Biology & Society

Are Laws Protecting Genetic Privacy Necessary?

The rapid development of new tools and techniques to analyze DNA makes it possible to test for alleles related to thousands of medical conditions. In theory, the results of genetic testing should benefit everyone. Accurate genetic data helps physicians select the proper treatments for patients. It may allow people with genes that place them at risk of certain conditions to minimize those risks.

At issue, however, is individual privacy. Once a test is done, who has access to the data, and how can they use it? Could employers refuse to hire people who might drive up their medical costs? Might insurance companies refuse to renew the policies of individuals with genes for certain disorders? These are not hypothetical questions. In 2005, managers of a professional basketball team asked one of its players to be tested for a gene linked to heart ailments. When he refused, they traded the player to another team. Dr. Francis Collins, director of the National Human Genome Research Institute, worries that "the public is afraid of taking advantage of genetic testing." Is he correct? Should genetic data be protected by law, or should it be open to public view?

The Viewpoints

Genetic Privacy Does Not Need Legal Protection
Other laws already protect individuals from discrimination on the basis of medical disability. Employers and insurance companies are nonetheless allowed to ask individuals if they smoke, use alcohol, or have a history of medical problems. Having this information allows employers to make intelligent choices about whom to hire. It also helps insurance companies maintain lower rates for their healthiest clients. Free access to genetic data should be a public right.

Many commercial laboratories test human DNA for genetic disorders.

Genetic Privacy Should Be Protected by Law
The Genetic Information Nondiscrimination Act (GINA) went into effect in 2009, and it provides important protections to personal privacy. Individuals may not take advantage of today's advances in genetic medicine if they fear their personal information might be used to deny them employment or insurance. We need such laws to realize the full benefits of modern medicine and to protect otherwise healthy individuals from genetic discrimination.

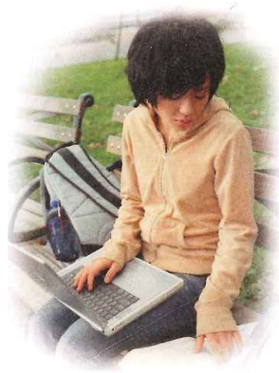
Research and Decide

1. Analyze the Viewpoints To make an informed decision, learn more about genetic testing by consulting library or Internet resources. Then, list the key arguments expressed by the proponents and critics of both points of view. Find out if laws preventing genetic discrimination have been proposed or passed in your state.

2. Form an Opinion Should access and use of genetic data be regulated? Weigh both sides of the issue. Who will benefit from the sharing of genetic data? Will anyone suffer? Do some arguments outweigh others? If so, which ones? Explain your answers.

14.3 Studying the Human Genome

THINK ABOUT IT Just a few decades ago, computers were gigantic machines found only in laboratories and universities. Today, many of us carry small, powerful computers to school and work every day. Decades ago, the human genome was unknown. Today, we can see our entire genome on the Internet. How long will it be before having a copy of your own genome is as ordinary as carrying a cellphone in your pocket?



Manipulating DNA

What techniques are used to study human DNA?

Since discovering the genetic code, biologists have dreamed of a time when they could read the DNA sequences in the human genome. For a long time, it seemed impossible. DNA is a huge molecule—even the smallest human chromosome contains nearly 50 million base pairs. Manipulating such large molecules is extremely difficult. In the late 1960s, however, scientists found they could use natural enzymes in DNA analysis. From this discovery came many useful tools. **By using tools that cut, separate, and then replicate DNA base by base, scientists can now read the base sequences in DNA from any cell.** Such techniques have revolutionized genetic studies of living organisms, including humans.

Cutting DNA Nucleic acids are chemically different from other macromolecules such as proteins and carbohydrates. This difference makes DNA relatively easy to extract from cells and tissues. However, DNA molecules from most organisms are much too large to be analyzed, so they must first be cut into smaller pieces. Many bacteria produce enzymes that do exactly that. Known as **restriction enzymes,** these highly specific substances cut even the largest DNA molecule into precise pieces, called restriction fragments, that are several hundred bases in length. Of the hundreds of known restriction enzymes, each cuts DNA at a different sequence of nucleotides.

In Your Notebook *Make a flowchart that shows the processes scientists use to analyze DNA.*

Key Questions

- ***What techniques are used to study human DNA?***
- ***What were the goals of the Human Genome Project, and what have we learned so far?***

Vocabulary

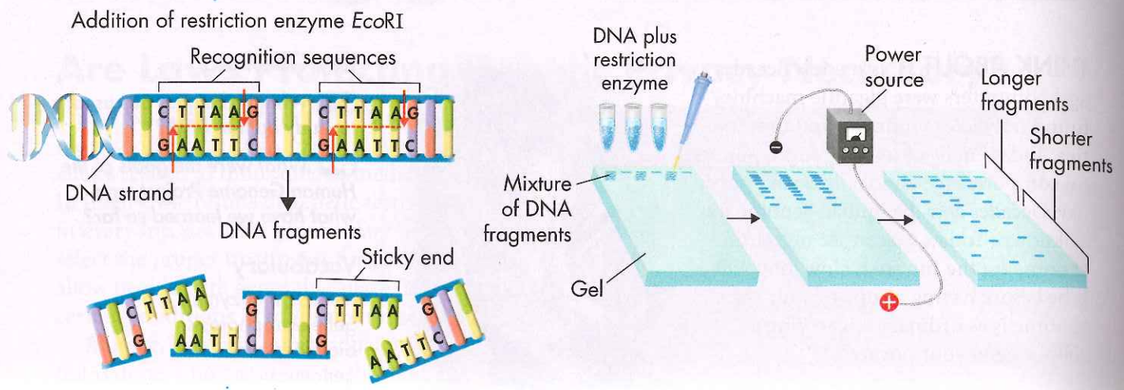
restriction enzyme
gel electrophoresis
bioinformatics
genomics

Taking Notes

Preview Visuals Before you read, look at **Figure 14–10,** and write down three questions you have about the figure. As you read, find answers to your questions.

Cutting DNA
A restriction enzyme is like a key that fits only one lock. The *Eco*RI restriction enzyme can only recognize the base sequence GAATTC. It cuts each strand of DNA between the G and A bases, leaving single-stranded overhangs with the sequence AATT. The overhangs are called "sticky ends" because they can bond, or "stick," to a DNA fragment with the complementary base sequence.

Separating DNA
Gel electrophoresis is used to separate DNA fragments. After being cut by restriction enzymes, the fragments are put into wells on a gel that is similar to a slice of gelatin. An electric voltage moves them across the gel. Shorter fragments move faster than longer fragments. Within an hour or two, the fragments all separate, each appearing as a band on the gel.



VISUAL SUMMARY

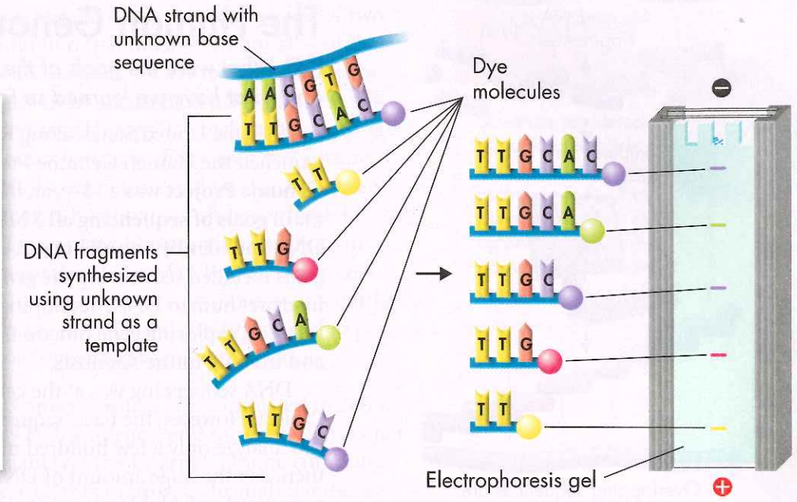
HOW SCIENTISTS MANIPULATE DNA

FIGURE 14–10 By using tools that cut, separate, and replicate DNA, scientists can read the base sequences in DNA from any cell. Knowing the sequence of an organism's DNA allows us to study specific genes.

Separating DNA Once DNA has been cut by restriction enzymes, scientists can use a technique known as **gel electrophoresis** to separate and analyze the differently sized fragments. **Figure 14–10** illustrates this simple, yet effective, method. A mixture of DNA fragments is placed at one end of a porous gel. When an electric voltage is applied to the gel, DNA molecules—which are negatively charged—move toward the positive end of the gel. The smaller the DNA fragment, the faster and farther it moves. The result is a pattern of bands based on fragment size. Specific stains that bind to DNA make these bands visible. Researchers can then remove individual restriction fragments from the gel and study them further.

Reading DNA After the DNA fragments have been separated, researchers use a clever chemical "trick" to read, or sequence, them. The single-stranded DNA fragments are placed in a test tube containing DNA polymerase—the enzyme that copies DNA—along with the four nucleotide bases, A, T, G, and C. As the enzyme goes to work, it uses the unknown strand as a template to make one new DNA strand after another. The tricky part is that researchers also add a small number of bases that have a chemical dye attached. Each time a dye-labeled base is added to a new DNA strand, the synthesis of that strand stops. When DNA synthesis is completed, the result is a series of color-coded DNA fragments of different lengths. Researchers can then separate these fragments, often by gel electrophoresis. The order of colored bands on the gel tells the exact sequence of bases in the DNA. The entire process can be automated and controlled by computers, so that DNA sequencing machines can read thousands of bases in a matter of seconds.

Reading DNA
A small proportion of dye-labeled nucleotides are used to make a complementary DNA strand. Each time a labeled nucleotide is added to the strand, DNA replication stops. Because each base was labeled with a different color, the result is color-coded DNA fragments of different lengths. When gel electrophoresis is used to separate the fragments, scientists can "read" the DNA sequence directly from the gel.

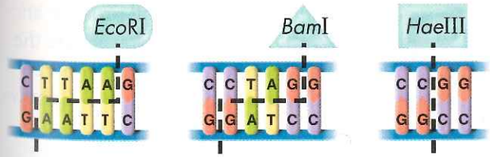


Base sequence as "read" from the order of the bands on the gel from bottom to top: **T G C A C**

Quick Lab
GUIDED INQUIRY

Modeling Restriction Enzymes

1. Write a 50-base, double-stranded DNA sequence using the bases A, C, G, and T in random order. Include each sequence shown below at least once in the sequence you write.
2. Make three copies of your double-stranded sequence on three different-colored strips of paper.
3. Use the drawings below to see how the restriction enzyme *Eco*RI would cut your DNA sequence. Use scissors to cut one copy of the sequence as *Eco*RI would.



4. Use the procedure in Step 3 to cut apart another copy of your sequence as the restriction enzyme *Bam*I would. Then, cut the third copy as the restriction enzyme *Hae*III would.
5. Tape the single-stranded end of one of your DNA fragments to a complementary, single-stranded end of a classmate's fragment. This will form a single, double-stranded DNA molecule.

Analyze and Conclude

1. Observe Which restriction enzyme produced the most pieces? The fewest pieces?

2. Evaluate How well did your model represent the actual process of using restriction enzymes to cut DNA? (*Hint*: Contrast the length of your model DNA sequence to the actual length of a DNA molecule.)

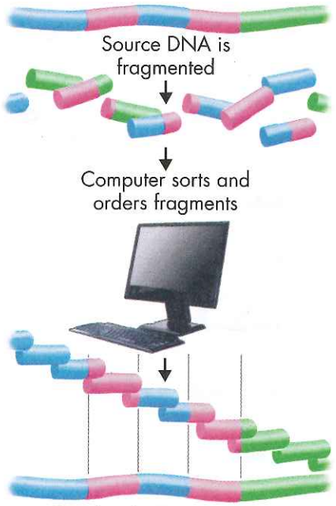


FIGURE 14–11 Shotgun Sequencing This method rapidly sorts DNA fragments by overlapping base sequences.

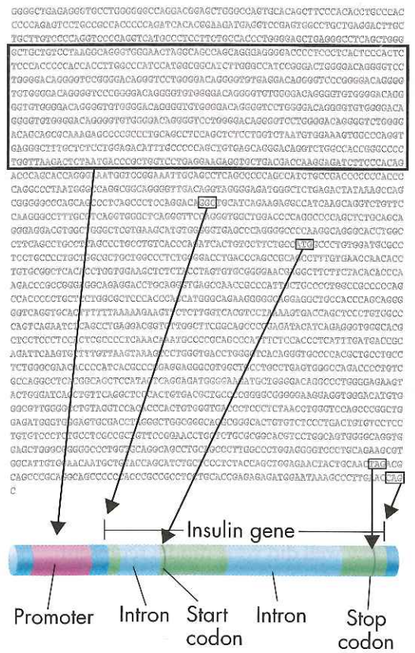


FIGURE 14–12 Locating a Gene A typical gene, such as that for insulin, has several DNA sequences that can serve as locators. These include the promoter, sequences between introns and exons, and start and stop codons.

The Human Genome Project

What were the goals of the Human Genome Project, and what have we learned so far?

In 1990, the United States, along with several other countries, launched the Human Genome Project. **The Human Genome Project was a 13-year, international effort with the main goals of sequencing all 3 billion base pairs of human DNA and identifying all human genes.** Other important goals included sequencing the genomes of model organisms to interpret human DNA, developing technology to support the research, exploring gene functions, studying human variation, and training future scientists.

DNA sequencing was at the center of the Human Genome Project. However, the basic sequencing method you saw earlier can analyze only a few hundred nucleotides at a time. How, then, can the huge amount of DNA in the human genome be sequenced quickly? First, researchers must break up the entire genome into manageable pieces. By determining the base sequences in widely separated regions of a DNA strand, they can use the regions as markers, not unlike the mile markers along a road that is thousands of miles long. The markers make it possible for researchers to locate and return to specific locations in the DNA.

Sequencing and Identifying Genes Once researchers have marked the DNA strands, they can use the technique of "shotgun sequencing." This rapid sequencing method involves cutting DNA into random fragments, then determining the base sequence in each fragment. Computer programs take the sequencing data, find areas of overlap between fragments, and put the fragments together by linking the overlapping areas. The computers then align these fragments relative to the known markers on each chromosome, as shown in **Figure 14–11.** The entire process is like putting a jigsaw puzzle together, but instead of matching shapes, the computer matches DNA base sequences.

Reading the DNA sequence of a genome is not the same as understanding it. Much of today's research explores the vast amount of data from the Human Genome Project to look for genes and the DNA sequences that control them. By locating sequences known to be promoters—binding sites for RNA polymerase—scientists can identify many genes. Shortly after a promoter, there is usually an area called an open reading frame, which is a sequence of DNA bases that will produce an mRNA sequence. Other sites that help to identify genes are the sequences that separate introns from exons, and stop codons located at the ends of open reading frames. **Figure 14–12** shows these sites on a typical gene.

Comparing Sequences If you were to compare the genomes of two unrelated individuals, you would find that most—but not all—of their DNA matches base-for-base with each other. On average, one base in 1200 will not match between two individuals. Biologists call these single base differences SNPs (pronounced "snips"), which stands for single nucleotide polymorphisms. Researchers have discovered that certain sets of closely linked SNPs occur together time and time again. These collections of linked SNPs are called haplotypes—short for haploid genotypes. To locate and identify as many haplotypes in the human population as possible, the International HapMap Project began in 2002. The aim of the project is to give scientists a rapid way to identify haplotypes associated with various diseases and conditions and to pave the way to more effective life-saving medical care in the future.

Sharing Data The Human Genome Project was completed in 2003. Copies of the human genome DNA sequence, and those of many other organisms, are now freely available on the Internet. Online computer access enables researchers and students to browse through databases of human DNA and study its sequence. More data from the human genome, and the genomes of other organisms, are added to these databases every day.

One of the key research areas of the Human Genome Project was a new field of study called **bioinformatics.** The root word, *informatics,* refers to the creation, development, and operation of databases and other computing tools to collect, organize, and interpret data. The prefix *bio-* refers to life sciences—specifically, molecular biology. Assembling the bits and pieces of the human genome would have been impossible without sophisticated computer programs that could recognize overlapping sequences and place them in the proper order, or immense databases where such information could be stored and retrieved. Without the tools of bioinformatics shown in **Figure 14–13,** the wealth of information gleaned from the Human Genome Project would hardly be useful. Bioinformatics also launched a more specialized field of study known as **genomics**—the study of whole genomes, including genes and their functions.

MYSTERY CLUE

Scientists can detect the sickle cell allele with a test for SNPs in the genes for the polypeptides that make up hemoglobin. What does this tell you about the sickle cell mutation?

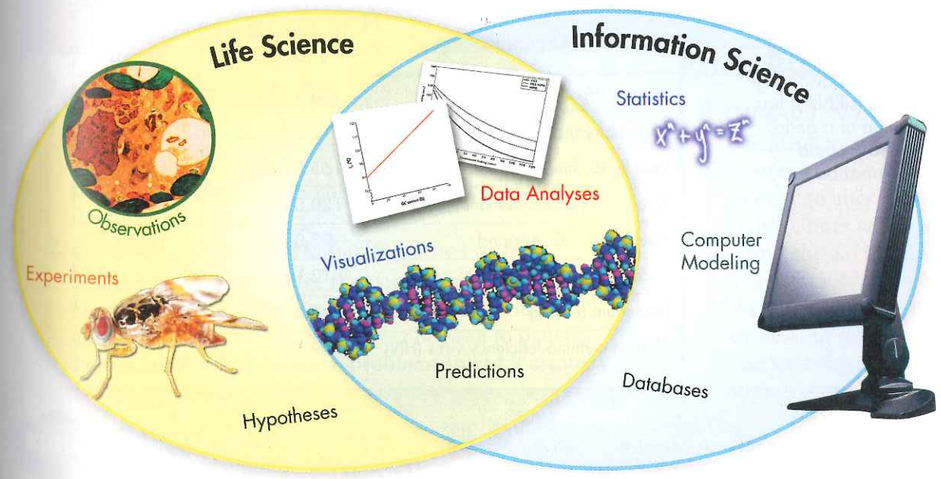


FIGURE 14–13 Bioinformatics Bioinformatics is a new field that combines molecular biology with information science. It is critical to studying and understanding the human genome.

FIGURE 14–14 Announcements The first details of the human genome appeared in two well-known scientific journals in February 2001, *Science* and *Nature* (shown here).

What We Have Learned In June 2000, scientists announced that a working copy of the human genome was complete. The first details appeared in the February 2001 issues of the journals *Nature* and *Science.* The full reference sequence was completed in April 2003, marking the end of the Human Genome Project—two years ahead of the original schedule. Coincidentally, that was also the fiftieth anniversary of Watson and Crick's publication of DNA structure that launched the era of molecular biology!

Besides finding that the human genome in its haploid form contains three billion nucleotide bases, the Human Genome Project uncovered a wealth of interesting, and sometimes surprising, information. For instance, only about 2 percent of our genome encodes instructions for the synthesis of proteins, and many chromosomes contain large areas with very few genes. As much as half of our genome is made up of DNA sequences from viruses and other genetic elements within human chromosomes. During the project, investigators completed the genomes of several other organisms, including unicellular ones. They found that more than 40 percent of the proteins coded for by our genome have strong similarity to proteins in many of those organisms, including fruit flies, worms, and even yeast. **Figure 14–15** compares the human genome with these and other model organisms.

By any standard, the Human Genome Project has been a great scientific success. **The Human Genome Project pinpointed genes and associated particular sequences in those genes with numerous diseases and disorders. It also identified about three million locations where single-base DNA differences occur in humans.** This information may help us find DNA sequences associated with diabetes, cancer, and other health problems. The Human Genome Project also transferred important new technologies to the private sector, including agriculture and medicine. By doing so, the project catalyzed the U.S. biotechnology industry and fostered the development of new medical applications.

FIGURE 14–15 Genome Size Comparisons The gene numbers in this table are not final. Some estimates include only protein-coding genes, while others include genes that code only for RNA. The discovery of small interfering RNAs (siRNAs) has complicated the definition of a gene. **Propose a Solution** ***How could you find updated information on genome sizes?***

Size Comparison of Various Genomes

Organism	Genome Size (bases)	Estimated Genes
Human (*Homo sapiens*)	3.2 billion	25,000
Laboratory mouse (*M. musculus*)	2.5 billion	24,174
Fruit fly (*D. melanogaster*)	165.0 million	13,600
Mustard weed (*A. thaliana*)	120.0 million	25,498
Roundworm (*C. elegans*)	97.0 million	19,000
Yeast (*S. cerevisiae*)	12.1 million	6,294
Bacterium (*E. coli*)	4.6 million	4,288
Human immunodeficiency virus (HIV)	9749.0	9

New Questions Throughout its duration, the Human Genome Project worked to identify and address ethical, legal, and social issues surrounding the availability of human genome data and its powerful new technologies. The issues, including privacy, fairness in the use of and access to genomic information, medical issues, and commercialization, are complex. For example, who owns and controls genetic information? Is genetic privacy different from medical privacy? Who should have access to personal genetic information, and how will it be used? Right now, these questions are hypothetical, but they may not be for long. In May 2008, President George Bush signed into law the Genetic Information Nondiscrimination Act, which prohibits U.S. insurance companies and employers from discriminating on the basis of information derived from genetic tests. Other protective laws may soon follow.

What's Next? Many more sequencing projects are underway, helped along by powerful new technologies. You can expect an ever-growing database of information from microbial, animal, and plant genomes in the years ahead. Each of these will have its own mysteries to be explored, not to mention the fact that we still don't fully understand the functions of as many as 50 percent of the human genes thus far discovered.

The 1000 Genomes Project, launched in 2008, will study the genomes of 1000 people in an effort to produce a detailed catalogue of human variation. Data from the project will be used in future studies of development and disease, and the information may hold the key to successful research on new drugs and therapies to save human lives and preserve health.

Perhaps the most important challenge that lies ahead is to understand how all the "parts" of cells—genes, proteins, and many other molecules—work together to create complex living organisms. Future efforts may provide a deeper understanding of the molecular processes underlying life and may influence how we view our own place in the global ecosystem.

14.3 Assessment

Review Key Concepts

1. a. **Review** How do molecular biologists identify genes in sequences of DNA?
   b. **Use Analogies** How is shotgun sequencing similar to doing a jigsaw puzzle?
2. a. **Review** What is the Human Genome Project?
   b. **Form an Opinion** Judge the potential impact of the Human Genome Project on both scientific thought and society. How might the project be used to benefit humankind? What potential problems might it create?

WRITE ABOUT SCIENCE

Persuasion

3. Scientists may one day be able to use genomics and molecular biology to alter a child's inherited traits. Under what circumstances, if any, should this ability be used? When should it not be used? Write a persuasive paragraph expressing your opinion. (*Hint:* Use specific examples of traits to support your ideas.)

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