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Signs of Neanderthals Mating With Humans

By NICHOLAS WADE

Neanderthals mated with some modern humans after all and left their imprint in the human genome, a team of biologists has reported in the first detailed analysis of the Neanderthal genetic sequence.

The biologists, led by Svante Paabo of the Max Planck Institute for Evolutionary Anthropology in Leipzig, Germany, have been slowly reconstructing the genome of Neanderthals, the stocky hunters that dominated Europe until 30,000 years ago, by extracting the fragments of DNA that still exist in their fossil bones. Just last year, when the biologists first announced that they had decoded the Neanderthal genome, they reported no significant evidence of interbreeding.

Scientists say they have recovered 60 percent of the genome so far and hope to complete it. By comparing that genome with those of various present day humans, the team concluded that about 1 percent to 4 percent of the genome of non-Africans today is derived from Neanderthals. But the Neanderthal DNA does not seem to have played a great role in human evolution, they said.

Experts believe that the Neanderthal genome sequence will be of extraordinary importance in understanding human evolutionary history since the two species split some 600,000 years ago.

So far, the team has identified only about 100 genes — surprisingly few — that have contributed to the evolution of modern humans since the split. The nature of the genes in humans that differ from those of Neanderthals is of particular interest because they bear on what it means to be human, or at least not Neanderthal. Some of the genes seem to be
involved in cognitive function and others in bone structure.

“Seven years ago, I really thought that it would remain impossible in my lifetime to sequence the whole Neanderthal genome,” Dr. Paabo said at a news conference. But the Leipzig team’s second conclusion, that there was probably interbreeding between Neanderthals and modern humans before Europeans and Asians split, is being met with reserve by some archaeologists.

A degree of interbreeding between modern humans and Neanderthals in Europe would not be greatly surprising given that the species overlapped there from 44,000 years ago when modern humans first entered Europe to 30,000 years ago when the last Neanderthals fell extinct. Archaeologists have been debating for years whether the fossil record shows evidence of individuals with mixed features.

But the new analysis, which is based solely on genetics and statistical calculations, is more difficult to match with the archaeological record. The Leipzig scientists assert that the interbreeding did not occur in Europe but in the Middle East and at a much earlier period, some 100,000 to 60,000 years ago, before the modern human populations of Europe and East Asia split. There is much less archaeological evidence for an overlap between modern humans and Neanderthals at this time and place.

Dr. Paabo has pioneered the extraction and analysis of ancient DNA from fossil bones, overcoming daunting obstacles over the last 13 years in his pursuit of the Neanderthal genome. Perhaps the most serious is that most Neanderthal bones are extensively contaminated with modern human DNA, which is highly similar to Neanderthal DNA. The DNA he has analyzed comes from three small bones from the Vindija cave in Croatia.

“This is a fabulous achievement,” said Ian Tattersall, a paleontologist at the American Museum of Natural History in New York, referring to the draft Neanderthal genome that Dr. Paabo’s team describes in Thursday’s issue of Science.

But he and other archaeologists questioned some of the interpretations put forward by Dr. Paabo and his chief colleagues, Richard E. Green of the Leipzig institute, and David Reich of Harvard Medical School. Geneticists have been making increasingly valuable contributions to human prehistory, but their work depends heavily on complex mathematical statistics that make their arguments hard to follow. And the statistical insights, however informative, do not have the solidity of an archaeological fact.

“This is probably not the authors’ last word, and they are obviously groping to explain what
they have found,” Dr. Tattersall said.

Richard Klein, a paleontologist at Stanford, said the authors’ theory of an early interbreeding episode did not seem to have taken full account of the archaeological background. “They are basically saying, ‘Here are our data, you have to accept it.’ But the little part I can judge seems to me to be problematic, so I have to worry about the rest,” he said.

In an earlier report on the Neanderthal genome, the reported DNA sequences were found by other geneticists to be extensively contaminated with human DNA. Dr. Paabo’s group has taken extra precautions but it remains to be seen how successful they have been, Dr. Klein said, especially as another group at the Leipzig institute, presumably using the same methods, has obtained results that Dr. Paabo said he could not confirm.

Dr. Paabo said that episode of human-Neanderthal breeding implied by Dr. Reich’s statistics most plausibly occurred “in the Middle East where the first modern humans appear before 100,000 years ago and there were Neanderthals until 60,000 years ago.” According to Dr. Klein, people in Africa expanded their range and reached just Israel during a warm period some 120,000 years ago. They retreated during a cold period some 80,000 years ago and were replaced by Neanderthals. It is not clear whether or not they overlapped with Neanderthals, he said.

These humans, in any case, were not fully modern and they did not expand from Africa, an episode that occurred some 30,000 years later. If there was any interbreeding, the flow of genes should have been both ways, Dr. Klein said, but Dr. Paabo’s group sees evidence for gene flow only from Neanderthals to modern humans.

The Leipzig group’s interbreeding theory would undercut the present belief that all human populations today draw from the same gene pool that existed a mere 50,000 years ago. “What we falsify here is the strong out-of-Africa hypothesis that everyone comes from the same population,” Dr. Paabo said.

In his and Dr. Reich’s view, Neanderthals interbred only with non-Africans, the people who left Africa, which would mean that non-Africans drew from a second gene pool not available to Africans.