

PLURAL LINEAGES IN HUMAN MTDNA GENOME

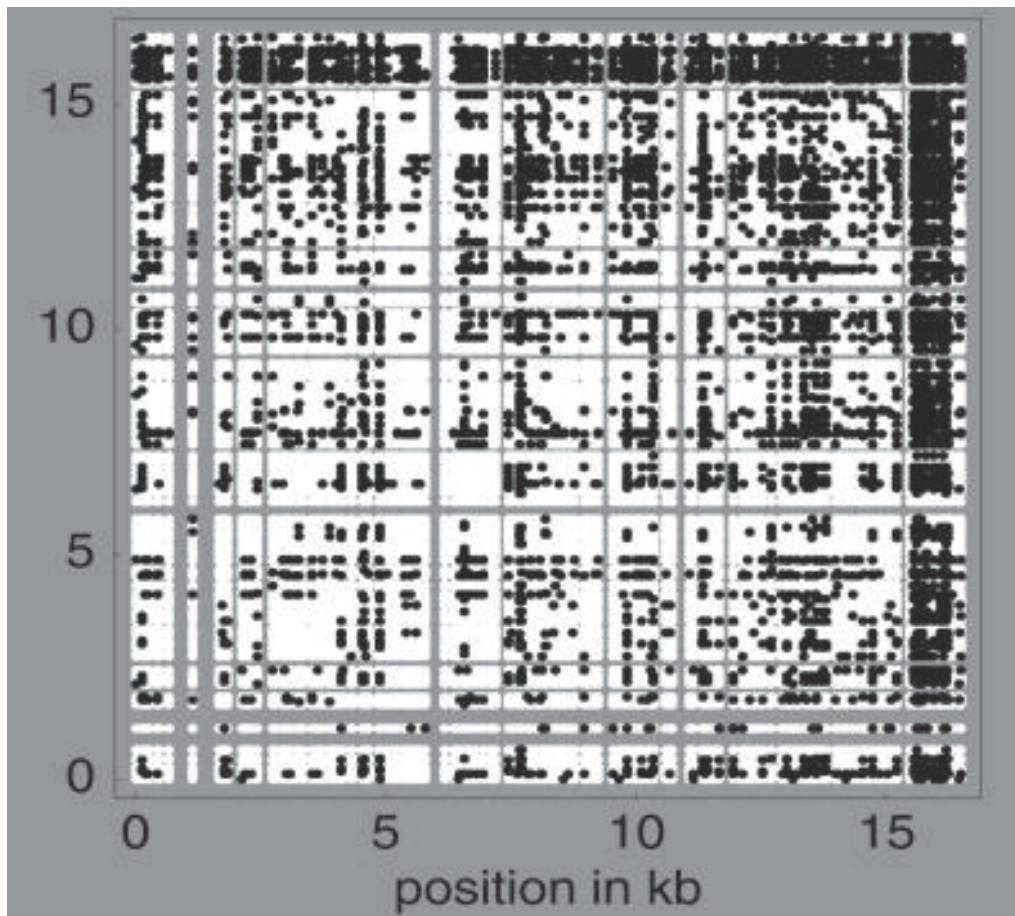
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There has been unanimous and vociferous insistence in the media on one critical Afrocentrist assumption: All modern human *mtDNA* is “so similar” that it had to come from a single woman. This is known to the public as the “African Eve” theory, and researchers speak of the same idea as “a single genealogy for the *mtDNA* genome.” Far from proving the out-of-Africa theory, this is an absolutely *necessary* pre-condition for their theory of Afro-replacement. However, recent research contradicts the claim that all human *mtDNA* is “so similar” it must come from one woman.

Consider the following sentence (italic emphasis mine) from the first paragraph of “Recombination or Mutational Hot Spots in Human *mtDNA*?” (Innan and Nordborg, 2002): “The argument for *recombination* is based on the observation that the pattern of polymorphism in *mtDNA* is incompatible with a *single genealogical tree* and *unique mutations*.”⁴¹ Thus, there are three things that might account for the observed pattern of polymorphisms: first, recombination; second, more than one lineage; or third, multiple mutations at *many* sites. Recombination, as the term applies to *mtDNA* replication, refers to the uncommon occurrence of *mtDNA* from the sperm becoming incorporated in the developing embryo. It was previously assumed that *only* the mother’s *mtDNA* was ever transmitted. For the purposes of this discussion it isn’t necessary to even understand what recombination in *mtDNA* means, let alone the details of how it is accomplished: We can simply treat it as an “object” in this analysis.

Look at figure two from the Innan paper, and you will see that the possibly recombined or repetitively mutated sites are scattered throughout the genome, and do not occur only in the hypervariable region. Note that the quote from Innan & Nordborg implies that the *less* recombination or repetitive, same-site mutation has affected the mitochondrial genome, then the *less* likely it is that there is a *single* genealogy for human *mtDNA*. In other words, unless there has been enough repetitive mutation or recombination to account for the observed pattern of polymorphism, then not all *mtDNA* is from the same source, despite the claims of proponents of the “African Eve,” or African radiation and replacement, theories.

FIG. 2 Evidence for recurrent mutation or recombination (or both) in human *mtDNA* (data of Ingman et al. 2000).² Each point represents the comparison between a pair of polymorphic sites. The point is black if the pattern of polymorphism for the pair of loci is such that either recombination must have occurred between the loci or recurrent mutation affected at least one of the loci. The point is white otherwise. If recombination has occurred, and (importantly) the probability of recombination increases with distance between sites, white points are expected to be clustered along the diagonal (because recombination is less likely to have affected closely linked sites). Recurrent mutations, on the other hand, might be expected to give rise to a pattern that does not depend on the distance from the diagonal, leading to black “crosses” against a white background. The D-loop is visible as a cluster of such crosses in the upper right corner (position 0 corresponds to the first position after the D-loop).



On the other hand, the more recombination, or repetitive mutation, that has occurred, then the more Eve's age has been underestimated. That is true because the effect of either repeated mutations at one site or recombination is to make the *mtDNA* genome appear younger than it really is. If we knew Eve's era from historical or anthropological data, we could compare that date with the one derived from the *mtDNA* coalescence algorithm. The difference between the calculated coalescence result and the historical date would reveal the combined effect of recombination and repetitive mutation. If that combined effect is obviously insufficient to account for the observed pattern of polymorphism, then we may infer a plural genealogy for the human *mtDNA* genome.

There is nothing in the historical or anthropological record to independently establish the era of a (supposed) speciation of humans in Africa. However, the situation in Eurasia is very different. There we can calibrate the coalescence date of Eurasian strains of *mtDNA* with an historical event, anthropological evidence, and research on the human y-chromosome. Thus we can infer the effect (hence extent) of recombination and repeated mutations by comparing those other dates with the result of the *mtDNA* coalescence calculation. If the fit on all these dates is fairly close we can be assured that little recombination or repetitive mutation has occurred, hence the observed pattern of polymorphism must be interpreted to reveal a plural genealogy for the *mtDNA* genome.

In "Natural Selection Shaped Regional *mtDNA* Variations in Humans" (Mishmar et al., 2003) macrohaplogroups (*mtDNA* lineages) M and N are discussed. Technically, one can't say that these are "Eurasian specific" lineages, only because they have also found their way into the African population. The authors admit that the Afrocentrists' attempts to explain the radiation and distribution of these macrohaplogroups is not convincing. Mishmar et al suggest that natural selection for cold-adaptation can explain the variation in lineages derived from M and N. However, as they admit, it is rather implausible that *only* these two haplogroups radiated from Africa, when all are present in NE Africa.³ To sum up: In the Afro-view, M and N lineages diverged from the African Eve's lineage, while in my view they are the oldest surviving Eurasian lineages. Either way, it doesn't affect the argument I am making in regard to determining the relative contribution of repeated mutation and/or recombination versus plural genealogy.

Mishmar calculates the M and N lineages are *both* 65,000 years old. I regard it as significant that these Eurasian Eves' ages are the same, as will be explained below. It doesn't matter for this part of the argument whether you think M and N split off the African lineage then, or whether you think they are the most ancient, indigenous Eurasian lineages still extant: either way, they are 65 kyr old, as calculated by the coalescence algorithm.

There was a lot going on in Eurasia around -65 kyr. The last common *paternal* ancestor of Europeans lived at -59 kyr, as calculated from y-chromosome data. So we have a date for the male counterpart of Eurasian Eve, *calculated from a different*

genome at a date within 10 percent of Eve's age. Then, true-human artifacts are found in Eurasia by around -50 kyr. Allowing for the fact that it is unlikely that we have found the very first artifacts, and that people may have been genetically modern for a while before developing human (*Homo sapiens sapiens*) culture, those dates are in remarkable agreement. Add to this that radiation into Australia by anatomically modern humans occurred at a date that may be as early as -60 kyr, and no doubt that radiation took some millennia.

It begins to look like that -65 kyr coalescence date is right on target, and we could claim there is *no* influence from recombination and repeated mutation, so all the sites reflected in figure two are evidence for plural genealogies: QED! However, that would be disingenuous, because I believe there has been *some* recurrent mutation, though it is possible Innan and Nordborg have fully accounted for it in their model, and *some* recombination, which they have *not* explicitly factored in. So, I would expect that coalescence date to be a little more recent than the era of a genetically significant event which actually caused modern humans to differentiate from a relatively advanced population of Eurasian archaic sapiens.

One reason we can expect the loss of *mtDNA* lineages existing before the modern type differentiated is that there must have been a population "bottleneck" associated with speciation itself. Moreover, there must have been a severe population loss in temperate or higher latitudes when Mt. Toba erupted around -74 kyr and caused a nuclear winter in Eurasia. That savage selection event alone would account for the loss of more-ancient Eurasian *mtDNA* lineages, especially if it was followed by the constriction attendant to speciation. It is significant that the two oldest Eurasian *mtDNA* lineages are the same age. The fact that they both date from the same era makes it most plausible that more-ancient lineages were lost in a specific selection event and/or population bottleneck rather than through "lineage sorting." It is comparatively unlikely that two *mtDNA* lineages would *simultaneously* diverge from a putative African lineage, and *both* (but only they) migrate from Africa and survive to the present. It is far more reasonable and parsimonious to assume that no more-ancient Eurasian lineages survived two severe bottlenecks, and subsequent lineage sorting, in the indigenous Eurasian population.

So, even if we assume that the actual population constriction occurred prior to the coalescence date, and associate it with the obvious selection event of Toba's eruption, the -65kyr coalescence date still calibrates quite closely. If we compare the dates, we note that -65 kyr is only about 12 percent less than the putative genetically significant date of -74 kyr. So we can see that both recombination and repeated mutations can only have had a small effect on the calculation of a coalescence date, and hence there are not many sites in the *mtDNA* genome that have experienced recombination or repeated mutation. But look at figure two, where many of the sites show evidence of either recombination or repeated mutation, or else of more than one genealogy for the *mtDNA* genome! Therefore most of the sites graphed in figure two *must be*

considered as evidence for more than one *mtDNA* genealogy. Therefore, M and N lineages are *not* derived from the African genome, but represent the most ancient, surviving lineages of the Eurasian type. Hence M and N are Eurasian specific lineages that only entered Africa through radiation, rather than coming from Africa.

In conclusion, recombination and repetitive mutations are not enough (by a wide margin) to explain the observed pattern of polymorphisms in the *mtDNA* genome. Therefore, there is more than one genealogy: There are two Eurasian maternal lineages, associated with the speciation of modern humans (*Homo sapiens sapiens*) in Eurasia, and another, African lineage. This falsifies the "Afro-radiation and replacement" theory, and the politically correct shibboleth that "we are all Africans." As explained in "Age and Origin of the Human Species" (posted at rafonda.com) the evidence already pointed to a recent, Eurasian origin for modern humans.⁴ Only the assertion of a single genealogy for *mtDNA* supported the African replacement theory, and that is now refuted.

Mishmar et al. are probably correct to attribute variation, in lineages derived from M and N, to natural selection. One supposes they theorize that *mtDNA* selection took place in the last 50 kyr (Wallace's date for an African radiation, quoted in Wade, "Ice Age Ancestry")⁵ in order to accommodate their theory to the constraints of assuming an African origin for modern humans. However, it is more plausible that such selection took place in very ancient times (when prehuman species were adapting to a cold climate) and was only retained at *high latitudes*, among people living at low culture levels, as the adaptations come at a fitness cost, and thus were lost when and where physical adaptations were superseded by elaborated clothing and shelter, in the temperate zone and the recent era.

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ENDNOTES

1. Hideki Innan and Magnus Nordborg, "Recombination or Mutational Hot Spots in Human *mtDNA*?" *Molecular Biology and Evolution*, 19 (7), 2002, 1122-1127.
2. Fig. 2 in Innan created from data in M.H. Ingman, et al, "Mitochondrial Genomic Variation and the Origin of Modern Humans," *Nature*, 408, 2000, 708-713.
3. Dan Mishmar et al., "Natural Selection Shaped Regional *mtDNA* Variation in Humans," *PNAS*, 100 (1), January 7, 2003, 171-176. www.pnas.org/cgi/doi/10.1073/pnas.0136972100.
4. Ronald Alan Fonda, "Age and Origin of the Human Species," *Mankind Quarterly*, XLII (2), Winter 2001, 189-199.
5. Nicholas Wade, "Ice Age Ancestry May Keep Body Warmer and Healthier," *New York Times*, January 9, 2004, 16.