Human history recorded in a single genome

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WANT to know the history of your ancestors? Look no further than your genome. It seems every one of us carries in our genes a million-year record of past human population size.

Analysing the ways that mitochondrial DNA sequences differ across a large number of living people has helped to establish prehistoric population trends, but this record stretches back only 200,000 years to the point where all humans alive today shared a common female ancestor. That's because mitochondrial DNA passes down from mother to child.

Richard Durbin of the Wellcome Trust Sanger Institute in Cambridge, UK, and Heng Li at the Broad Institute in Cambridge, Massachusetts, can push the record back five times as far by reading a single genome.

Taking advantage of the handful of complete human genome sequences now available, the pair looked at how alleles - the two copies of each gene we inherit from our parents - differ within a genome. Many differences between the two copies suggest that they separated some time ago, while similar copies have a more recent separation date.

By reading thousands of alleles and estimating mutation rates, the duo can work out the separation date for each allele and calculate past population sizes. For instance, evidence that many alleles share the same separation date suggests the population was small and genetically similar at the time.

Durbin and Li analysed seven complete sequences: one each from China and Korea, three of European origin and two from west Africa. The pair concludes that European and Chinese populations both suffered a severe bottleneck between 10,000 and 60,000 years ago, while African populations endured a milder bottleneck at that time (Nature, DOI: 10.1038/nature10231).

"The idea that each human genome contains information about the history of its ancestors' population size has been known theoretically, but we have never had the data or methods to pull out that information until now," says John Novembre of the University of California, Los Angeles.
Ryan Gutenkunst of the University of Arizona in Tucson is also impressed by the study. "The method is really spectacular. Previous methods have taken averages across the genome, but here they are looking at variation from one location to another location and getting good results from even a single individual."

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