection acting most notably on some anatomical regions of populations living in extreme cold environments (e.g., Roseman 2004, Harvati and Weaver 2006, Hubbe et al. 2009). Under this model, there is an underlying assumption that the rate of morphological differentiation in the past was uniform across the planet. However, in a previous study (Hubbe et al. 2011) we suggested this might not be the case: Late Pleistocene specimens from Europe, East Asia and South America shared a common morphological pattern, which was quite distinct from the morphologies of recent groups from their geographical regions. Although based on a limited number of specimens, these results favor the scenario that the actual morphological diversity seen among *Homo sapiens* worldwide is the result of a late event of differentiation, probably taking place during the Holocene.

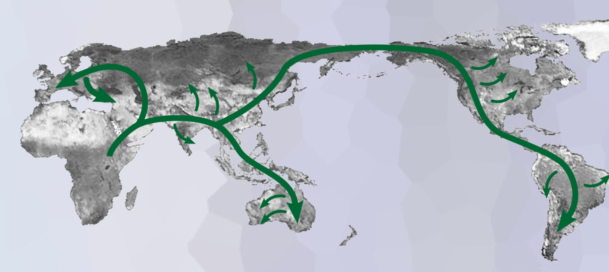
Our goal in this study is to test whether worldwide human cranial morphological variation fits a model of progressive geographic diversification associated with slow and constant phenotypic change, or a model of rapid initial dispersion followed by a later process of morphological diversification.

2. DISPERSION SCENARIOS AND EXPECTATIONS



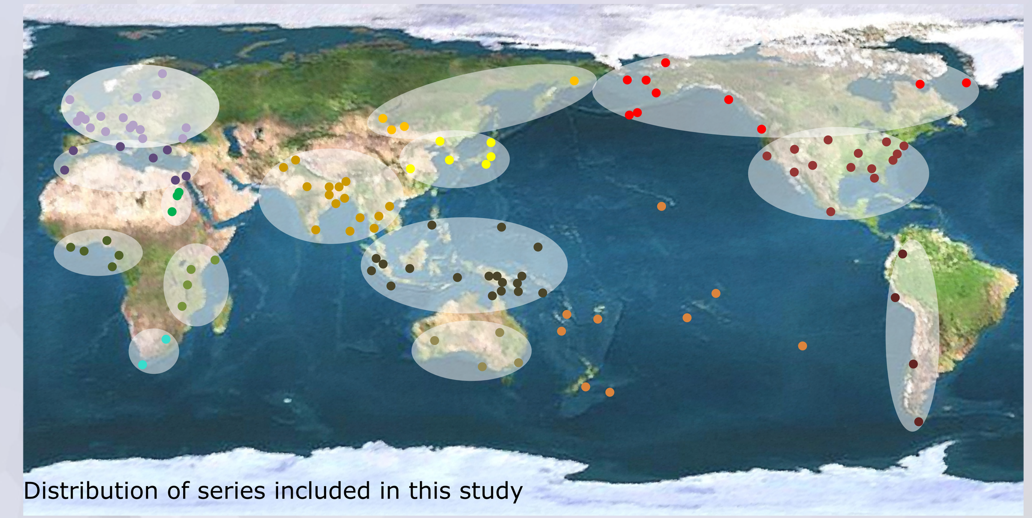
Scenario 1 – Constant rate of morphological differentiation

Expectations: Under this model, we would expect morphological differentiation to occur as a process of recurrent small bottlenecks and isolation by distance. This model predicts that morphological differences between groups will be largely the result of their temporal or geographical separation from their last common ancestor, and consequently, a strong geographic structure is expected in the pattern of differentiation of groups between regions.



Scenario 2 – Rapid expansion followed by late events of differentiation

Expectations: For this scenario we also expect similar end results to those of Scenario 1, i.e., geographic organization of morphological variation between regions and progressive loss of variation with increased distance from the ancestral populations. However, in this case, morphological differentiation is not a process of progressive bottlenecks and isolation by distance alone, but it is the outcome of local events after the initial occupation of the region. For our immediate goals, regional morphologies would derive from a relatively similar cranial morphology present in every major geographic region across the planet.



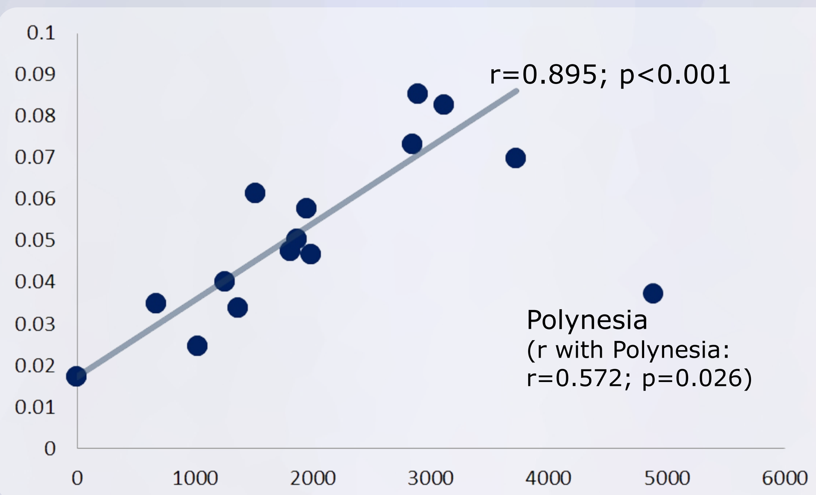
Distribution of series included in this study

3. MATERIALS

We analyzed a large sample of male individuals from 135 human series from Hanihara's dataset. This database has been previously used to study the effects of intra-group variation loss relative to distance from Africa (Manica et al. 2007, Betti et al. 2009) and the effects of climate on anatomical regions of the skull (Hubbe et al. 2009). Craniometric variation was assessed with 33 linear measurements for each pair of the series. Size effect was removed following Darroch and Mosimann (1985)

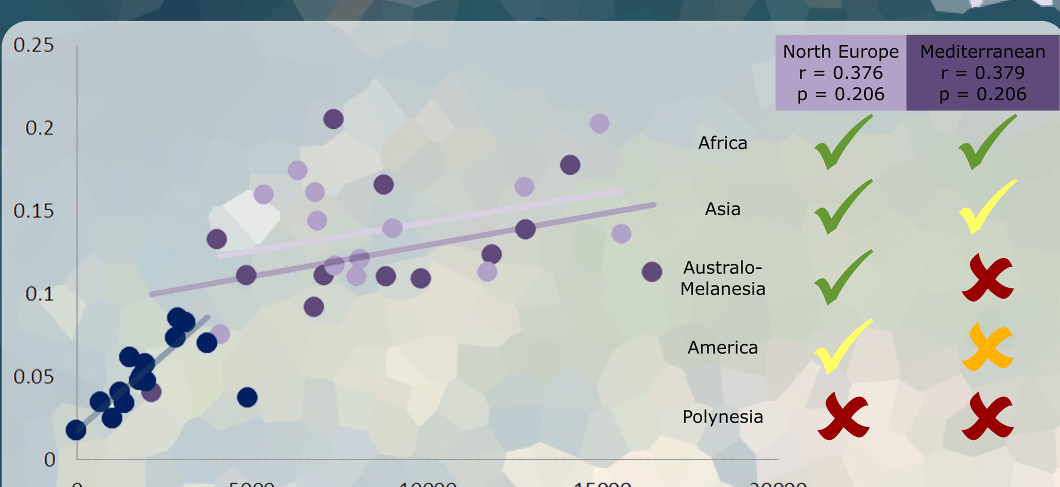
4. METHODS

Fst was calculated between pair of series.
Series were grouped into 15 geographic regions (as detailed in Hubbe et al. 2009).
First, correlations between Fst and geographic distances were calculated for the average values within each geographic region.
Second, similar correlations were calculated between regions.
Third, an expected ratio of morphological differentiation per kilometer (Fst/km) for the within-region Fst values was calculated, and a 95% confidence interval was estimated and used to compare with the between-region fst/km ratio.
Finally, ratios between regions were contrasted to this confidence interval.

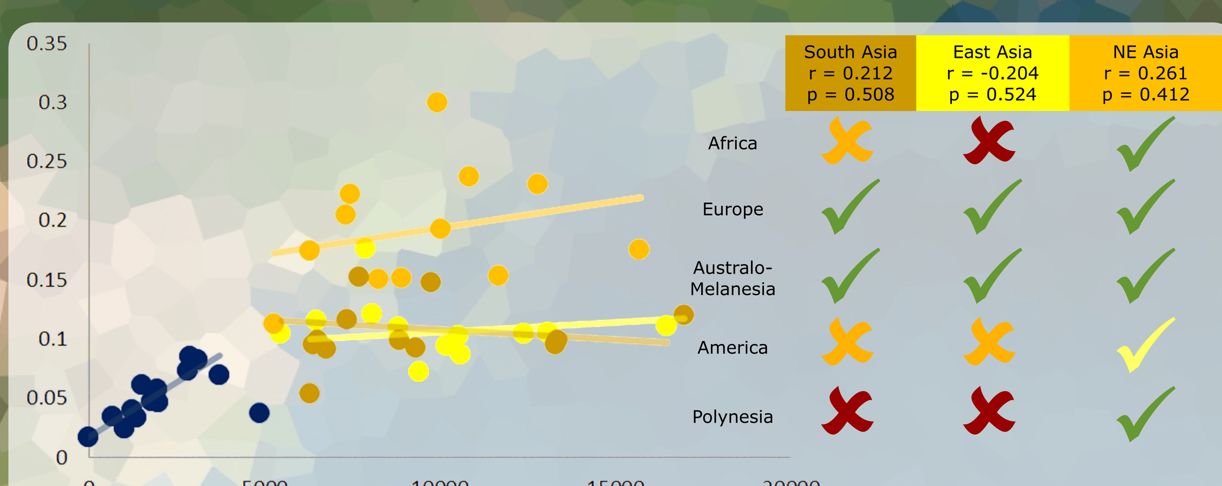


5. WITHIN REGIONS MORPHOLOGICAL DIFFERENTIATION

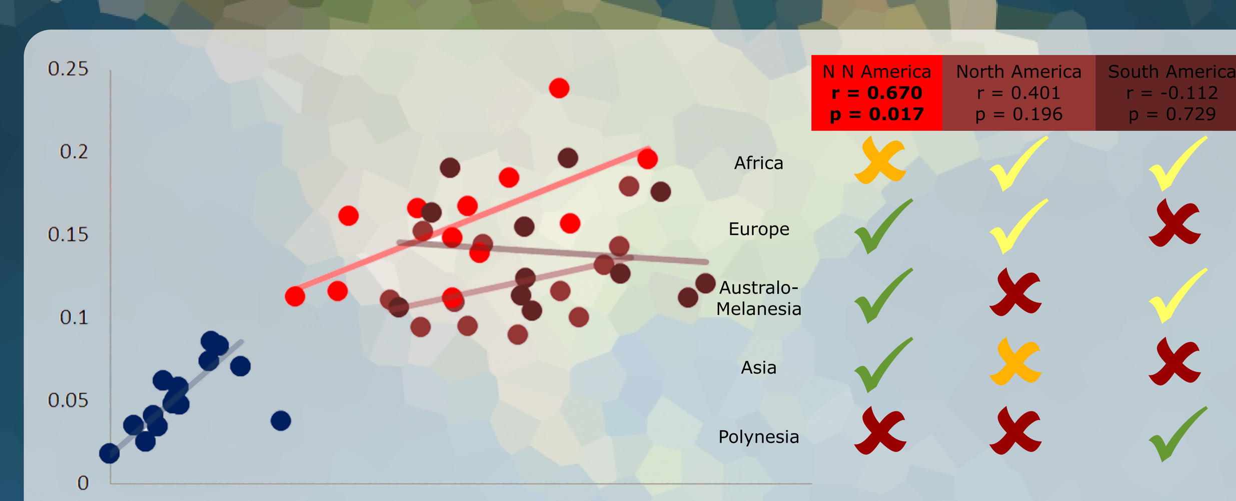
Within-region results indicate a very strong linear correlation between average Fsts and average distance (r=0.895; p<0.001), as represented in the graph to the left. The only region that is an outlier in this relationship is Polynesia, with lower Fst values than expected given the average distance between populations. This outlier position of Polynesia groups is expected due to the maritime transportation that connected the islands, effectively reducing the geographic distance between these insular groups. This strong linear association suggests that within each of the 15 geographic regions included in this study, geographic distance explains most of the morphological differences between groups.



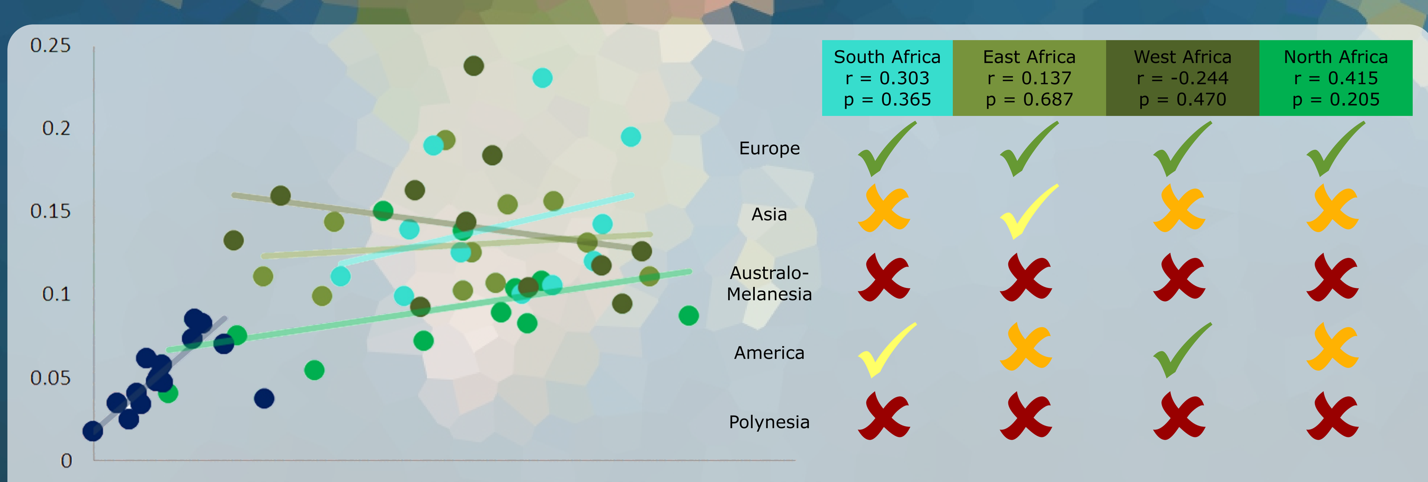
Europe: Geographic distance is not correlated with Fst between European series and the other regions of the planet.
Fst between North Europe and all regions but Polynesia fall inside the within-regions ratio confidence interval. For Mediterranean series, only distances to Africa and some Asian series fall inside the within-regions ratio interval.



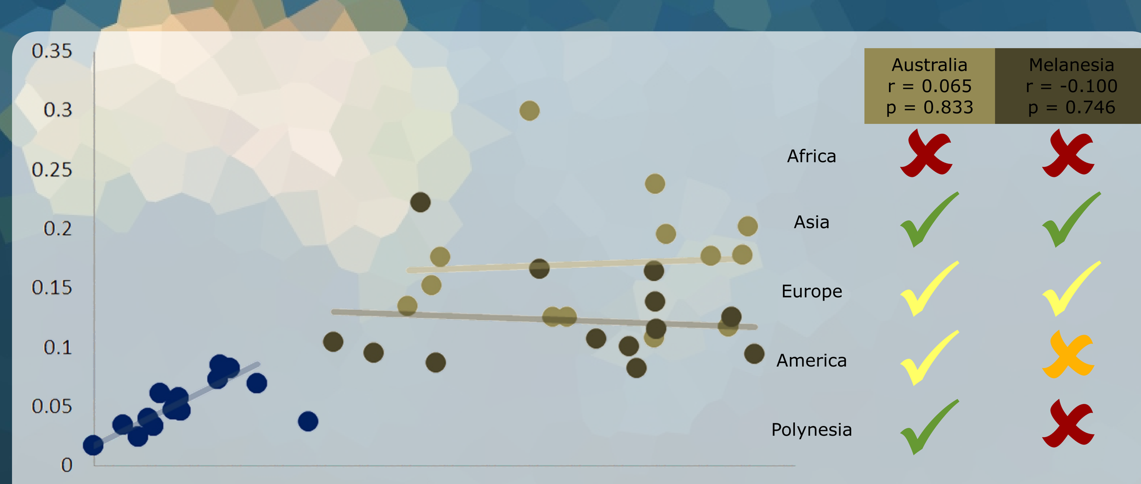
Asia: Geographic distance is not correlated with Fst between Asian series and the other regions of the planet.
Fst between NE Asia and other regions fall inside the within-regions ratio confidence interval. For South Asian and East Asian series only distances to Europe and Australo-Melanesia fall inside the within-regios ratio interval.



America: Geographic distance is correlated with Fst between Northern North America series and the other regions of the planet, but not for the other American regions.
Fst between Northern North America and Europe, Australo-Melanesia and Asia fall inside the within-regions ratio confidence interval. North and South America have more complex patterns of relationships with the other regions.



Africa: Geographic distance is not correlated with Fst between African regions and the other regions of the planet.
Fst between African regions and Europe regions fall inside the confidence interval of the within-regions ratio. For the remaining regions, there is a tendency that Fst values with African regions are lower than expected by the within-regions ratio.



Australo-Melanesia: Geographic distance is not correlated with Fst between Australo-Melanesian series and the other regions of the planet.
Fst between Australia and all regions but Africa fall inside the within-regions ratio confidence interval. For Melanesian series, only distances to Asia and Europe series fall inside the within-regions ratio interval.

Australo-Melanesia: Geographic distance is not correlated with Fst between Polynesian series and the other regions of the planet.
Fst between Polynesia and all regions but Australo-Melanesia fall below the within-regions ratio confidence interval.

KEY TO THE SYMBOLS:

- Fst distance with **all** the regions in this continent fall **inside** the estimated within-regions fst/km ratio
- Fst distance with **most** of the regions in this continent fall **inside** the estimated within-regions fst/km ratio
- Fst distance with **most** of the regions in this continent fall **below** the estimated within-regions fst/km ratio
- Fst distance with **all** the regions in this continent fall **below** the estimated within-regions fst/km ratio

6. BETWEEN REGIONS MORPHOLOGICAL DIFFERENTIATION

When average Fst values between regions were calculated, a complete rupture of the linear relationship between Fst and geographic region was apparent (see graphs for each continent above). In fact, none of the regression lines between regions (with the exception of Northern North America) have coefficients statistically different from 0.

These results show that the geographic pattern of differentiation observed within regions is not typical of phenotypic differences between regions. Although these results could be indicating a slower impact of differentiation between regions, it could also be the result of a non-linear relationship between morphological differences and geographic distances. It is reasonable to assume that this relationship would be better described by a logarithmic function than a linear function. However, independent of the nature of the expected relationships, these results show distinct processes of differentiation within and between regions.

7. FST/KM RATIOS

None of the Fst/km ratios between regions are above the expected range observed within regions, indicating that in no case differences between regions are more pronounced than differences within regions when geographic distance is taken into account.

Interestingly, we see that some regions show most of their ratios falling within the expected differentiation process, while others have most of the ratios below the expected range. The regions that fall best within the range are the northernmost regions (North Europe, NE Asia and Northern North America), which have been shown previously to be under stronger selective pressures to cold climate (Hubbe et al. 2009) and consequently may be reflecting an increased rate of morphological differentiation due to selection. On the other hand, African, East Asian, Melanesian, Polynesian and North American regions show lower than expected Fst/km ratios in most of their comparisons. Moreover, the results demonstrate no clear monotonic geographic pattern (i.e., closer regions do not show different patterns from distant regions) in most of the comparisons.

Together, these results suggest that some of the regions with the strongest evidence of natural selection acting on skull shape are the only regions that follow a level of differentiation per distance similar to the observed within regions. The rest of the regions tend to show a reduced differentiation per distance than what is observed within regions.

8. CONCLUSIONS

The preliminary results presented here indicate that the processes associated with the morphological differentiation within and between continents may have differed considerably between regions. This is more in accordance with a process triggered by local events (natural selection or localized microevolutionary processes), rather than a constant and common pattern of differentiation worldwide. Although these processes mimic the differentiation pattern expected from random stochastic processes across time, the assumption of a common and progressive rate of morphological change does not hold.

References cited

Betti L, Balloux F, Hanihara T, Manica A. 2009. Distance from Africa, not climate, explains within-population phenotypic diversity in humans. Proc R Soc Biol Sci Ser B 276: 809–814.
Darroch JN, Mosimann JE. 1985. Canonical and principal components of shape. Biometrika 72:241–252.
Harvati K, Weaver T. 2006. Human cranial anatomy and the differential preservation of population history and climate signatures. Anat Rec 288A:1225–1233.
Hubbe M, Hanihara T, Harvati K. 2009. Climate signatures in the morphological differentiation of worldwide modern human populations. Anat Rec 292A:1720–1733.
Hubbe M, Neves WA, Harvati K. Paleoamerican Morphology in the Context of European and East Asian Late Pleistocene Variation: Implications for Human Dispersion Into the New World. Am J Phys Anthropol 144:442–453.
Manica A, Amos W, Balloux F, Hanihara T. 2007. The effect of ancient population bottlenecks on human phenotypic variation. Nature 448:346–348.
Relethford JH. 2004. Global patterns of isolation by distance based on genetic and morphological data. Hum Biol 76:499–513.
Roseman CC. 2004. Detection of interregionally diversifying natural selection on modern human cranial form by using matched molecular andmorphometric data. Proc Natl Acad Sci USA 101:12824–12829.