Human evolution

Out of the garden of Eden

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A PAPER by R. L. Cann, M. Stoneking and A. C. Wilson on page 31 of this issue[1] reports that Eve was alive, well and probably living in Africa around 200,000 years ago. In considering this striking claim we should bear in mind the words of Thomas Hood in the first stanza of 'A Black Job':

> The history of human-kind to trace
> Since Eve — the first of dupes — our doom unriddled,
> A certain portion of the human race
> Has certainly a taste for being diddled.

Cann *et al.* base their proposed identification of Eve on a detailed analysis of the mitochondrial DNA types in a diverse group of human populations. Human mitochondrial DNA, a circular molecule of about 16,500 base pairs[2,3], has a strictly maternal inheritance pattern. Thus, the mitochondrial DNA type of an individual is inherited from the mother, the maternal grandmother and so on.

Mitochondrial DNA evolves rapidly with a mutation rate of up to ten times higher than that of nuclear DNA[4], giving rise to much variation between the mitochondrial DNA sequences of different individuals[5]. Cann *et al.* have determined some of the differences in mitochondrial DNA sequences from 147 individuals in 5 populations by high-resolution mapping with 12 restriction enzymes. They find 133 different DNA types and present an evolutionary tree that relates these types both to each other and to a derived ancestral mitochondrial DNA type.

The mitochondrial DNA tree has two primary roots of descent, the first leading exclusively to Africans and the second to some Africans and to all other population groups. This tree is consistent with limited nuclear DNA data that suggest[6] an early division of populations between an African group and a Caucasian–Mongoloid group. A more difficult problem is the question of which population group is the ancestral one. The study of Cann *et al.*[1] represents the strongest molecular evidence so far in favour of the African population being ancestral. The two supporting lines of evidence are the evolutionary tree itself, which clearly suggests an African origin, and the fact that Africans seem to have more mitochondrial DNA diversity than other populations (the population which has been around longest would be expected to have more DNA sequence changes). It seems likely that modern man emerged in Africa and, as discussed in a previous News and Views article[7], that subsequently a founder population left Africa and spread throughout Europe, Asia and the Americas. Further studies of the mitochondrial DNA types of African populations would be valuable to provide corrobatory evidence for this 'Out of Africa' hypothesis, based by Cann *et al.* largely on studies of black Americans.

As an extension of the analysis, Cann *et al.* conclude that the mitochondrial DNAs stem from a single woman. This conclusion is not dependent on the data presented but rather is the consequence of the method of analysis. It assumes that the heterogeneity of mitochondrial DNA at the time of ancestral type was no less than in present-day populations, otherwise the present-day mitochondrial DNA types might well have descended from a large number of women who were monomorphic for a particular DNA type.

The case against the 'mother Eve' hypothesis has now been put by Latorre and colleagues, who have just published a fascinating study[8] of the mitochondrial DNA types of the fruitfly *Drosophila subobscura*, which has recently made a dramatic expansion into the New World. Latorre *et al.* point out that in a few thousand years time all the *D. subobscura* flies may have mitochondrial DNAs derived from the type they call morph I. Clearly this does not mean that mitochondrial DNA of the descendants derives from only one *D. subobscura* currently living, as morph I is found in 44 per cent of the present-day population.

The high mutation rate of mitochondrial DNA (compared with nuclear DNA) and its inability to undergo recombinations are undoubted bonuses for the population geneticist bent on tracing ancestral lines. Nevertheless it should not be forgotten that — except for the tiny mitochondrion — the nuclear genome comprises all of our genetic makeup and stores many secrets of our murky past. For example, we inherit our mitochondrial DNA from just one of our sixteen great-great grandparents, yet this maternal ancestor has only contributed one-sixteenth of our nuclear DNA. The recently identified common female ancestor should more correctly be recognized simply as our 'mitochondrial Eve' as she has contributed little, if anything, to our nuclear DNA. Clearly, evolutionary studies of nuclear and mitochondrial DNA are complementary, although in certain circumstances they can give divergent results depending on the breeding pattern of the species.

Cann *et al.* conclude that mitochondrial Eve lived between 140,000 and 290,000 years ago. This calculation is based on an assumption of 2–4 per cent divergence in mitochondrial DNA sequence per million years. The dating is obviously rather crude but it is tempting to relate the occurrence of the ancestral mitochondrial DNA type back to a severe constriction in population size (bottleneck). If this assumption is correct, the timing of such bottlenecks may correlate with major evolutionary events. It would be interesting to see where a more precise dating puts the ancestral mitochondrial DNA type in relation to our own evolution.

But there is an alternative explanation for the data, which is that one ancestral mitochondrial DNA type has reached fixation by random genetic drift. This is another point emphasized by Latorre *et al.*[8] in their discussion of a population of 15,000 unrelated females which apparently has a probability of 50 per cent that the mitochondrial DNA of all individuals living 18,000 generations later will have derived from a single female[9]. This is equivalent to about 360,000 years for humans. Nevertheless, it still seems possible that the date of the mitochondrial Eve is associated with the most recent population bottleneck.

Again, these types of calculation show that many Eves have contributed to our nuclear DNA. So far, most evolutionary studies of nuclear DNA have been concerned with restriction fragment length polymorphisms and sequence comparisons from autosomal loci. However, work has now commenced on the construction of our paternal ancestry with the use of Y-chromosome-specific probes which might enable a common paternal ancestor to be identified[10]. It could turn out that Adam and Eve were contemporaries after all, in which case they might even have met.

Where does this leave molecular biology as a tool for studying human evolution? There are now powerful methods for studying population affinities and prehistoric migrations, but it is less clear how we can estimate the size, date and place of bottlenecks and their significance in evolution. The combined use of mitochondrial DNA, autosomal nuclear DNA and Y-chromosome-specific DNA markers has enormous potential for the genetic analysis of human populations. □

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