The Y Chromosome and the Replacement Hypothesis

Ke et al. (1) raised old questions about the origins and evolution of modern humans in East Asia. Unfortunately, they have shed no new light on the subject, their study amounting only to a large-scale genotyping of three previously observed Asian Y chromosome alleles (2). The authors provided no explicit statistical tests of any of their conclusions, which in any event are not new (3). Nor did they address the relationship of the information from the Y chromosome, a single genetic locus in the classical sense, to other genetic loci that might provide independent evidence for their preferred phylogenetic scheme—other than to assert that other loci are inadequate for such inferences by virtue of their mutational dynamics. The inadequacy of scientific and historical perspectives in the paper may leave many readers wondering how any competent researcher could possibly hold a view contrary to that of these authors.

Many do disagree, however. In particular, no information provided by Ke et al. contradicts the studies of Hammer et al. (4) and Templeton (5), which viewed Y chromosome variation from a multilocus perspective. These earlier studies concluded that a global replacement of ancient Y chromosome variation did occur, probably from an African source. This ancient event apparently did not result in the replacement of the entire nuclear or mitochondrial genomes, but instead reflected a more limited genetic phenomenon upon a background of worldwide gene flow and isolation by distance. Furthermore, the Y chromosome variation appeared to indicate a later population movement out of Asia into Africa. It is not clear whether the ultimate explanation for Y chromosome variation is selection or male reproductive dynamics, but whatever is the case, there need be no a priori expectation that Y chromosome variation must be the only element of genetic variation requiring explanation, as assumed by Ke et al. (1). Perhaps Ke et al. disagree with the conclusions of these earlier papers, but they presented no reasons to disbelieve them, and any argument based only upon data from a single locus can never fully address the evolutionary history of the people who carried that gene.

Also, notwithstanding the implication of Ke et al., fossils and archaeology are not silent on this issue. Although diagnostic skeletal remains between 120,000 and 50,000 years old within China are rare, the supposed absence of “missing links” (1) is not particularly relevant. The important morphological and archaeological observations relevant to the possibility of population replacement are (i) the occurrence of distinctive morphologies in both archaic and recent Asian populations, and (ii) the complete absence of any archeological evidence for an influx of a new, presumably African population (6–8). Absent extraordinary circumstances—such as the replacement of archaic East Asians only by the Africans who look most similar to them, and only by those willing to abandon their own material culture and pick up the indigenous material culture—the most reasonable explanation for these facts is a partial contribution of archaic East Asians to the present human population. Remarkably, the only citations provided by Ke et al. (1) to contest the continuity of morphology and archaeology in East Asia were the previous assertions by some of their own coauthors, all laboratory geneticists.

At the moment, at least, the only strong evidence for replacement of East Asian populations comes from the Y chromosome. It is important not to overinterpret this single source of evidence, but instead to view it in light of data from other genetic sources and archaeological and fossil sources into a comprehensive view. Such a balanced view argues against a simple hypothesis of replacement for East Asians or, indeed, for any ancient human population.

John Hawks
Department of Anthropology
University of Utah
Salt Lake City, UT 84112, USA

References

Response: Our recent study (1) showed that all 12,127 Y chromosomes sampled from 163 Asian populations carry a mutation (M168T) that traces to Africa. That observation supports a complete replacement of local archaic humans in Asia by modern humans from Africa, as suggested in the “Out-of-Africa” or replacement hypothesis (1, 2). Hawk criticizes this work, arguing that (i) the history of the Y chromosome may not reflect the evolutionary history of the nuclear and mitochondrial genomes, (ii) our study provided no explicit statistical test to support its conclusions, and (iii) our discussion of historical and fossil evidence was inadequate. None of these criticisms dilutes the strength of our conclusion. The power of Y chromosome markers for testing hypotheses of modern human origins lies in the specific qualities of the male sex chromosome—nonrecombination, low mutation rate, and a high sensitivity to population events such as migrations (1, 3–5). The logic underlying the study design was simple: Based on the solid phylogenetic relationship among worldwide Y chromosome haplotypes—in which only Africans possess the ancient lineages, and all non-African populations share a relatively recent mutation at locus M168 (4)—one would expect the ancient lineages to be found in modern Asian populations if a local contribution did exist. With a fairly large sample size and well-represented population coverage, our data refuted the existence of any local contribution as suggested by advocates of the multiregional hypothesis. The statistics used were likewise simple: The probability of not observing a 0.1% local contribution due to sampling error was 5.4 × 10⁻⁶. In other words, a local contribution even as small as 0.1% does not seem to exist in modern Asian populations. Of course, Y chromosome data reflect only the population history of males. Because a robust phylogeny is essential to test the hypothesis that indigenous archaic humans were completely replaced by modern humans of African origin, the Y chromosome markers are the best choice so far. Other genetic systems, such as mitochondrial and autosomal markers, have certain limits: Recombinations would blur the phylogenetic connections among mutations on autosomes, and the high recurrent mutation rate in the mitochondrial DNA control region, the most extensively studied mtDNA segment, creates the same problem. However, the recent effort of whole-genome sequencing of mitochondrial DNA in worldwide populations provides a good resource for identifying non-African-specific markers in the coding region of mtDNA that can be used to test the “Out-of-Africa” hypothesis in the female lineage (6). Furthermore, mutations located in the autosomal regions that lack recombination would also be candidates for such study.

Historical and fossil evidence is indeed valuable as a view of the fixed moments of historical events. But it can hardly illuminate whether human beings living at that moment actually contributed to the extant populations. The rich hominid fossil remains unearthed in China and other parts of East Asia have been taken as strong support for the independent origin of modern humans there. The significant gap of human fossils between 100,000 and 40,000 years ago in East Asia (5), however, cannot be circumvented by the simple
explanation of a “missing link,” in view of the abundance of human fossils excavated in that area. The dating of those fossils was conducted by archaeologists, not geneticists; nonetheless, we argue that the time frame of the fossil gap coincides with the duration of the last Ice Age and is consistent with the hypothesized extinction of archaic humans and the entry of modern humans of African origin into East Asia.

The origins of modern humans is indeed a very old subject—and a still-unsolved mystery that calls for new scientific insights from both genetic and archaeological evidence. A large-scale systematic genetic analysis of hominid fossils (i.e., ancient DNA) may provide the opportunity to reconcile the differences between genetic and paleoanthropologic evidence.

Li Jin
Bing Su*
Department of Environmental Health
University of Cincinnati
3223 Eden Avenue
Cincinnati, OH 45267–0056, USA
and
State Key Laboratory of Genetic Engineering
Institute of Genetics
School of Life Sciences
Fudan University
220 Handan Road
Shanghai, China 200443

*Also Kunming Institute of Zoology, the Chinese Academy of Sciences, Kunming, China

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