Gene Lineages and Human Evolution

In a recent article, Francisco J. Ayala (*1*) does an excellent job of debunking many of the myths surrounding the "mitochondrial Eve" hypothesis. However, one myth is perpetuated. In discussing the "multiregional model" of gene flow, Ayala (*1*) correctly notes that this requires "persistent migrations and interbreeding between populations from different continents", but then incorrectly adds that there is "no direct evidence" of this in the genetic data. However, strong and statistically significant evidence of recurrent genetic exchange between prehistoric human populations on different continents does exist. When geographical data are overlayed on the mitochondrial DNA (mtDNA) haplotype tree with rigorous statistical evaluation and interpreted with the use of explicit and objective criteria (*2*), a fractal-like pattern of continental distributions is observed that recurs throughout the entire mtDNA genealogy at all time depths (*3*, *4*). Such a pattern could have arisen from recurrent gene flow (albeit at low levels in this case), but could not have arisen from a single episode of range expansion, such as an out-of-Africa replacement (*2*, *3*, *4*) (although no intercontinental population expansions were detected in these studies, regional ones were, such as an expansion across Europe).

Ayala (*1*) states that African and non-African populations "split" about 156,000 years ago. This "split" hypothesis is based on the genetic distances (the distances created by allele frequencies that differ between populations) found in nuclear DNA. If such a split between human populations had occurred, the genetic distances would be a reflection of the time since the split. An alternative explanation is that the genetic distances reflect a pattern of recurrent but restricted gene flow among human populations throughout recent human evolutionary history, with lower amounts of gene flow leading to larger genetic distances. One mechanism for restricted gene flow is isolation-by-distance, which predicts that the most geographically distant populations would have the lowest amount of effective gene flow. These two explanations make different and testable predictions. A split between African and non-African populations would affect not only nuclear genetic distances, but also the geographical overlay on the mtDNA haplotype tree. Explicit and objective criteria exist for inferring splits with gene tree data (*2*), but no split is detected from the human mtDNA data (*3*, *4*). A split between Africans and non-Africans would also imply that Asians and Europeans would be equally distant genetically from Africans, and Asians and Europeans should be equidistant from their common ancestral node. In contrast, the restricted gene flow hypothesis with isolation-by-distance (the model indicated by the mtDNA analysis *3*, *4*) predicts that Asians should be the most distant genetically from Africans, with Europeans lying in between. The nuclear data sets referenced by Ayala (*1*), and many other sets, support the restricted gene flow hypothesis and often provide a statistically significant rejection of the African–non-African split hypothesis as well (*3*, *4*). Thus, we know of no evidence that supports the hypothesis of an African–non-African population split in either the mtDNA or the nuclear DNA data. Rather, research indicates that all humans constitute a single evolutionary lineage, with populations showing (i) regional genetic differentiation because of restricted, but recurrent, gene flow along with (ii) some recent, regional range expansions (*3*, *4*).

What do these conclusions imply for the origin of anatomically modern humans? Different modern traits could have evolved in different geographical regions, and then spread throughout all of humanity by the combined effects of gene flow and selection (*4*, *5*). Alternatively, modern traits could have arisen first in a single geographical location (within the range of ancient humans, which includes Africa) and then spread throughout all of humanity (again by the combined effects of gene flow and selection). Because gene flow was restricted, regional genetic differentiation among human populations would be expected and could persist even as the genes for anatomically modern traits were spreading (*4*).

Recent debates (*6*) about the humanoid fossil data are based on the myth that replacement of one physical feature in a fossil series with another feature can only be created by one population replacing another (by exterminating them, for example), but such fossil patterns could be a reflection of one genotype replacing another through gene flow and natural selection. Morphological replacement should not be equated with population replacement when one is dealing with populations that can interbreed.

The mtDNA and nuclear DNA data show that gene flow occurred in Old World human populations throughout recent human evolution, but these data do not help one to discriminate between the multi-region and single-region gene flow hypotheses (discussed two paragraphs above). This can only be done with the use of fossil data. The two data sets are complementary: Fossils can reveal the patterns of evolution, and genes can reveal the processes that could have lead to the observed patterns. The most important information that the genetic data have yielded so far is that humans evolved into their modern form as a single unit, despite past and current regional differentiation for some traits.

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Response: Hominid evolution from *Australopithecus* to *Homo habilis* to *Homo erectus* occurred in Africa. Shortly after its emergence somewhat more than 1.8 million years ago (Ma), *H. erectus* spread to other continents. Fossil remains of *H. erectus* from faraway Java and the Caucasus have been dated to between 1.6 and 1.8 Ma. The transition from *H. erectus* to *H. sapiens* occurred around 400,000 years ago, and the origin of anatomically modern humans somewhat before 100,000 years ago.

The origin of modern humans is debated. Some scientists argue that it occurred in Africa, whence they spread throughout the world, there replacing any preexisting humans. Proponents of the multi-regional model argue, instead, that the transition from *H. erectus* to modern humans occurred consonantly in various parts of the Old World. This explanation postulates "persistent migrations and interbreeding between populations from different continents, of which no direct evidence exists," as I wrote in my article (*1*). Templeton disagrees with my "no direct evidence" statement. He then argues that his statistical analysis of published mtDNA data demonstrates that some regional expansions occurred, such as an expansion across Europe, "although no intercontinental population expansions were detected. . . . " But this conclusion of Templeton's (no intercontinental expansions) appears, in fact, to support my point (no direct evidence of intercontinental migration).

In any case, Templeton's mtDNA analyses are largely irrelevant to the issue at hand. What is at stake is the continuity

from early dispersed *H. erectus*, dated 1.6 to 1.8 Ma, to anatomically modern *Homo sapiens*. The mtDNA data embrace much more recent times, the last 200,000 years in the prevailing view.

In my article (*1*), I quoted the conclusion of Goldstein *et al.*, derived from the analysis of 30 DNA polymorphisms, that the deepest split separating African from non-African populations occurred 156,000 years ago (*2*). But I also quoted estimates for the origin of anatomically modern humans, derived from mtDNA, of 200,000, 143,000, 298,000, and 622,000 to 889,000 years ago; and estimates derived from Y chromosome studies of 270,000, 168,000, and 37,000 to 49,000 years ago. I did not favor any particular date, but rather pointed out that the discrepancies underscore "the need for more extensive and accurate data" (*1*).

I do not find it surprising that disparate estimates exist for the origin of anatomically modern humans or for the split between African and non-African populations. The estimates depend on many uncertainties, including the assumption that rates of molecular evolution are constant and that we know precisely enough what rate to apply in each particular case. Rather, what I find surprising is the "assurance with which some molecular evolutionists assert the precise dates they infer from their analyses." (*3*).

Templeton states that he knows of "no evidence for a split" between African and non-African populations. But there is plenty of evidence. Cavalli-Sforza *et al.* (*4*), for example, have analyzed 120 genes in 42 populations broadly representative of the world, and shown a deep split between African and non-African populations. The split is statistically robust, present in about 83% of bootstrap replications. When the 42 populations are collapsed into nine compact clusters, the bootstrap value rises to 98%. Perhaps Templeton would argue that this split and other evidence is not conclusive of the African replacement hypothesis (because the split can also be explained by models that assume restricted gene flow between populations); this happens to be my view as well. The weight of evidence, I wrote, favors a recent African origin for modern humans, but the replacement may not have been complete everywhere. Many uncertainties remain, so that only the future will "provide more definitive and precise answers" (*1*).

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