

Genomics study in Africa—demographic history and deleterious mutations

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Scientists from the Institut Pasteur set out to understand how the demographic changes associated with the Neolithic transition also influenced the efficacy of natural selection. By comparing the genome diversity of more than 300 individuals from groups of forest huntergatherers (pygmies) and farmers (Bantu-speaking peoples), from western and eastern Central Africa, they discovered that the reason pygmies did not suffer from excessive deleterious mutations was because of their genetic diversity and their admixture with the Bantu peoples.

Research into the diversity of the human genome and any rare or frequent <u>mutations</u> is vital in identifying mutations that increase susceptibility to complex diseases such as infectious or autoimmune disorders. "The elimination of these mutations that are harmful for human health is largely conditioned by the evolutionary history of populations, especially their demographic fluctuations," explains Lluis Quintana, Head of the Human Evolutionary Genetics Unit and author of the study. Throughout their history, human populations have undergone significant and extremely varied changes in terms of numbers of individuals, giving rise to differences in the number and severity of mutations carried by these populations. The accumulation of diseasecausing genetic variants, known as the "burden of deleterious mutations", varies from one population to another depending on its past. "The aim of our project is to understand how demographic events such as the rise and fall in numbers of individuals, as well as genetic admixtures, have had an impact on the efficacy of some evolutionary mechanisms such as purifying selection."



Comparing the genome diversity of two populations

The scientists focused their research on a comparison of populations that lived through one of the most significant socio-cultural transitions in human history: the transition to agriculture. By comparing the genome diversity of more than 300 individuals from groups of forest hunter-gatherers (pygmies) and farmers (Bantu-speaking peoples) in western and eastern Central Africa, the study aimed to shed light on the way in which demographic changes associated with the Neolithic transition also influenced the efficacy of <u>natural selection</u>. "We therefore characterized the genetic structure of these populations using exome sequencing data [editor's note: the exome is a fraction of the genome that contains protein-coding regions] and used computer modeling techniques to reconstruct changes in population size over the past 200,000 years," continues Lluis Quintana.

Against all expectations, his team observed that the current groups of forest hunter-gatherers are descendants of prosperous ancestral populations with a genetic size comparable to that of the farmers' ancestors. Simulations also indicate the existence of genetic admixture between the groups more than 20,000 years ago.

"It was more recently, over the past 20,000 years, that the demographic development of these two populations became diametrically opposed, with the hunter-gatherer <u>population</u> size falling dramatically [approximately 80%] while the number of early farmers tripled."

Measuring the impact of extreme demographic changes

In a bid to understand and quantify the impact of these extreme demographic changes on the burden of deleterious mutations, the



scientists measured the efficacy of purifying selection in the two groups and examined its dynamics over time. "This enabled us to measure the rise and fall in the burden of deleterious mutations as the <u>population size</u> contracted and expanded," explains the scientist. "Based on the study, we were also able to predict that, in the specific case of African huntergatherers and farmers, the current burden of mutations is similar in the two groups, despite their different demographic histories and lifestyles."

What prevented the hunter-gatherers suffering from an excess of deleterious mutations was both their high ancestral genetic diversity and a strong and constant admixture with farmers. The scientists' predictions were then verified using DNA sequencing data to empirically quantify the number of deleterious mutations carried by each individual. No differences were detected in the number and severity of mutations carried by forest hunter-gatherers and farmers.

More information: Marie Lopez et al, The demographic history and mutational load of African hunter-gatherers and farmers, *Nature Ecology* & *Evolution* (2018). DOI: 10.1038/s41559-018-0496-4

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