

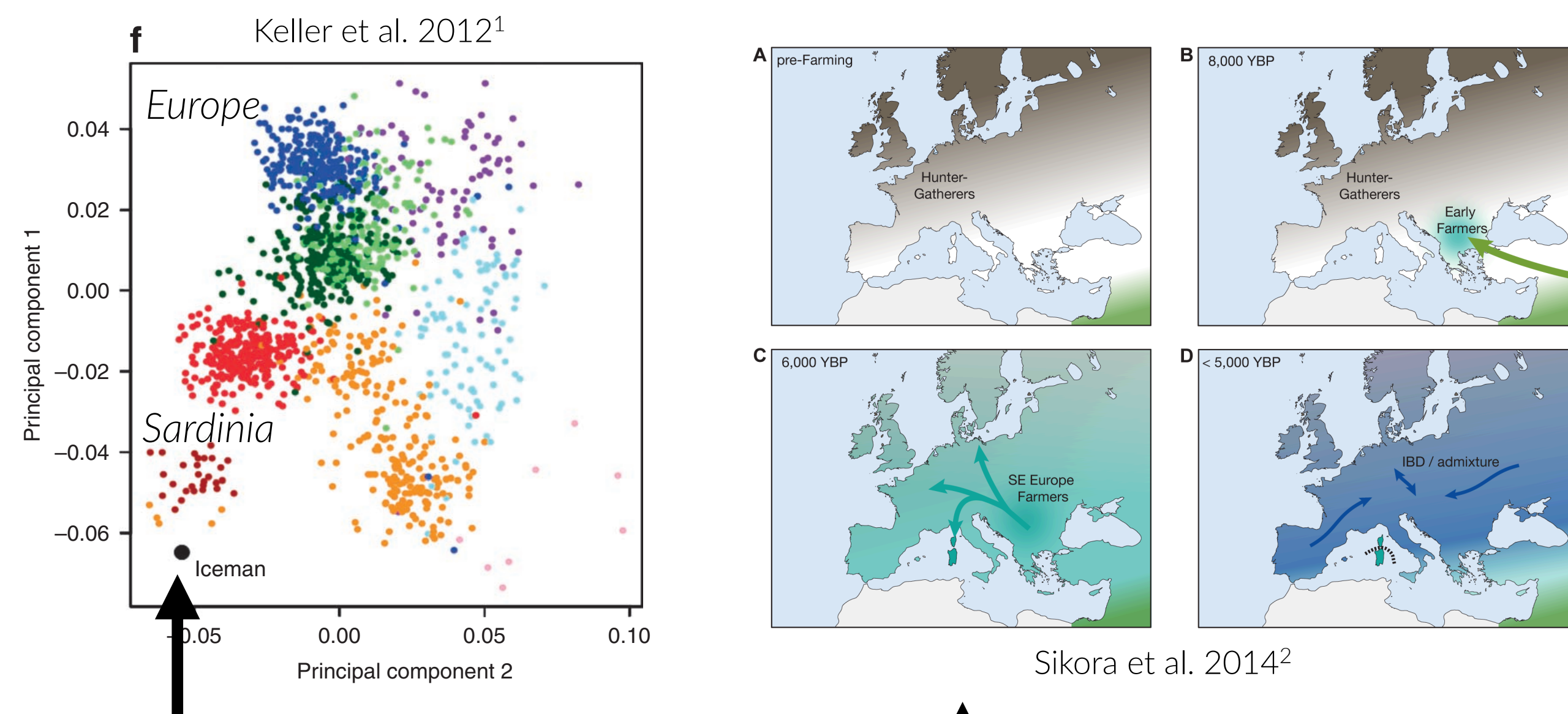
# Using ancient DNA from Sardinia to assess population stability from the Neolithic to present

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## Background

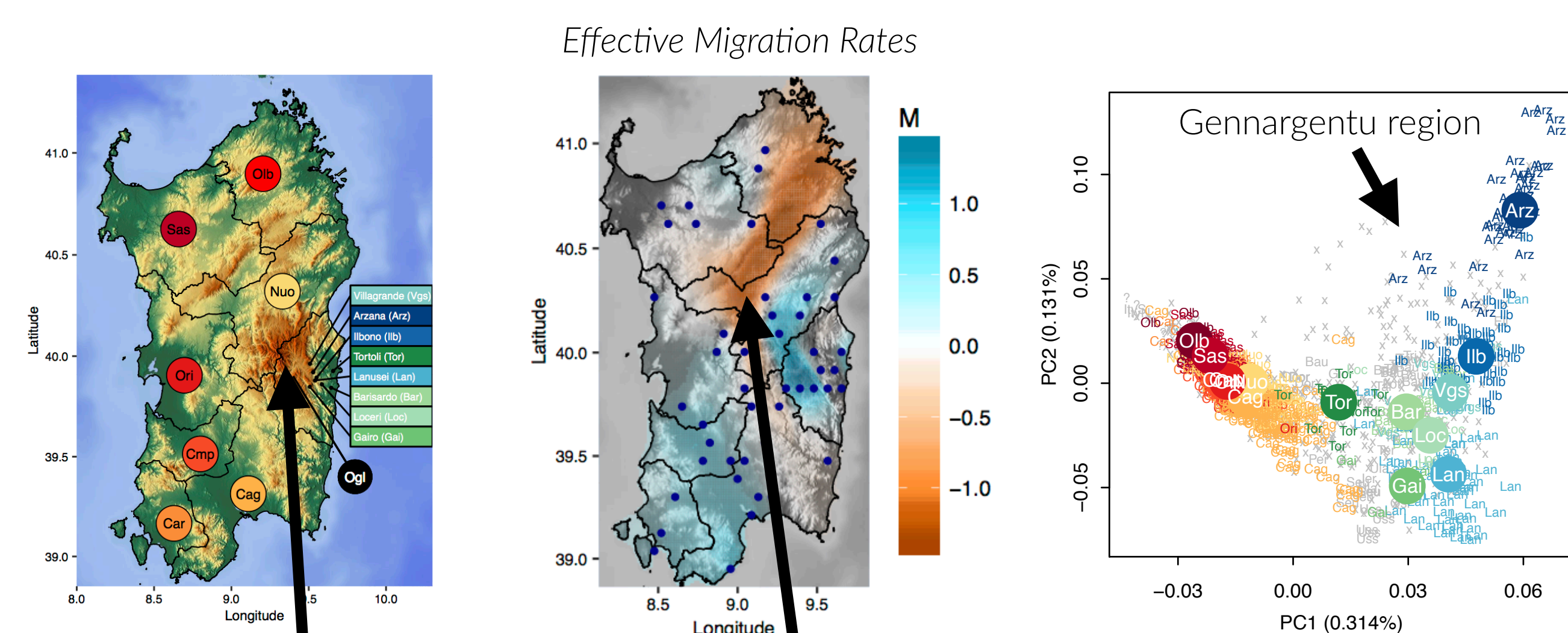
Genetic studies of the Sardinian population have revealed insights into the peopling of Europe



Ötzi, the Tyrolean Ice-Man, genetically clusters with contemporary Sardinians even though he was found in the Ötztal Alps (hundreds of km away). This observation, in addition to other recent studies, suggests a model of a large-scale recent migration event into Europe.

This model has been extended in recent studies to include other large scale migration events contributing to modern variation in Europe, particularly from populations with Steppe ancestry.<sup>3,4,5</sup>

Fine-scale population structure within Sardinia (Chiang et al. 2017<sup>6</sup>)



Geographical features within Sardinia include large mountains in the Gennargentu region.

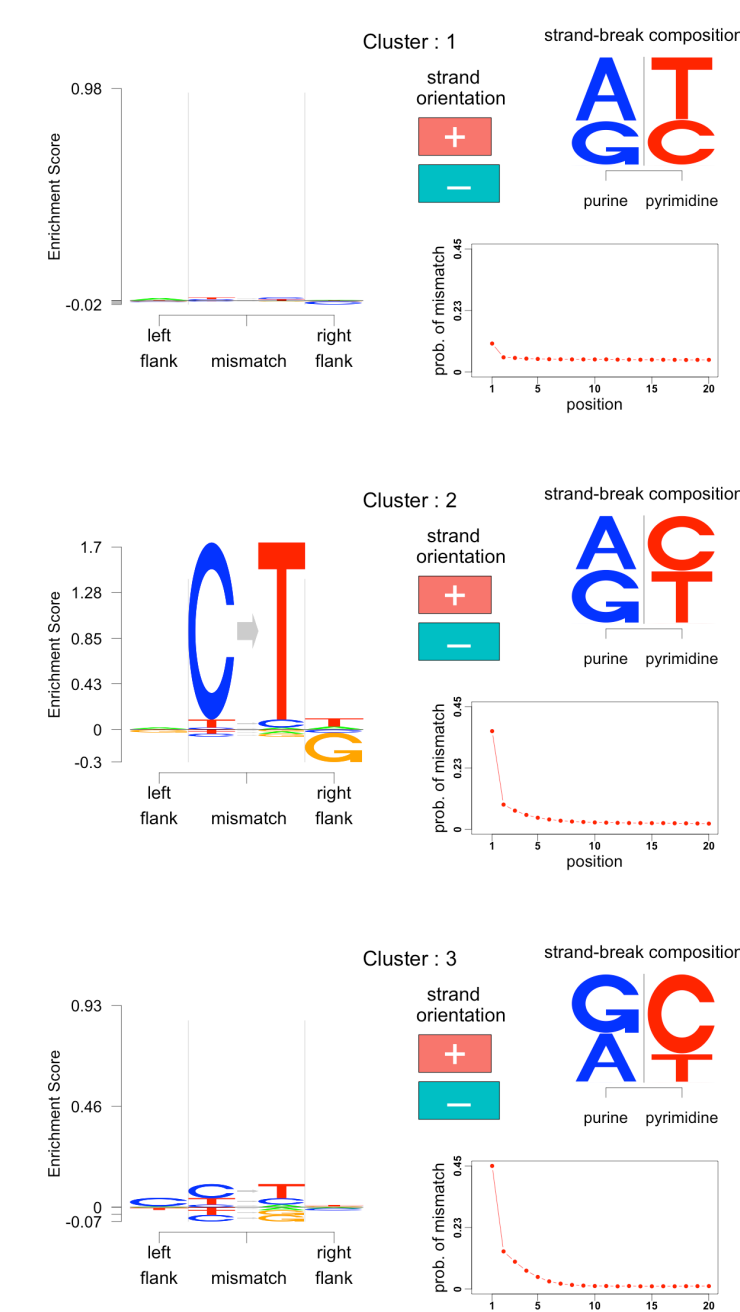
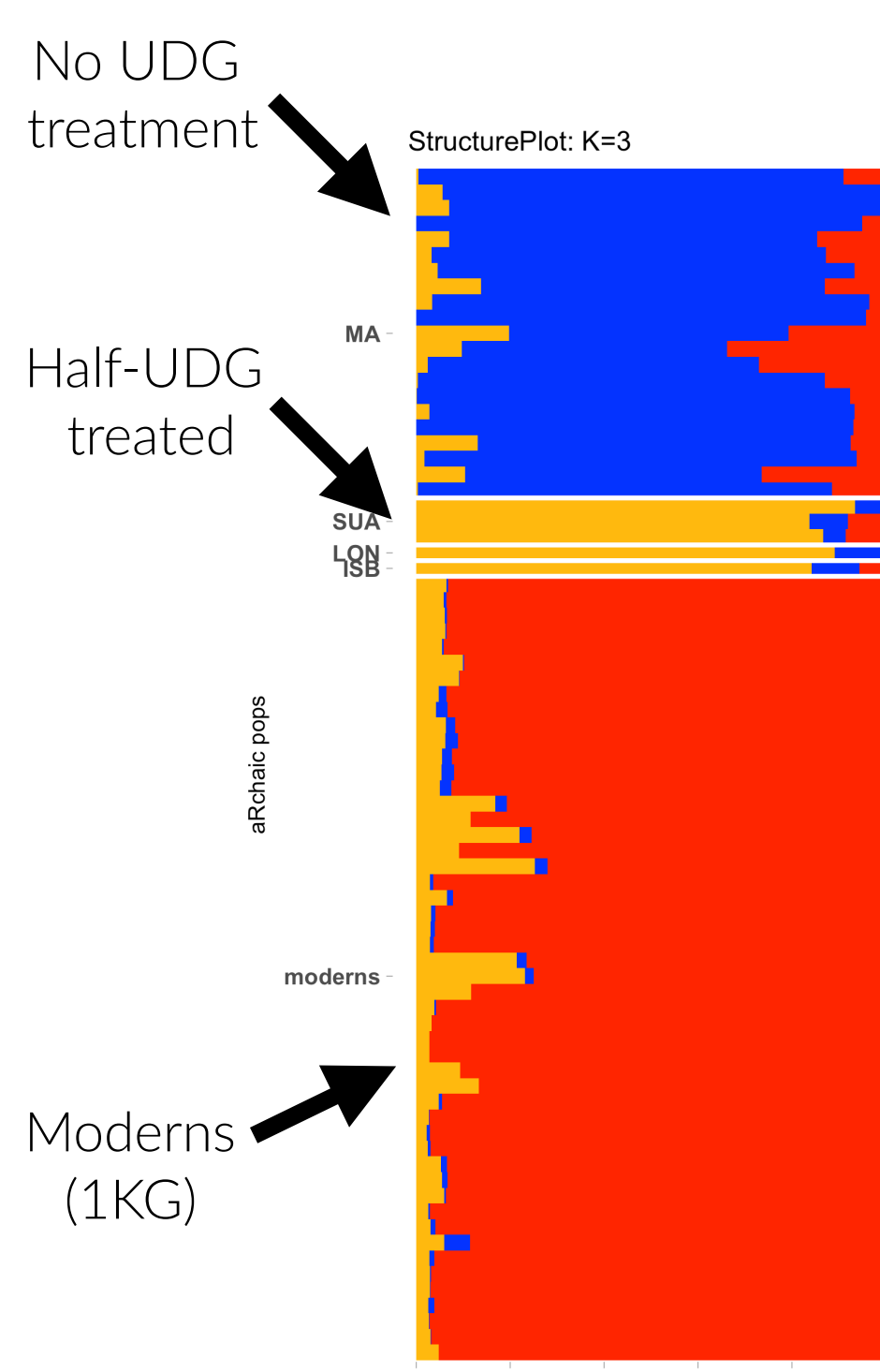
Restricted gene-flow between individuals from the Ogliastra region and elsewhere in Sardinia is shown in both estimated effective migration rates and PCA.

The goal of this study is to use ancient DNA to better understand the population history of the Sardinian people.

## Sample Info / Quality Control



We generated low-coverage capture data at a targeted set of ~1240k SNPs in 26 individuals from different time periods in Sardinia's pre-history.



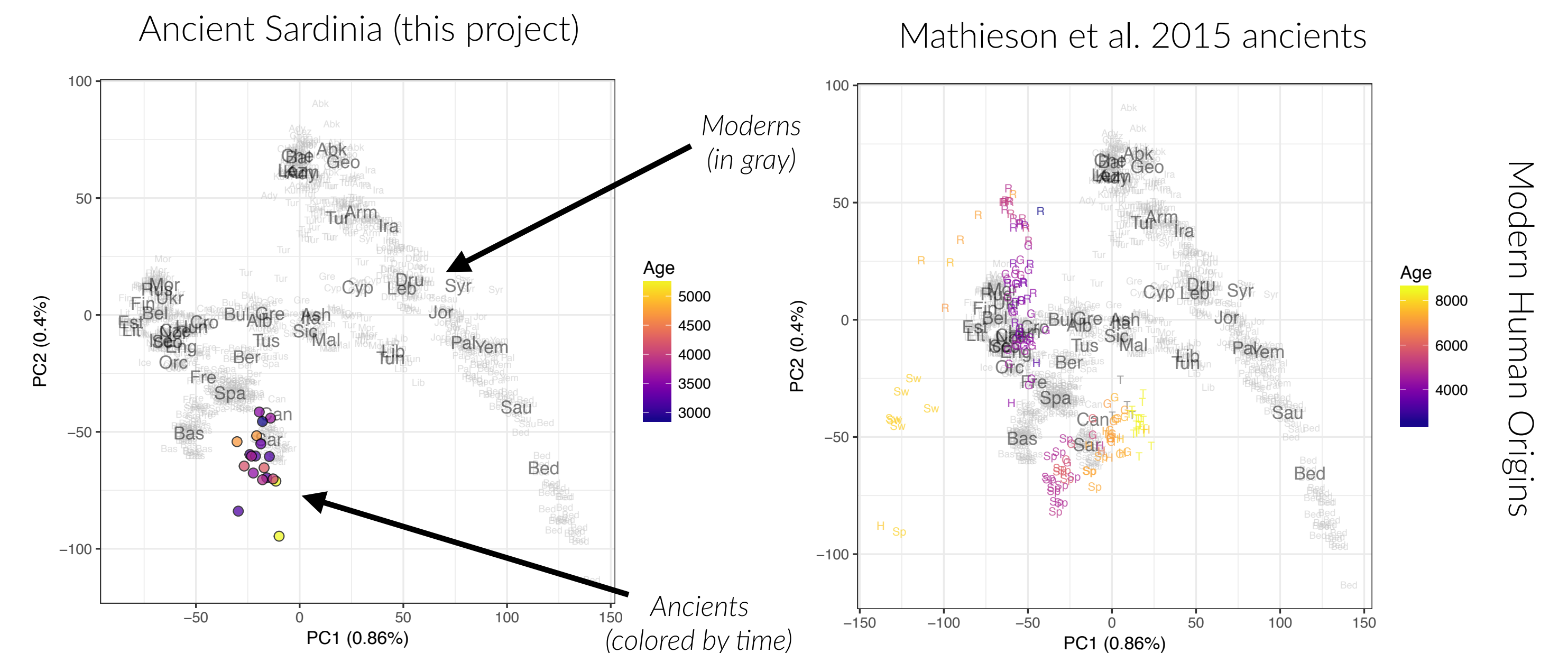
Visualizing ancient DNA damage patterns with a mixed-membership model using *aRchaic*<sup>7</sup> shows ancients have a distinct damage profile when compared to moderns.

see ... <https://github.com/kkdey/aRchaic>

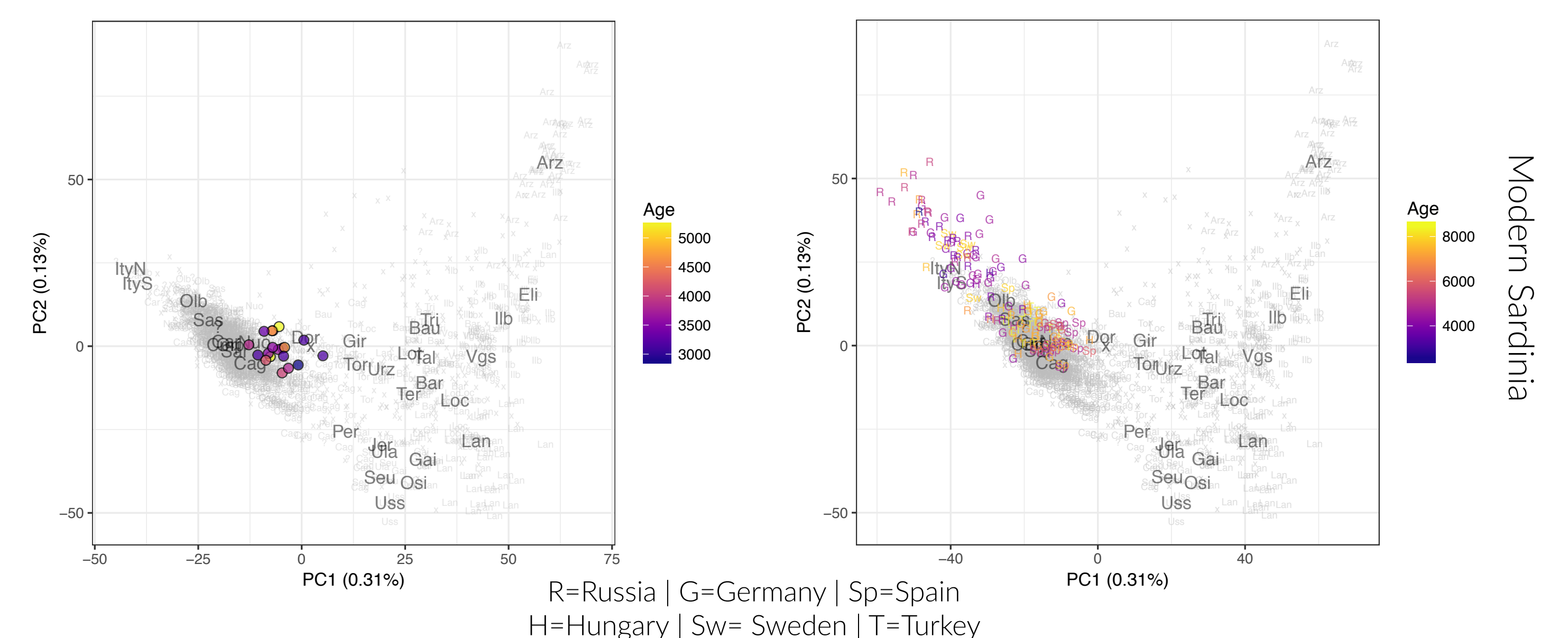
We additionally performed a number of QC steps to filter out individuals who have high contamination rates or too few covered SNPs.

## Population Structure

Projecting ancient individuals onto axes defined by modern genetic variation using PCA

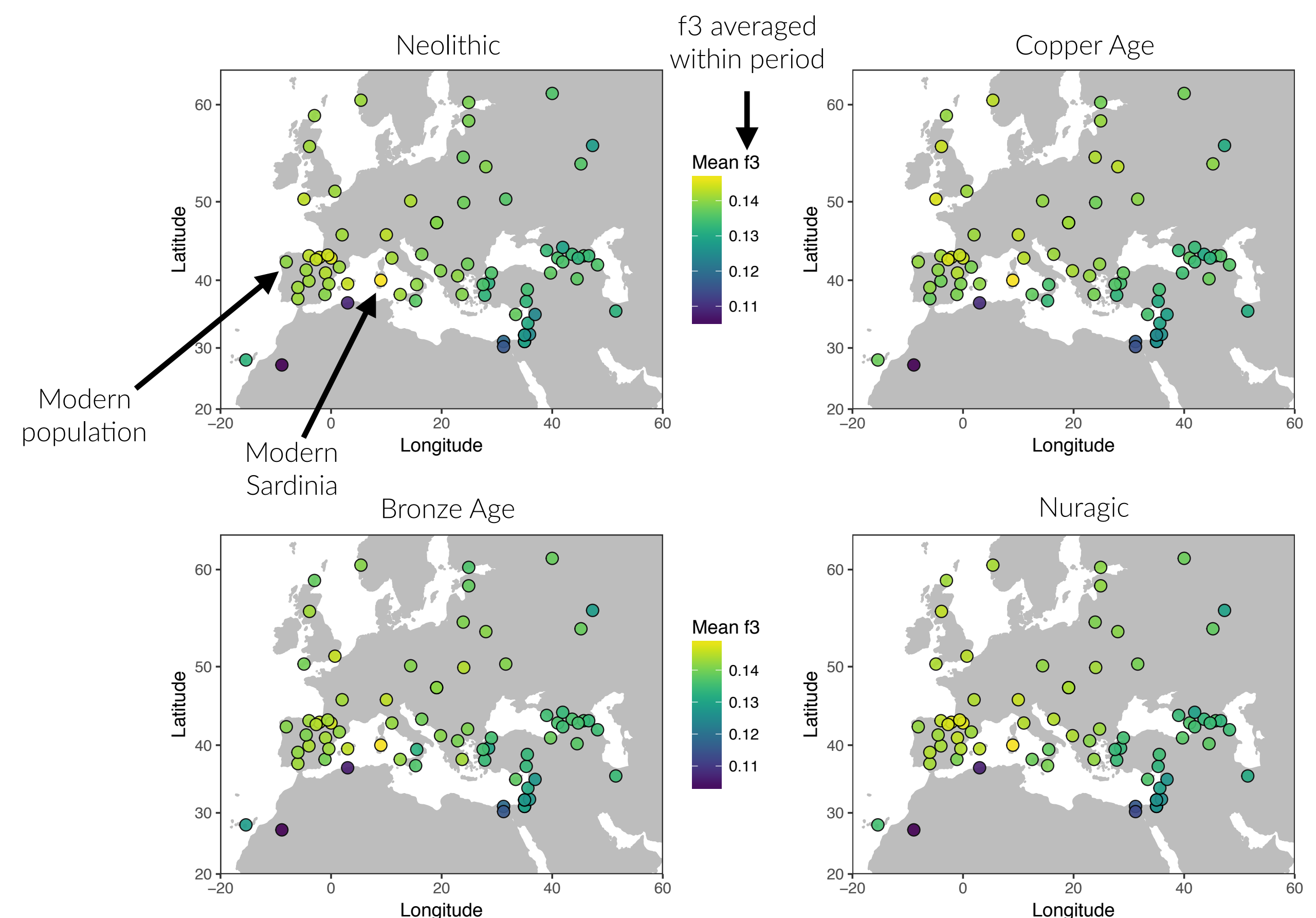


Ancient Sardinians from different time periods cluster with modern Sardinians. This type of temporal stability is not as persistent in mainland Europe.



Ancient individuals project close to Sardinian sub-populations outside the Gennargentu region.

Visualizing shared genetic drift between ancient Sardinia and modern populations through time

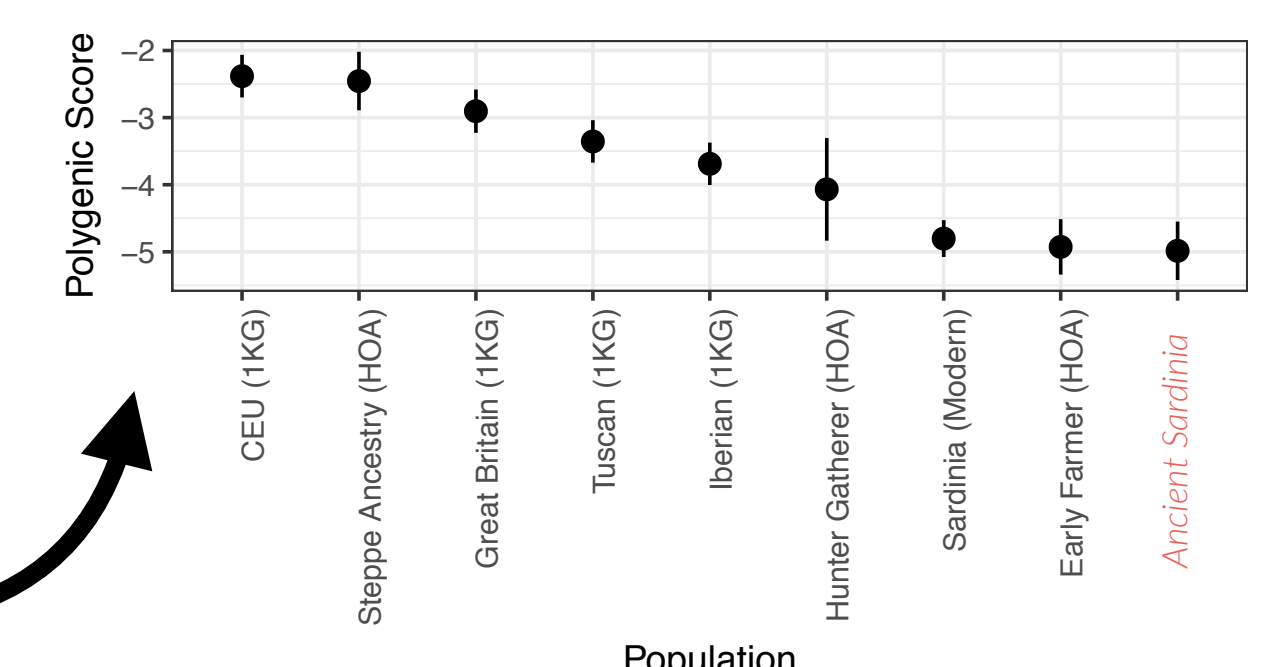


Modern Sardinia and Basque populations shows the strongest shared genetic drift with ancient Sardinia through multiple time periods.

## Conclusions & Future Directions

We observe temporal stability within Sardinia when contrasted to other ancient individuals from mainland Europe.

We are conducting an ongoing study of the evolutionary forces acting on quantitative traits through time in Sardinia (particularly height and auto-immune diseases)



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References

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