

# NEWS RELEASE

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Oct. 26, 2005

## NC State Researcher Participates in Mapping of Human Genome

### FOR IMMEDIATE RELEASE

A team of international scientists that includes a researcher from North Carolina State University has succeeded in creating a “map” of the human genome that will help scientists find the genetic causes of common diseases like diabetes and Alzheimer’s.

Dr. Bruce Weir, William Neal Reynolds Distinguished Professor of Statistics and Genetics and director of NC State’s Bioinformatics Research Center, is one of more than 60 scientists from around the world involved in the international effort to create a haplotype map of the human genome – a map that pinpoints genetic differences between people.

The researchers’ findings appear in the Oct. 27 issue of the journal *Nature*.



Dr. Bruce Weir

A haplotype is a short piece of a chromosome. Human DNA contains 23 chromosomes, and these chromosomes are almost identical from person to person. However, there are places along the genome – the genetic content within these chromosomes – where variations occur. Scientists refer to these positions along the genome as SNPs (single nucleotide polymorphisms) or “snips.”

The aim of the haplotype map, or “HapMap,” is to provide scientists and medical researchers with “addresses” along the map that will show them where these genetic variations occur.

“Most of our diseases have a genetic component,” Weir says. “We need to find out what these genes are, and to do that we first must discover where they are. The HapMap basically identifies landmarks along the chromosomes so that researchers can eventually find the genes responsible for diseases like Alzheimer’s, diabetes and others.”

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These results represent the end of the first phase of the International HapMap Project, a study of the genetic constitution of 269 people of varying ethnicity: 90 people of European descent, 90 members of the Yoruba tribe in Nigeria, 45 Chinese residents of Beijing and 44 Japanese residents of Tokyo.

A partnership of scientists from Canada, China, Japan, Nigeria, the United Kingdom and the United States began the project in 2002. The researchers' findings are available to the public via their Web site: *www.hapmap.org*.

As a statistician, Weir's role in the HapMap project was to help make sense of the raw data. "Basically, we had all these numbers and letters in a giant computer file of data," he says. "Our team needed to figure out how this data should best be organized in order to help other scientists and researchers use it, and then to do the organizing.

"Phase I of the HapMap project identified 1 million SNPs," Weir says. "We believe that there are 10 million total positions along the genome where variations occur, but that identifying a fraction of them should be sufficient for our purposes. One of the HapMap goals is to identify that fraction."

Phase II of the HapMap project should be finished in the near future. There are also plans to extend the study to other world populations. Weir is excited about the future ramifications of the project.

"This is really big science," he says. "Sixty scientists from around the world working toward a common goal that will have a huge impact on mankind."

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**Note to editors:** An abstract of the *Nature* paper follows.

## "A Haplotype Map of the Human Genome"

*Authors:* The International HapMap Consortium, which includes Bruce Weir, North Carolina State University

*Published:* Oct. 27, 2005, in *Nature*

**Abstract:** Inherited genetic variation plays a critical but as yet largely uncharacterised role in human disease. We report a public database of common variation in the human genome: more than one million single nucleotide polymorphisms (SNPs) for which accurate and complete genotypes have been obtained in 269 DNA samples from four populations, including ten 500 kb regions in which essentially all information about common DNA variation has been extracted. These data document the generality of recombination hotspots, a block-like structure of linkage disequilibrium, and low haplotype diversity, leading to substantial correlations of SNPs with many of their neighbours. We show how the HapMap resource can guide the design and analysis of genetic association studies, shed light on structural variation and recombination, and identify loci that may have been subject to natural selection during human evolution.