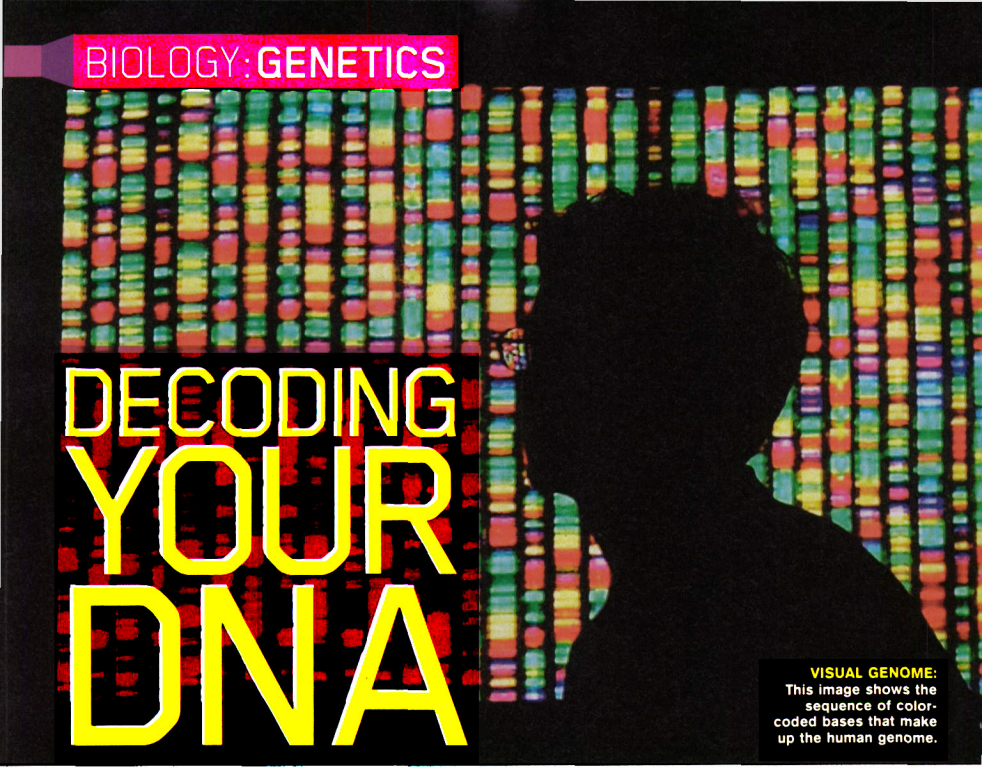
DECODING YOUR DNA



VISUAL GENOME: This image shows the sequence of color-coded bases that make up the human genome.

It's becoming easier to unravel a person's entire genetic code. What will this information reveal about you?

Back in 1990, biologists began an epic task. They set out to decode the entire human *genome*—the blueprint of life encoded in molecules of *DNA*. DNA is the hereditary material found in every one of our cells. The Human Genome Project cost $3 billion and took more than a decade for scientists to complete.

Although the project was a great success, science has come a long way since then. Now, a machine about the size of a computer printer can *sequence*, or translate, a person's genome in a single day for an estimated $1,000.

Your genome is like your own personal instruction manual: It carries the directions that determine almost everything about you, from the color of your eyes to your blood type. With access to your complete genetic code, you can learn about not only these types of traits, but also about your ancestry—and, perhaps most important, your health.

"Today, sequencing is being used to better understand diseases, their treatment, and their prevention," says Bob Fulton, a molecular biologist at the Genome Institute at Washington University in St. Louis, Missouri.

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CRACKING THE CODE

DNA is made up of four building blocks called *bases*: adenine (A), thymine (T), guanine (G), and cytosine (C). A molecule of DNA looks like a twisted ladder (*see The Human Genome, right*). The ladder's rungs are the bases from each of the ladder's *strands*, or sides, that have paired up—A always with T and G always with C.

Sequencing a person's genetic code means figuring out the order of the 3 billion bases along one of the strands. Written out, a chunk of a translated genome

THE HUMAN GENOME: YOUR INSTRUCTION MANUAL

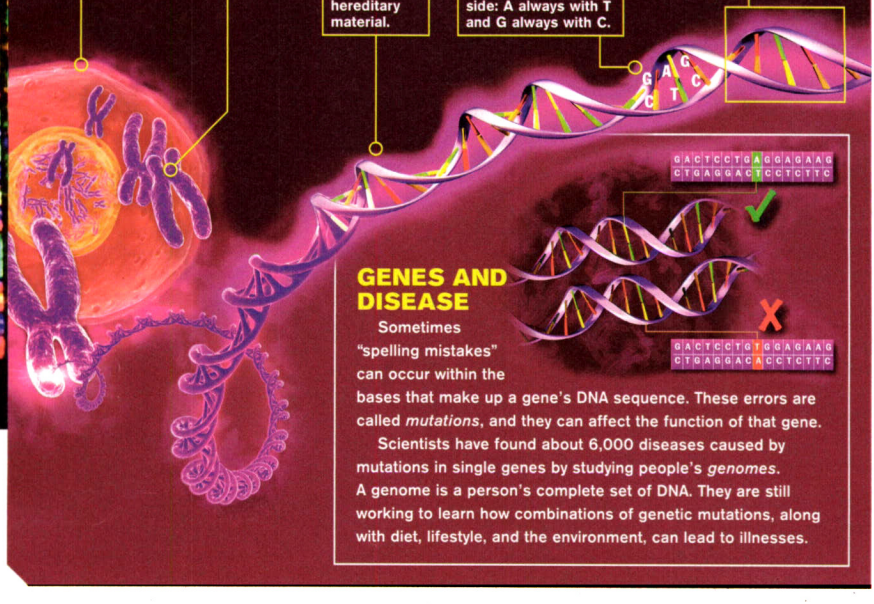
CELLS: The human body contains trillions of *cells*, the basic units of life.

CHROMOSOMES: Packed inside the *nucleus*, or center, of each cell are 46 structures called *chromosomes*.

DNA: Each chromosome contains a long, tightly coiled molecule of *DNA*, a person's hereditary material.

BASES: DNA is made up of a sequence of four *bases*, abbreviated A, G, T, and C. Bases on one side of a DNA molecule pair up with partners on the other side: A always with T and G always with C.

GENES: Series of bases are organized into *genes*, which carry the instructions to make proteins. Proteins keep cells functioning and determine a person's traits.



GENES AND DISEASE

Sometimes "spelling mistakes" can occur within the bases that make up a gene's DNA sequence. These errors are called *mutations*, and they can affect the function of that gene.

Scientists have found about 6,000 diseases caused by mutations in single genes by studying people's *genomes*. A genome is a person's complete set of DNA. They are still working to learn how combinations of genetic mutations, along with diet, lifestyle, and the environment, can lead to illnesses.

would look something like this: ACGACTTAAGTAGTACATG. An entire human genome contains enough letters to fill roughly 400 paperback books, each 500 pages long (*see Size Comparison, p. 22*).

Within a genome's sequence are sections that form *genes*. Genes carry the directions to make *proteins*. These molecules have many jobs in the body. Proteins guide how cells function and ultimately determine a person's different traits. It's estimated that humans have about 25,000 genes.

Sometimes mistakes can crop up in the DNA sequence that codes for a gene. These errors are called *mutations*. Single altered genes cause some illnesses, like the lung disease *cystic fibrosis*. But others, like diabetes, are the result of multiple mutated genes working together—along with factors like diet, lifestyle, and the environment.

For the most part, people's genetic codes are pretty similar. "Between any two individuals, about one in 1,000 bases will be different," says Fulton. He and other scientists use genome sequencing to compare the genetic codes of people who have a certain disease with those who don't. Where the genomes vary could indicate a mutation linked to that disease.

CHEAPER AND FASTER

Sequencing a person's complete genome for $1,000 had long been a goal for scientists. "It puts genome sequencing within the price range of other standard hospital-based diagnostic tests, like an MRI or a CT scan," says Andy Felton, a

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chemist and director of marketing for the company Ion Torrent. This year, Ion Torrent launched the first sequencing machine that hit the $1,000 target. It can sequence a human genome quickly and affordably, thanks to a tiny microchip.

HOW SEQUENCING WORKS

To sequence a person's DNA, its two strands are separated. One strand is then cut into short pieces. Each fragment goes into one of 660 million wells on the microchip's surface. Inside the machine, the chip gets washed with solutions of different bases. These bases match up with their complementary bases on the DNA fragments. This causes a chemical reaction that the chip detects, allowing it to record the position of the bases.

Once the DNA fragments have been sequenced, a computer re-creates the genome. "It's like a giant jigsaw puzzle that you have to put back together," says Felton.

TAILORED MEDICINE

There's still a long way to go before genome sequencing becomes a routine part of a doctor visit. But someday, sequencing could allow physicians to customize health care to fit each patient.

From a person's genetic code, a doctor could learn what diseases the patient is at risk for and take measures to help him or her avoid getting sick. Since genes are inherited, physicians could screen patients' genomes for genetic diseases that might be passed on to their children.

Genes can even affect how a person responds to certain medications. Sequencing could help a doctor figure out the most effective prescription to give a patient.

CHEAP CHIP: This microchip allows a new sequencing machine to decode a person's genome for about $1,000.

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RISKS VS. BENEFITS

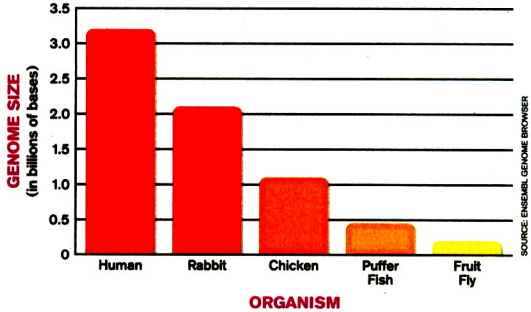
Is there a downside to having your genome sequenced? Some people may not want to know if they have a high risk of developing a condition that is currently untreatable. Others worry about whether their genetic information could be used against them. This concern prompted Congress to pass the Genetic Information Nondiscrimination Act in 2008. It protects people from things like being denied insurance coverage or being fired based on their genes.

Right now, scientists are studying the human genome to figure out how to best help people use the wealth of information it contains. "There's endless work to be done yet," says Fulton. But he predicts that within your lifetime, decoding people's genetic codes will be commonplace. "The cheaper DNA sequencing gets," he says, "the more readily available it becomes for everyone to have their genome sequenced."

— Cody Crane

SIZE COMPARISON

Species' genomes vary in size. But just because a species is small doesn't mean its genome is too. A single-celled amoeba has the largest known genome, at 670 billion bases long. Compare that with humans'.



WHAT DO YOU THINK?

Would you choose to have your genome sequenced to screen for diseases? Explain why or why not.