Gene researchers find more clues to human individuality

By Stefanie Olsen, CNET News.com
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Scientists at Emory University have developed a new type of map of the human genome that could one day lead to breakthroughs in personalized medicine.

By mapping the genome--the Human Genome Project completed in 2003--scientists have already shown that people share the same basic genetic information. As much as 99 percent of human code is identical--it's the last 1 percent that determines individual traits like difference in appearance, life expectancy, resistance to drugs and susceptibility to disease.

On Thursday, a team of scientists in Emory's School of Medicine said that they've advanced the study of the last 1 percent of genetic variation by producing a map of more than 400,000 insertions and deletions (INDELs) in the human genome. In simpler terms, they've shown 400,000 different naturally occurring variations in genetic code, or so-called polymorphisms, which can help explain idiosyncrasies in humans.

"That's where all this genetic research is headed--variation research to be able to predict someone's future health based on their genetic sequence," said Scott Devine, an assistant professor of biochemistry at Emory who led a team of seven scientists in the research.

"Imagine 10 years from now we can resequence your genome to show that the one gene will vary and predict that you could have heart disease when you're 40 years old. Doctors could then tailor treatments to that person depending on their genetics."

Several years ago, an international team of researchers called the HapMap Consortium produced the first map of natural genetic variations called single nucleotide polymorphisms (SNPs), of which there were more than 2 million in the human population.

SNPs are single variations in the base pairs of chemical building blocks of humans--adenine, thymine, cytosine and guanine (A, T, C, G). (Human DNA contains 3 billion base pairs of A, T, C, G, which are strung together in different combinations in long chains within 23 pairs of chromosomes.)

To draw an analogy, SNPs are like changing one letter in those building blocks. So an SNP might be A, A, C, G.

In contrast, INDELs either insert or delete hundreds of letters, or chemical building blocks. When those changes happen, a human's genetic makeup is altered.

"If the genome is an instruction book, SNPs are places where the instruction book varies by one letter, like a misspelling," Devine said. "INDELs are more like a word or paragraph inserted and deleted."

Devine's team developed computer-based algorithms to locate the INDELs in compiled genetic data on 36 humans, in a research project spanning the last two years.

"For the first time, we characterized these types of variations in human populations and built an initial map of it," Devine said. He believes the map of INDELs will grow to 2 million naturally occurring variations. In contrast, the map of SNPs will likely grow to 10 million.

Of course, scientifically the variations are not correlated to specific traits yet, but that's what scientists like Devine hope the research will lead to. Devine also aims to combine the SNP and INDEL maps so that scientists have a broader perspective to make correlations on individual variations.

Emory's research has been posted online and will appear in the September issue of the journal Genome Research.
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