

## Detection of archaic hominin introgression in modern African populations

Miguel Navascués and Carina Schlebusch

miguel.navascues@ebc.uu.se

carina.schlebusch@ebc.uu.se

Human Evolution, Evolutionary Biology Centre

<https://www.iob.uu.se/research/human-evolution>

During many years, there were two competing hypotheses about the origins of anatomically modern humans. The out-of-Africa hypothesis suggested a single origin in Africa followed by worldwide dispersal. On the other hand, the multiregionalism hypothesis proposed multiple populations of archaic ancestors across continents, connected by extensive gene flow, which evolved into modern humans. Nowadays, the most accepted model includes elements of both hypotheses: modern humans first appeared in Africa and spread worldwide, they encountered archaic hominin populations and admixed with them. Some genetic diversity patterns in modern human populations (long stretches of DNA in linkage disequilibrium) had suggested this ancient admixture<sup>1</sup>, but it was the sequencing of ancient DNA from Neanderthals and Denisovans that has confirmed this model. The reference genomes of these archaic humans has allowed putting in evidence introgression of 1 to 4% of Neanderthal DNA in Eurasians and Native Americans and around 6% of Denisovan DNA in Australians, Melanesians and some Southeast Asian populations.

African populations have been shown to present the same linkage disequilibrium patterns suggesting ancient admixture. However, they do not show any trace of introgression from Neanderthals or Denisovans. Are these patterns really a signature of introgression from an archaic hominin (other than the Neanderthal/Denisovan lineage)? As the reference genome from other potential candidates is not available, this question can only be addressed by thoroughly assessing the observed genetic diversity patterns with different demographic models including/excluding introgression from a “ghost” population. Recent work<sup>2</sup> developing such approach supports the presence of archaic introgression in modern-day African, in a limited number of samples.

Building on such approaches, the student will adapt and apply them to a larger and more diverse panel of African populations. The objective of the stage will be to produce a detailed picture of this introgression, specifically answering the following questions: Is there a single or multiple archaic sources? Do different African population present different levels of introgression?

Requirements: Some knowledge of population genetic models and the measurement of genetic diversity are necessary for this project. Introgression inference will be based on a simulation based approach, commonly known as Approximate Bayesian Computation<sup>3</sup>. It consist the the generation of a training data set through simulation that will be used to train a machine learning algorithm (typically random forest). A minimum knowledge on R and bash is required, knowledge of Python will be ideal, to put into practice these analyses.

Keywords: approximate Bayesian computation, human evolution, admixture, archaic humans

### References:

<sup>1</sup> Plagnol V, Wall JD (2006) Possible Ancestral Structure in Human Populations. PLoS Genet 2(7): e105. <https://doi.org/10.1371/journal.pgen.0020105>

<sup>2</sup> Durvasula A, Sankararaman S (2019) Recovering signals of ghost archaic introgression in African populations. bioRxiv <https://doi.org/10.1101/285734>

<sup>3</sup> Csilléry, Blum, Gaggiotti, François (2010) Approximate Bayesian Computation (ABC) in practice. Trends in Ecology & Evolution. <https://doi.org/10.1016/j.tree.2010.04.001>