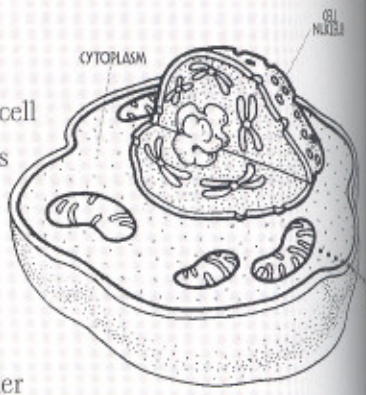
Mitochondrial DNA



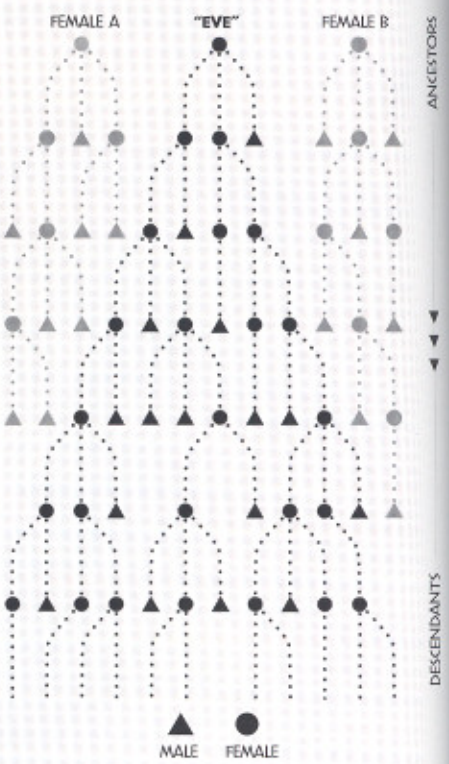
THE CELL

Mitochondrial DNA (mtDNA) is a small length of DNA found in the mitochondria, cell structures outside the nucleus that are involved in the cell's energy production. It is of particular interest to anthropologists and biologists because it is inherited only through the mother. This happens because when the sperm fertilizes the egg, only the nuclear DNA of the sperm is injected into the egg. The rest of the sperm, including its mtDNA, remains outside the egg and is eventually absorbed by the mother's tissue. Your mtDNA came only from your mother, who obtained it from her mother, and so on. This property of mtDNA allows patterns of genetic relationship to be studied without the complication of genetic recombination that occurs every generation for the rest of your DNA (nuclear DNA).

DNA analysis has been used to look at the genetics of ancient humans dating back many tens of thousands of years. The question of the origin of anatomically modern humans has been studied not only with the fossils that have been found, but also with mtDNA of modern groups of people. Mitochondrial DNA is obtained from blood samples collected from geographic populations of modern humans around the world and the differences and similarities of the sequences of the bases (DNA building blocks) are compared among the groups.

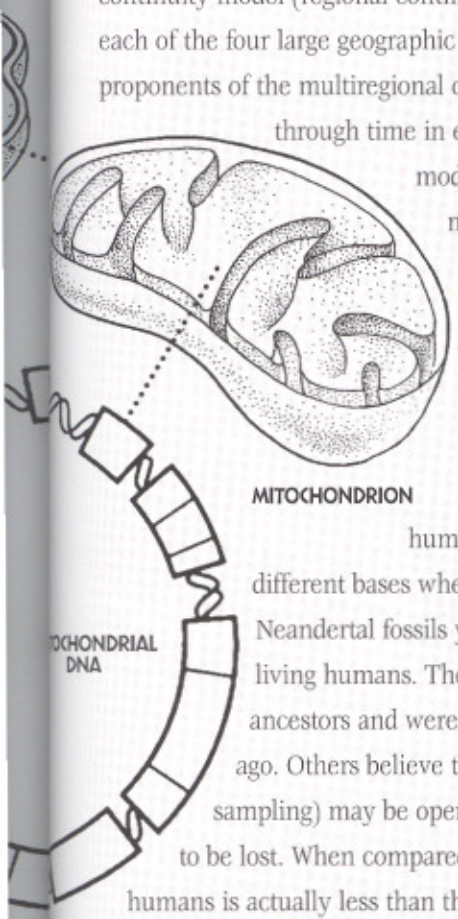
Studies of mtDNA show that the world's populations today form two basic clusters. One cluster consists of people with African ancestry, and the other cluster contains people of different ancestries, both African and non-African. Additional studies use genetic trees to estimate time of divergence—the time when two different populations split from an ancestral population. The degree of variation among the Africans is greater than that within the second group, which indicates that the African group has been established longer. One conclusion from this work is that the original female ancestral lineage was African. These findings have been interpreted as reflecting population history: an initial origin of modern humans in Africa followed by a subsequent split of non-African populations. An estimate of the time of origin of anatomically modern humans can be made with these genetic trees. The common mitochondrial DNA ancestral lineage of all living humans arose roughly 200,000 years ago in Africa.

Not all paleoanthropologists agree that the time, 200,000 years ago, and the place, Africa, are correct. The African replacement model (complete replacement model), as this interpretation is called, had been criticized for its sampling techniques, not taking into consideration the presence of population bottlenecks and the possibility of irregularity of the mtDNA clock. A competing model, called the multiregional



This mitochondrial "tree" shows how one female's mtDNA will be inherited by all her female descendants. Females A and B have no female descendants after the 4th and 5th generations respectively.

continuity model (regional continuity model), says that *Homo erectus* evolved into modern *Homo sapiens* in each of the four large geographic areas of Africa, Asia, Europe, and Australasia. Besides the above criticisms, proponents of the multiregional continuity model emphasize anatomical similarities of the fossils found through time in each of the areas. The mtDNA analyses do not rule out the multiregional model. However, the African replacement model is not only supported by the mtDNA data, but the majority of the nuclear DNA and Y chromosome data add credence to an African origin of modern humans.



Can mtDNA distinguish Neandertal from anatomically modern humans (*Homo sapiens*)? Mitochondrial DNA has been extracted from three Neandertal fossils and their DNA sequences are quite different from those of living humans. In the analysis of the first fossil, 27 differences were found between the Neandertal sample of 378 DNA bases and a living human sample of the same number of bases. This compares to an average of 8 different bases when one looks at the variation between pairs of living people. The other two Neandertal fossils yield essentially the same result—genetic differences are high relative to living humans. These studies have led some to conclude that the Neandertals are not our ancestors and were likely a separate species that became extinct in Europe about 28,000 years ago. Others believe that the situation is not that simple. Factors such as genetic drift (chance sampling) may be operating which would cause certain Neandertal and/or modern human lineages to be lost. When compared to nonhuman primates, the difference between Neandertals and living humans is actually less than that found in some of the comparisons of chimpanzee subspecies. This would lead you to think Neandertal might be a different subspecies and still be among our ancestors. Clearly, more Neandertal fossils need to be sampled.

As it has been discussed above, mtDNA has contributed much to the understanding of our past. Analyses of mtDNA of living populations have also been an exciting source of data that has been used to reconstruct past migrations of various groups of the world. Global surveys of human mtDNA variation have led to valuable new insights into the relationships of various groups in different parts of the world. For example, genetic diversity and distance studies using mtDNA have described structure and history of populations in the Pacific; have introduced new ideas about the peopling of the New World; and have produced one of the most complex genetic pictures of the continent of Africa that has ever been seen. These data supplement and complement whatever archaeological or historical information is available. In some cases, they introduce completely novel ways of thinking about our recent past. New methodology and additional surveys will produce even more interesting ways of looking at ourselves and our ancestors.

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