

# History-hunting geneticists can still follow familiar trail

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As the world's first explorers branched away from humanity's birthplace in east Africa some 65,000 years ago, distinct mutations accumulated in the DNA of each population, essentially providing a genetic trail for modern researchers to follow.

Recently some scientists have raised doubts about this classic genetic system to study ancient migrations of people and to estimate the populations of people or animals as they existed tens of thousands of years ago.

But University of Florida researchers writing this month in an online edition of *Science* validate the approach, which involves tracking sequences of mitochondrial DNA, also known as mtDNA.

"The study of mtDNA has helped to demonstrate the African origin of our species and the relationship between living humans and the Neanderthals," said Connie Mulligan, an associate professor of anthropology in the College of Liberal Arts and Sciences and an assistant director of the UF Genetics Institute. "MtDNA data have also been used to establish the time and route of major events in human history, such as the expansion of Neolithic farmers into Europe, and the settlement of the New World."

MtDNA has made headlines recently because of initiatives such as the National Geographic Project, a multimillion-dollar endeavor to reconstruct humanity's ancient migrations, and because of well-

publicized efforts to track the ancestral roots of Oprah Winfrey and other personalities.

Located within the hundreds of energy-producing mitochondria that lie outside the nucleus of our cells, mtDNA is unlike the DNA inside the nucleus of a cell that contains genes from both of our parents -- in people and animals mtDNA is exclusively passed from mothers to their children.

For humans, this means that all of the mtDNA in our cells are copies of our mothers' mtDNA, which in turn were copies of their mothers' mtDNA. In this way, mtDNA progresses through the ages, springing from what many scientists believe was a common ancestral mother.

But over the eons, random mutations enter the genetic code of all species. By tracking similarities and differences of mtDNA, scientists gain insight about the size of groups and how they moved around the world.

"When you look at ancient migration, you're always asking the question, 'How big was the population, how many were there?'" said Michael Miyamoto, a professor and associate chairman of zoology in UF's College of Liberal Arts and Sciences. "The field has worked from the premise that the more mtDNA variation you saw, the larger the population was that carried that variation, just like there would be a greater diversity of T-shirts or shoes within a larger population than a smaller population."

However, the connection between mtDNA and population size was questioned this year when French scientists analyzed vast groups of gene sequences from more than 3,000 animal species. They speculated that an evolutionary tendency for species to keep helpful genes and sift out detrimental ones, called "natural selection," preferentially affects

mitochondrial diversity, making mtDNA less useful for population size estimates.

"From a conservation perspective, when scientists look at census counts of animals and how the population size may be increasing or decreasing, the study of mtDNA tells us about the level of genetic diversity in a population, which is important in making conservation decisions on endangered species," Mulligan said. "If this approach were not credible, it could potentially have a bearing on future policy decisions, as well as affect literally hundreds of previous studies on humans and other mammals."

UF Genetics Institute scientists analyzed publicly available mtDNA datasets of 47 species of mammals -- a subset of the animals that were in the French study -- as well as associated data on protein diversity in the same species. A greater variety of proteins indicates more diverse DNA, because DNA contains a species' blueprints for manufacturing protein -- and the French scientists agreed that protein diversity did correlate with population size.

All that remained for UF researchers to do to reinstate mtDNA diversity as indicative of population size was to determine that protein diversity and mtDNA diversity were correlated.

"The researchers showed a correlation between mitochondrial DNA and genetic variation in a way that has never been done before," said Marc Allard, an associate professor of biology at George Washington University who was not involved in the study. "Population geneticists have used mitochondria for all kinds of work for 20 years, and to think that mtDNA didn't correlate with population size was clearly going against the dogma. This study shows the dogma is safe in mammals and probably in vertebrates, as well."

Source: University of Florida

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