



IMAGE SOURCE

HUMAN EVOLUTION

Multigenome coalescence for modern demographic insights

Throughout evolution, human genomes are shaped by both recombination during sexual reproduction and spontaneous mutations. Hence, genomes of modern humans harbour signatures of their history, and the interpretation of these signatures to reconstruct past demographic events is a major goal. A new study uses multiperson genome comparisons to obtain insights into more recent events in human population history than were previously possible.

Coalescence analysis involves tracing chromosomal lineages back in time until the most recent common ancestor is reached. Such analyses can infer historical population sizes and the timings of population splits. Building on the pairwise sequentially Markovian coalescent (PSMC)

model for the analysis of two haploid genomes (that is, haplotypes) in one individual, Schiffels and Durbin developed the multiple sequentially Markovian coalescent (MSMC) model for the simultaneous analysis of up to eight haplotypes from four individuals. As MSMC characterizes the first historical coalescence between any pair of the haplotypes, this provides information on more recent events than the limit of PSMC (~20,000 years ago).

The authors first tested their model by simulating various human genome evolution scenarios and showed that the MSMC model can process the resultant genomes to reconstruct information about population splits, migration and population size changes as recently as 2,000 years ago.

Schiffels and Durbin then analysed real genome sequences of modern humans from nine geographically diverse populations. The multigenome comparisons refined numerous aspects of human history. They showed that the genetic 'out-of-Africa' split of non-Africans from modern African populations probably also reflects splits within Africa long before ~50,000 years ago.

Furthermore, they traced subsequent population splits and size changes, which extended to the split between southern Italian and northern European populations only 6,000–5,000 years ago.

Future studies will reveal whether tools to compare more than eight haplotypes can provide insights into even more recent events.

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ORIGINAL RESEARCH PAPER Schiffels, S. & Durbin, R. Inferring human population size and separation history from multiple genome sequences. *Nature Genet.* <http://dx.doi.org/10.1038/ng.3015> (2014)