

The Scientist: NewsBlog:

African genomes sequenced

Posted by [Jef Akst](#)

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Scientists have sequenced the genomes of five individuals from indigenous populations in southern Africa, including famed South African Bishop Desmond Tutu, revealing new genetic variation among humans that they say will advance medical genomics research, according to a study published this week in *Nature*.

"It's the first genome sequence of a minority population in Africa," said human geneticist [Sarah Tishkoff](#) of the University of Pennsylvania School of Medicine, who was not involved in the research. "I'm excited to see that somebody has finally sequenced the genome of an underrepresented population."

Previous research has suggested that "there's an extraordinarily high level of variation among indigenous populations in Africa," Tishkoff explained, but to date, no full sequence data were available for these groups. Thus, most of the genome-wide association studies (GWAS) that look for correlations between genetic variants and particular phenotypes or diseases have been based on genetic variation identified in European and Asian populations. "So there's a huge bias when you then look at the African populations," Tishkoff said.



Bushmen of southern Africa
Image: Stephan C. Schuster

Using next generation sequencing technology, [Stephan Schuster](#) of Pennsylvania State University and his colleagues sequenced the complete genomes of an indigenous hunter-gatherer from Namibia's Kalahari Desert and Archbishop Tutu, a Bantu from southern Africa, as well as the coding regions of individuals from three other hunter-gatherer populations. The results confirmed the higher level of variation among these groups, documenting 1.3 million new sequence nucleotide polymorphisms (SNPs) not previously identified in the known Asian and European genomes, or the Nigerian genome [sequenced in 2008](#).

"I think it's quite amazing" to discover this number of new variants in these individuals, Schuster said at a press briefing this morning. These results help researchers to "understand just how broad human genetic variation can be and what the implications are."

Furthermore, "[these] variants could be used to develop new SNP arrays that will cover variation that may be specific to Africa," Tishkoff said, which "would be very informative for doing disease association studies, particularly in southern Africa." Indeed, the authors of the current study are already working on developing new arrays that incorporate the newly discovered genetic differences, and plan to use the tool to study variation across southern Africa, they said in the press conference.

Another interesting aspect of this study is that all of the individuals sequenced were quite old, molecular biologist and engineer [Jonathan Rothberg](#), founder and CEO of Ion Torrent

Systems in Connecticut, wrote in an email to *The Scientist* -- with the exception of the 78-year-old Archbishop, they were all at least 80. "So you know this diversity does not cause any fatal early onset diseases," he said. In addition, with medical history available for the sequenced individuals, we can get "a very good feel for the medical outcome of those [genetic] differences," Schuster said.

This study was limited to only one individual per group, however, Tishkoff noted. "There's only a limited amount we can say based on a single individual." The next step, she added, is to look at many more individuals to identify any additional variation that can be missed when you "just sample one individual out of a population."

"We will have to do as many genomes as cities in Google Earth to really get a complete picture of human history," Rothberg agreed, but this study is a great start, he said. "Future studies will fill in huge groups of individuals and tie the work in to give us a fantastic history of human migration and interactions."

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