Chapter 7

Genetic Technologies

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7.1 The Human Genome

As soon as the structure of DNA was known, scientists began to develop tools for studying—as well as manipulating—this genetic material. Advances in DNA technology now allow scientists to study the total genetic material of a living organism—the organism's **genome**. One of the first genomes scientists chose to study was the human genome.

The Human Genome Project, which was completed in 2003, determined the DNA sequence of the entire human genome. As a result of this project, we now know that there are about 3.2 billion nucleotide pairs in the human genome. Amazingly, more than 99.9% of these are identical in all humans. We also know that the human genome is not made up exclusively of genes. In fact, less than 2% of the genome carries genetic instructions for building proteins. Most of the genome—at least 50%—consists of repeat sequences that appear over and over again. Although repeat sequences do not have a direct function, they may occasionally rearrange the human genome, creating new genes or reshuffling existing ones.

We also know that humans have a total of about 19,000 protein-producing genes. Genes are often found in clusters at random spots along our chromosomes, with large sections of non-protein-coding DNA between them. Some genes give rise to RNA transcripts that are processed in different ways, so that one gene can provide the instructions for building multiple proteins. The function of more than half of our genes is still completely unknown.

Scientists have found numerous locations in the genome where the nucleotide sequence differs among human beings. These differences are called *single-nucleotide polymorphisms*, or *SNPs*. SNPs make every person's DNA unique, which is how forensic scientists are able to identify individuals based upon samples containing any trace DNA. SNPs are also helpful in allowing scientists to identify genes related to human diseases.

READING CHECK

Scientists believe that most mutations have little effect on genes or organisms. Can you use what you learned about the human genome to support this statement?

CHECK YOUR ANSWER

Genes make up less than 2% of the human genome. This suggests that, just by chance, the majority of mutations that strike human DNA will not affect any genes.



Learn more at the website of the Human Genome Project here:

https://www.genome.gov/human-genome-project



