
HEALTH

The map of our DNA is finally complete. Here's what that means for humanity.



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USA TODAY

Published 3:14 p.m. ET March 31, 2022 | Updated 10:35 p.m. ET April 10, 2022

Scientists are finally done mapping the human genome, more than two decades after the first draft was completed, researchers announced Thursday. About 8% of genetic material had been impossible to decipher with previous technology.

Completing the final pieces is like adding the continent of Africa to a map of the globe that lacked it, said Michael Schatz, who participated in the research and is a professor of computer science and biology at Johns Hopkins University.

Even missing that 8%, scientists were able to get the gist of the story of human genetics, said Jonas Korf, chief scientific officer of Pacific Biosciences, the company whose technology was used to fill the gaps.

If genetics were a detective story, "precisely the pages where you would find out who the murderer is were missing," he said.

Several teams of American researchers published six papers in the journal *Science* on Thursday that fill the gaps in a single human genome, compare those areas with some of our closest ape relatives and begin to explain the role of those newly described pieces.

It will be years before there's a concrete payoff to that additional information, researchers said, but those previously missing bits could offer insights into human development, aging and diseases such as cancer, as well as human diversity, evolution and migration patterns across prehistory.

"In some ways, these publications might be considered the long-awaited closing ceremony" of the Human Genome Project, which began in 1985, said Dr. Eric Green, director of the National Human Genome Research Institute at the National Institutes of Health.

He and several scientists involved in the Telomere-to-Telomere consortium research held a call with media several hours before the papers were published.

Mapping this genetic material should help explain how humans adapted to and survived infections and plagues, how our bodies clear toxins, how individuals respond differently to drugs, what makes the brain distinctly human and what makes each of us distinct, said Evan Eichler, a geneticist at the University of Washington School of Medicine who helped lead the research.

"In principle, this will allow us to better understand how we form as an individual organism and how we vary, not just between other humans but other species," said Eichler, who had hoped for decades to fill in the gaps. "For me, it's like a dream come true."

Earlier maps, he said, were missing entire chapters of the book of life. Now, "we can continuously read the book with almost no errors," he said, "we can get from Page 1 to the final chapter."

DNA, the blueprint of life, consists of four base pairs of nucleotides, simplified as the letters A, C, T and G. An individual's genome is the complete set of these sequences.

In the initial map, researchers discovered there were about 3 billion of these letter pairs in the human genome. Sections of five chromosomes were missing, mainly areas that contained a lot of repeated genetic letters.

The way earlier mapping technology worked, researchers sequenced short bits, then overlapped them – like piecing together a book from sentence fragments.

In the original Human Genome Project, researchers could map about 500 pairs of letters at a time. Newer technology, led by PacBio, can read up to about 100,000 pairs and detect repetitions.

Reading those longer pieces, Korlach said, allows scientists to "eavesdrop on what happens in nature."

Critical functions are controlled by these repeats, Eichler noted, including genes that enabled the human brain to become bigger with more folds. The repeats are involved in the production of ribosomes, the factories that allow cells to make proteins, transforming the genetic code into action.

Repeats play a role in the centrosomes, the pinched area in the middle of chromosomes that are involved in accurately copying genetic material, as one cell divides into two.

Problems with this process are implicated in diseases such as Down syndrome, a genetic condition in which children are born with an extra chromosome, leading to intellectual and physical challenges and a shortened lifespan.

Repeated sequences differ among species, Korf said. In so-called murder hornets, for instance, 30% to 40% of the genome is made up of repeats while butterflies have hardly any repeats.

Much remains to be explained about their role.

"We're more excited about what we don't know and the opportunity for discovery," said Karen Miga, co-chair of the research team and associate director of the UCSC Genomics Institute at the University of California, Santa Cruz.

Developing this single genome took about four years and cost several million dollars, said Adam Phillippy, co-chair of the consortium that conducted the work and head of the NHGRI Genome Informatics Section.

He hopes within a year to have a fuller version and to map many more human genomes, so scientists can explore human diversity.

It's not clear whether human variation revealed by the work causes disease, but "the fact that there's an entire class of variation that's never been seen before is extremely exciting to me," Schatz said.

George Church, a geneticist at Harvard University who was not involved in the new work, described it as a major scientific achievement.

"It's a huge milestone that should be celebrated," he said.

But, he added, "we probably won't know all the great things that come from it for a while," in the same way that it's taken decades to see the medical benefits of the original map. "We

shouldn't expect miracles."

The cost of mapping the human genome has come down to less than \$1,000 and in some cases to \$300, allowing it to be used in medical care. Though it's early days, the human genome is routinely used in determining cancer treatment, reactions to certain drugs and identifying inherited genetic diseases.

Phillippy hopes that within a decade, doctors and patients will have regular access to their complete genome – including the recently filled gaps – for less than \$1,000, so it can provide more routine benefits in medical care.

"This is a milestone on that pathway," Church said. "We'll get to celebrate multiple times" before that becomes reality, he said.

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Health and patient safety coverage at USA TODAY is made possible in part by a grant from the Masimo Foundation for Ethics, Innovation and Competition in Healthcare. The Masimo Foundation does not provide editorial input.