università di ferrara
DA SEICENTO ANNI GUARDIAMO AVANTI.

A very, very schematic view of the evolution of human genetic diversity

Guido Barbujani
Dipartimento di Biologia ed Evoluzione, Università di Ferrara
g.barbujani@unife.it

This is an attempt to assemble a coherent, if admittedly oversimplified, picture of human population history.

I am aware that the synthesis thus operated is brutal. Many other demographic processes, migrations and phenomena of isolation ad extinction, must have happened in the meantime. Yet, if this model is just vaguely accurate, it explains why human alleles are basically either African or cosmopolitan; why so many indexes of genetic diversity are associated with distance from Africa; why human genetic diversity is largely clinal across the Old World; and why sharp genetic boundaries between populations and groups thereof are so rare that it proved impossible to agree on a list of human races.

Panel 1 represents the ancestral populations when anatomically modern humans were restricted to Africa, some 100,000 years ago; the different colours represent different genotypes. The rest of the world was not devoid of humans; there were anatomically archaic people in Asia and Europe, but it is unlikely that they have contributed to the modern gene pool, and in any case that contribution must have been minimal (Foley 1998).

In **panel 2** we represent an expansion of the African population, which is accompanied by the generation of new alleles by mutation. Humans could not produce food at that time, and moved around looking for bearable living conditions. In the course of their movements, some of them reached north Africa, and in this scheme they were mostly carrying yellow and orange genotypes.

When, around 60,000 years ago (Liu et al. 2006), these people crossed into Eurasia (here we did not represent a possible Southern route of dispersal in the Arab peninsula, through the horn of Africa: Macaulay et al. 2005), they entered a territory with greater resources and very low population density (**panel 3**). The main consequence was an improvement in living conditions, resulting in a demographic growth which, however, affected only the descendents of the people who left Africa, here represented by yellow, orange, and green genotypes.

In **panel 4** the African emigrants' descendents have already ventured into Central Asia, and in **panel 5** some of them are sharing (peacefully, or not-so-peacefully) part of the European territory with Neandertal people.

By 10,000 years ago, the first peopling of the planet is almost complete (**panel 6**). Not represented in these figures is the migration that brought (perhaps 15,000 years ago) the first humans into the Americas, probably through a land bridge crossing what now is the Bering Strait. During all this lapse of time, mutations occurred, both in Africa and outside. However, at the end of the major expansion process (which was certainly accompanied and followed by other momentous demographic changes) the African alleles had dispersed worldwide.

The pie diagrams in **panel 7** are meant to represent variation at a typical human locus, in which Africa shows a large number of alleles, both continent-specific and cosmopolitan. By contrast, each of the gene pools of the non-African populations is largely (although not exclusively) composed of a different subset of African alleles, sometimes brought to high frequencies by genetic drift. By effect of the repeated founder effects, the yellow genotype forms a West-East cline encompassing all Eurasia, with maximal frequencies in China.

THREE REFERENCES

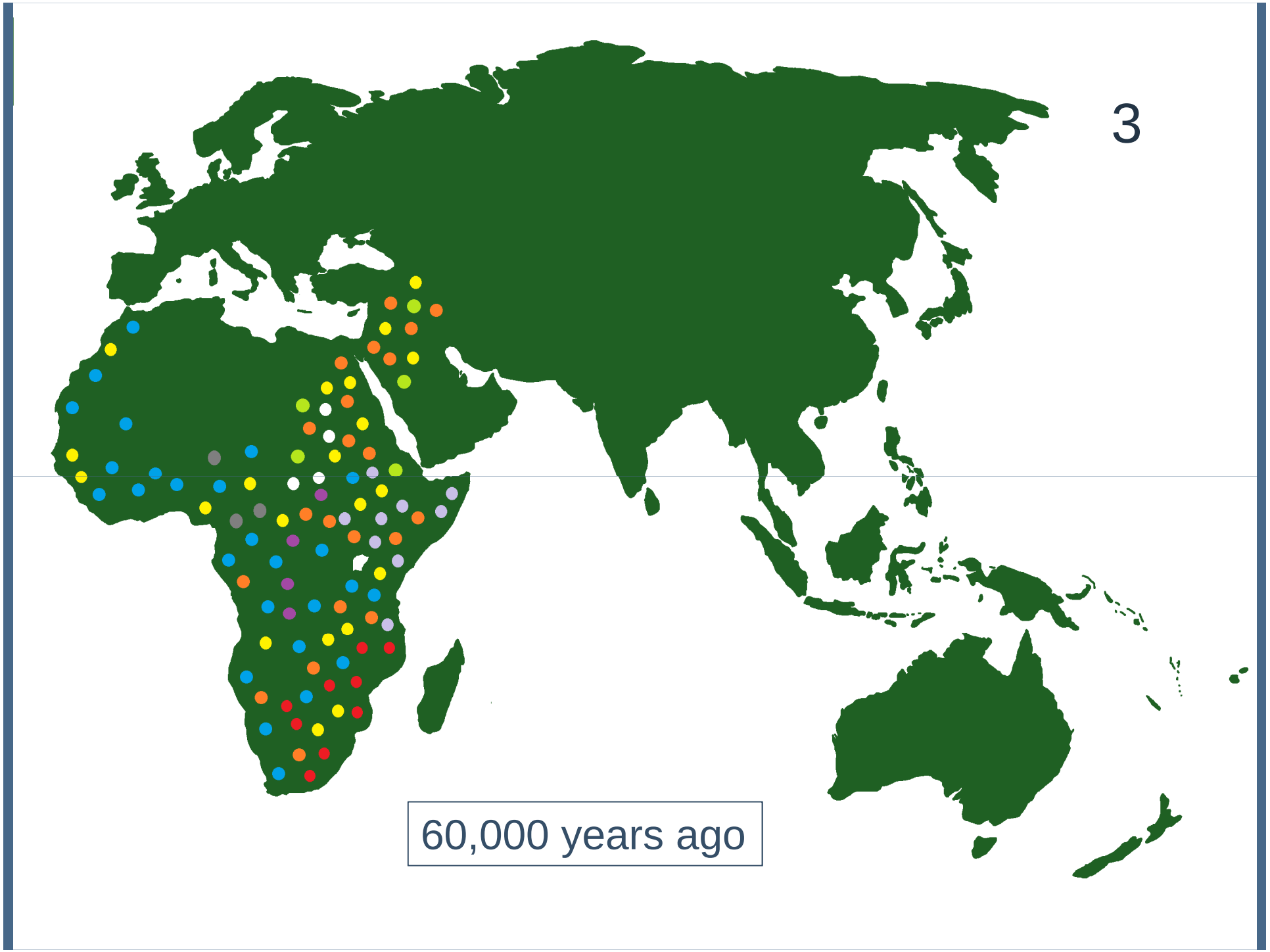
Barbujani G, Colonna V (2010) Human genome diversity: Frequently asked questions. *Trends in Genetics*, in press

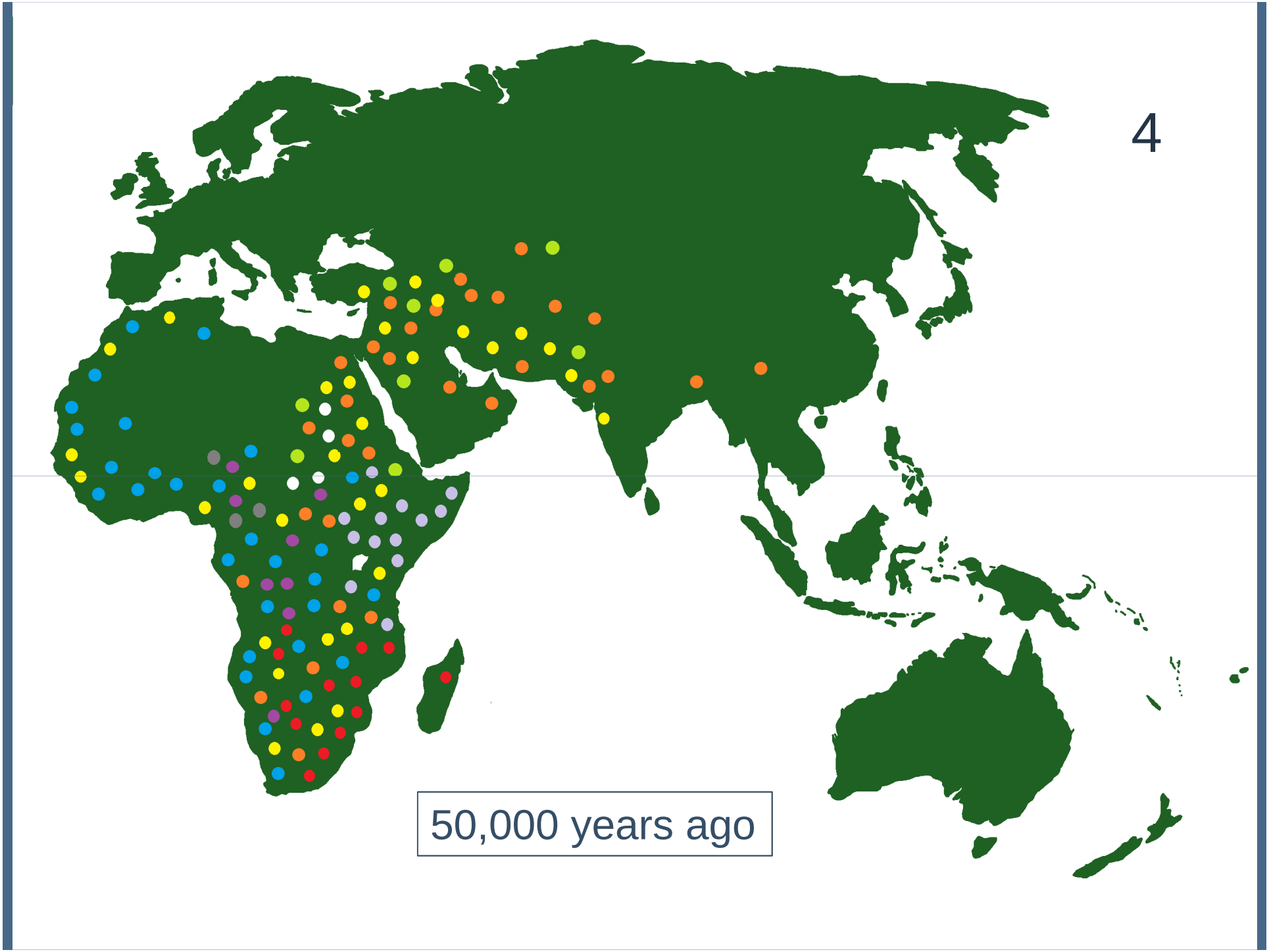
Foley R (1998) The context of human genetic evolution. *Genome Research* 8:339-347

Liu H, Prugnolle F, Manica A, Balloux F (2006) A geographically explicit genetic model of worldwide human-settlement history. *American Journal of Human Genetics* 79:230-237

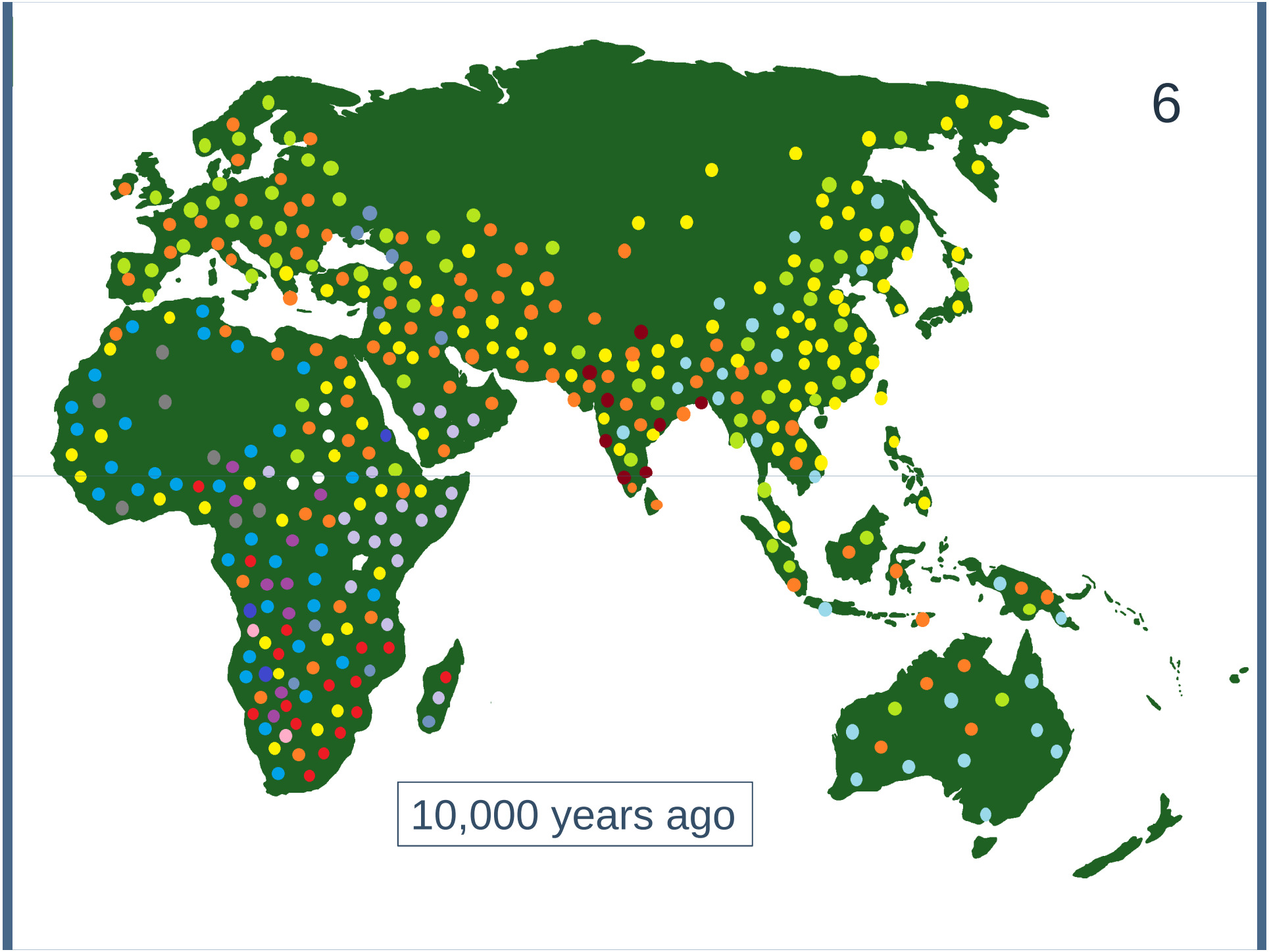

1
100,000 years ago

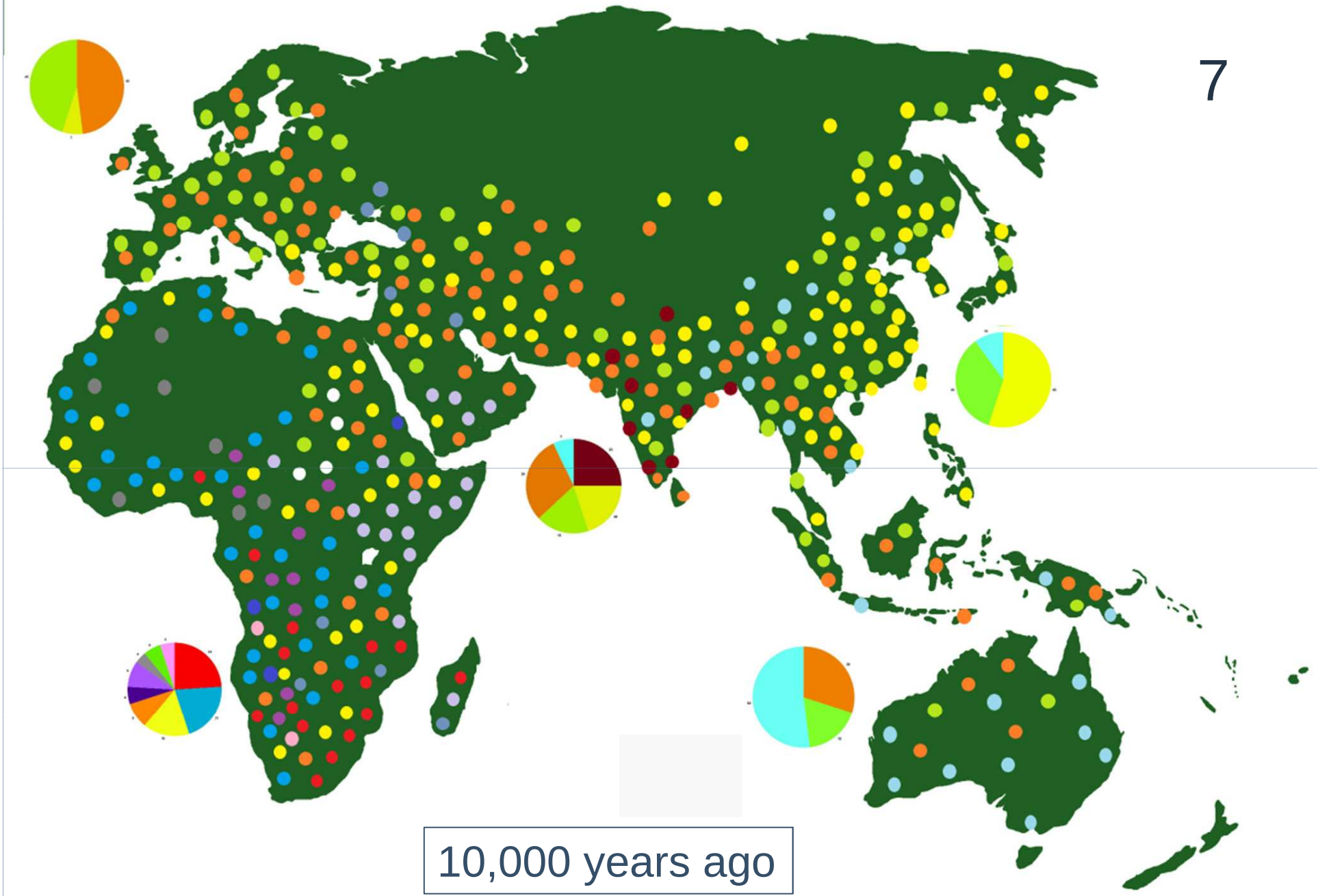

2
70,000 years ago


3
60,000 years ago


4
50,000 years ago


30,000 years ago


10,000 years ago



10,000 years ago