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SMART NEWS

Scientists Have Finally Sequenced a ‘Gapless’ Human Genome

Scientists have deciphered the missing eight percent of our genetic blueprint, setting the stage for new discoveries in human evolution and disease

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Filling in genome gaps could help scientists better the genetic basis for certain diseases and lead to new medical discoveries. Kateryna Kon Science Photo Library via Getty Images

Around 20 years ago, researchers completed a draft of the human genome: a sequence of roughly three billion letters. But the blueprint was only partially

complete, missing eight percent of the genes. Now, scientists have finally finished their quest.

“Hallelujah, we finally finished one human genome, but the best is yet to come,” says Evan Eichler, a University of Washington geneticist and one of the leaders of the mapping project, in a news briefing Thursday.

Having a gap-free sequence of a human genome could lead to new medical discoveries and reveal clues about our evolutionary past. The genome mapping effort was a collaboration of nearly 100 researchers from government, academic and private organizations, called the Telomere-to-Telomere consortium. Their work was published in the journal *Science*.

“I am thrilled that we got the job done,” Eichler says in a press release. “The complete blueprint is going to revolutionize the way we think about human genomic variation, disease and evolution.”

Deciphering a genome is a lot like putting together a puzzle. Scientists slice the DNA into pieces ranging from hundreds to thousands of letters long. They then employ the help of machines to read the individual letters in each snippet, which scientists try to put in the right order, according to the *Guardian*’s Hannah Devlin.

When researchers published their first draft of a human genome in the early 2000s, genetic sequencing technology was far more limited. DNA sequencing technology could only produce short fragments of genetic code, about 500 letters at a time, reports Evan Bush for NBC News. Scientists now have access to “long-read” sequencing machines that decode larger slices of genetic material at once, allowing them to fill in missing gaps accounting for roughly 400 million letters.

“In the future, when someone has their genome sequenced, we will be able to identify all of the variants in their DNA and use that information to better guide their healthcare,” says Adam Phillippy of the National Human Genome Research Institute, and co-chair of the consortium, in a press release. “Truly finishing the human genome sequence was like putting on a new pair of glasses. Now that we can clearly see everything, we are one step closer to understanding what it all means.”

Knowing the previously-missing slices of human DNA could help medical professionals pinpoint genetic variations that lead to certain diseases, and allow

scientists better understand how humans evolved particular traits that distinguish us from our great ape ancestors.

Those behind the genome mapping project note that this is “a” human genome, not “the” human genome. The genetic information was derived decades ago from cell tissue that contains only paternal chromosomes—half the genetic information need to make a full human—which means it does not represent a real person that ever lived, according to the *Washington Post’s* Joel Achenbach. While that approach meant scientists only need to sequence only one set of DNA, the consortium next plans to sequence genetic information with both paternal and maternal chromosomes.

The team also hopes to sequence genomes of people from around the world to better understand the full variation of human genetics.

“We’ll start with dozens and then hundreds and ultimately have thousands, so we will better represent the diversity of human genomes,” Eichler tells Ed Cara of *Gizmodo*.

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