Advances in Genetics

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CHAPTER 1 Advances in Genetics

Lesson Objectives

- Explain the significance of the Human Genome Project.
- Describe human genetic disorders.
- Identify methods and uses of biotechnology.

Lesson Vocabulary

- biotechnology
- gene therapy
- genetically modified organism (GMO)
- genetic disorder
- genome
- Human Genome Project

Introduction

The science of genetics has come a long way since Mendel's laws were rediscovered in 1900. There have been many advances in genetics. One of the most impressive advances was sequencing the human genome.

Sequencing the Human Genome

A species' **genome** consists of all of its genetic information. The human genome consists of the complete set of genes in the human organism. It's all the DNA of a human being.

The Human Genome Project

The **Human Genome Project** was launched in 1990. It was an international effort to sequence all 3 billion bases in human DNA. Another aim of the project was to identify the more than 20,000 human genes and map their locations on chromosomes. The logo of the Human Genome Project in **Figure** 1.1 shows that the project brought together experts in many fields.

The Human Genome Project was completed in 2003. It was one of the greatest feats of modern science. It provides a complete blueprint for a human being. It's like having a very detailed manual for making a human organism.

Applications of the Sequence

Knowing the sequence of the human genome is very useful. For example, it helps us understand how humans evolved. Another use is in medicine. It is helping researchers identify and understand genetic disorders. You can

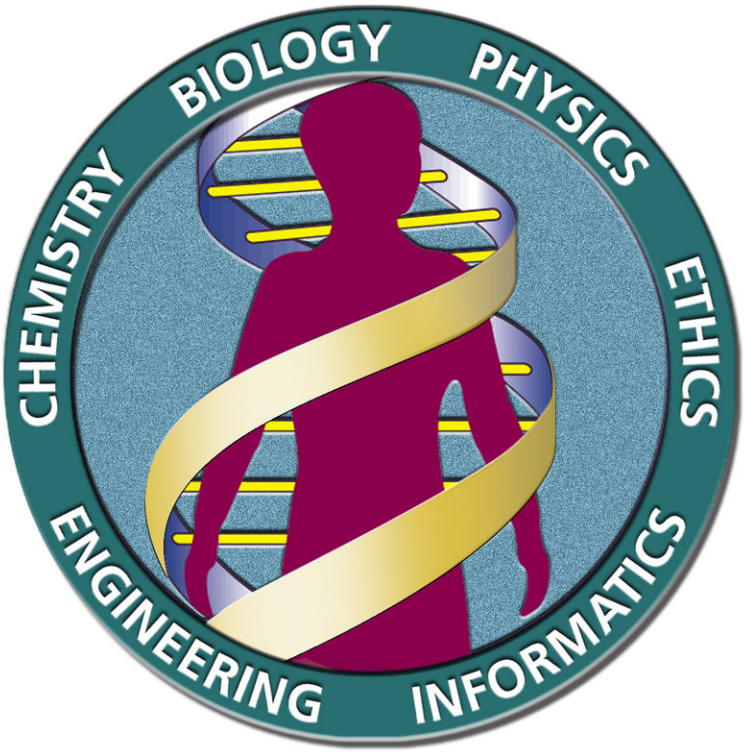

FIGURE 1.1
Human Genome Project logo

learn more about the Human Genome Project and its applications by watching this funny, fast-paced video: http://www.youtube.com/watch?v=F5LzKupeHtw .

MEDIA
Click image to the left or use the URL below.
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Human Genetic Disorders

Sequencing the human genome has increased our knowledge of genetic disorders. **Genetic disorders** are diseases caused by mutations. Many genetic disorders are caused by mutations in a single gene. Others are caused by abnormal numbers of chromosomes.

Disorders Caused by Single Gene Mutations

Table 1.1 lists some genetic disorders caused by mutations in just one gene. It include autosomal and X-linked disorders. It also includes dominant and recessive disorders.

TABLE 1.1: Examples of human genetic disorders caused by single gene mutations

Genetic Disorder	Effect of Mutation	Signs of the Disorder	Type of Trait
Marfan syndrome	Defective protein in tissues such as cartilage and bone	Heart and bone defects; unusually long limbs	Autosomal dominant
Cystic fibrosis	Defective protein needed to make mucus	Unusually thick mucus that clogs airways in lungs and ducts in other organs	Autosomal recessive
Sickle Cell Anemia	Defective hemoglobin protein that is needed to transport oxygen in red blood cells	Sickle-shaped red blood cells that block blood vessels and interrupt blood flow	Autosomal recessive
Hemophilia A	Reduced activity of a protein needed for blood to clot	Excessive bleeding that is difficult to control	X-linked recessive

Relatively few genetic disorders are caused by dominant alleles. A dominant allele is expressed in everybody who inherits even one copy of it. If it causes a serious disorder, affected people may die young and fail to reproduce. They won't pass the allele to the next generation. As a result, the allele may die out of the population. One of the exceptions is Marfan syndrome. It is thought to have affected Abraham Lincoln. He's pictured in **Figure** 1.2. His very long limbs are one reason for the suspicion of Marfan syndrome in this former U.S. president.

Recessive disorders are more common than dominant ones. Why? A recessive allele is not expressed in heterozygotes. These people are called carriers. They don't have the genetic disorder but they carry the recessive allele. They can also pass this allele to their offspring. A recessive allele is more likely than a dominant allele to pass to the next generation rather than die out.

Chromosomal Disorders

In the process of meiosis, paired chromosomes normally separate from each other. They end up in different gametes. Sometimes, however, errors occur. The paired chromosomes fail to separate. When this happens, some gametes get an extra copy of a chromosome. Other gametes are missing a chromosome. If one of these gametes is fertilized and survives, a chromosomal disorder results. You can see examples of such disorders in **Table** 1.2

TABLE 1.2: Disorders caused by abnormal numbers of chromosomes

Genetic Disorder	Genotype	Phenotypic Effects
Down syndrome	Extra copy (complete or partial) of chromosome 21	Developmental delays, distinctive facial appearance, and other abnormalities
Turner's syndrome	One X chromosome and no other sex chromosome (XO)	Female with short height and inability to reproduce
Klinefelter's syndrome	One Y chromosome and two or more X chromosomes (XXY, XXXY)	Male with abnormal sexual development and reduced level of male sex hormone

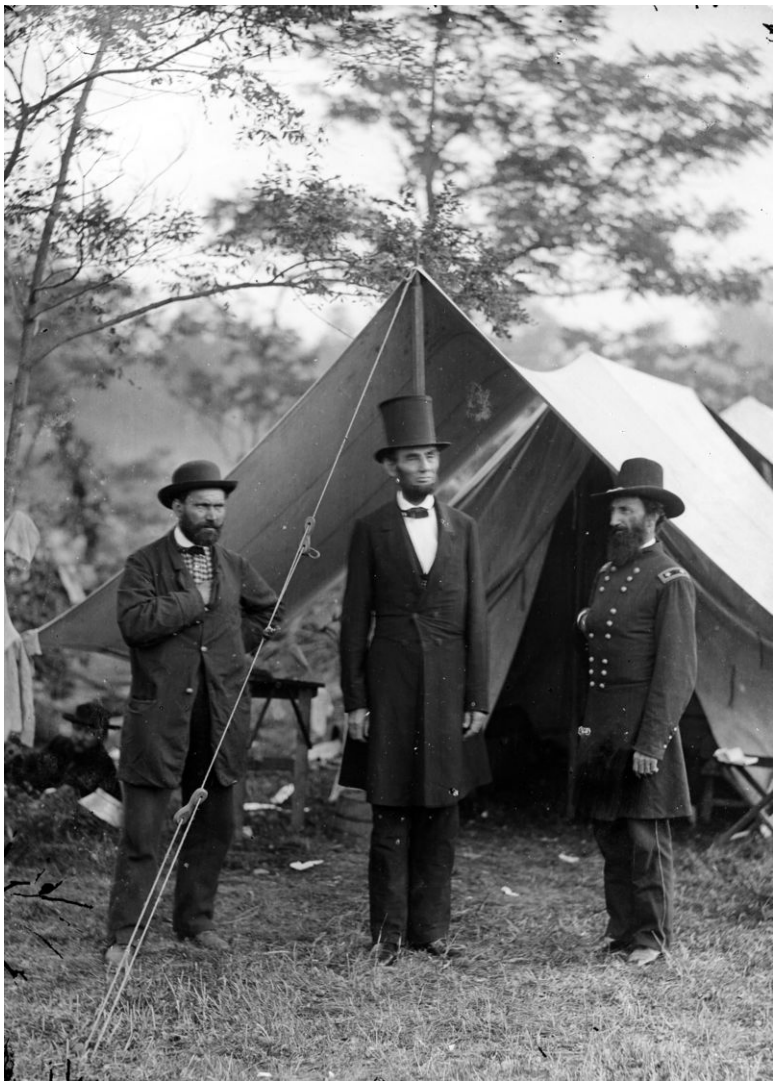

FIGURE 1.2

Abraham Lincoln (center) may have had the genetic disorder Marfan syndrome

Most chromosomal disorders involve the sex chromosomes. Can you guess why? The X and Y chromosomes are very different in size. The X is much larger than the Y. This difference in size creates problems. It increases the chances that the two chromosomes will fail to separate properly during meiosis.

Biotechnology

Treating genetic disorders is one use of biotechnology. **Biotechnology** is the use of technology to change the genetic makeup of living things for human purposes. It's also called genetic engineering. Besides treating genetic disorders, biotechnology is used to change organisms so they are more useful to people.

Methods in Biotechnology

Biotechnology uses a variety of methods, but some are commonly used in many applications. A common method is the polymerase chain reaction. Another common method is gene cloning.

- The polymerase chain reaction is a way of making copies of a gene. It uses high temperatures and an enzyme to make new DNA molecules. The process keeps cycling to make many copies of a gene.
- Gene cloning is another way of making copies of a gene. A gene is inserted into the DNA of a bacterial cell. **Figure** 1.3 shows how this is done. Bacteria multiply very rapidly by binary fission. Each time a bacterial cell divides, the inserted gene is copied.

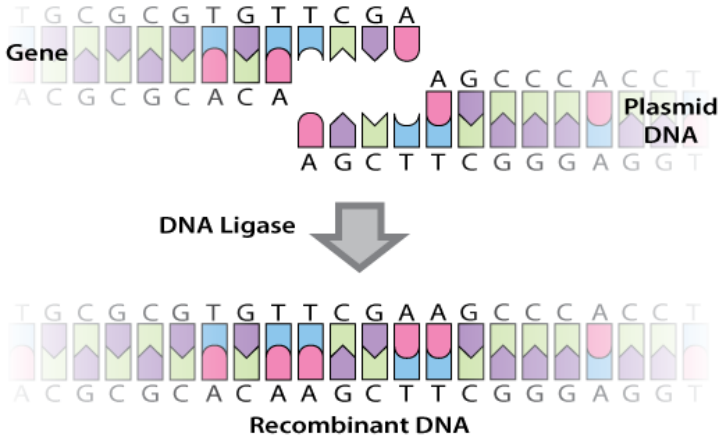

FIGURE 1.3

The enzyme DNA ligase joins together a gene and bacterial (plasmid) DNA. The DNA that results is called recombinant DNA.

Uses of Biotechnology

Biotechnology has many uses. It is especially useful in medicine and agriculture. Biotechnology is used to

- treat genetic disorders. For example, copies of a normal gene might be inserted into a patient with a defective gene. This is called **gene therapy**. Ideally, it can cure a genetic disorder.
- create **genetically modified organisms (GMOs)**. Many GMOs are food crops such as corn. Genes are inserted into plants to give them desirable traits. This might be the ability to get by with little water. Or it might be the ability to resist insect pests. The modified plants are likely to be healthier and produce more food. They may also need less pesticide.
- produce human proteins. Insulin is one example. This protein is needed to treat diabetes. The human insulin gene is inserted into bacteria. The bacteria reproduce rapidly. They can produce large quantities of the human protein. You can see another example in **Figure** 1.4.

Concerns about Biotechnology

Biotechnology has many benefits. Its pros are obvious. It helps solve human problems. However, biotechnology also raises many concerns. For example, some people worry about eating foods that contain GMOs. They wonder if GMOs might cause health problems. The person in **Figure** 1.5 favors the labeling of foods that contain GMOs. That way, consumers can know which foods contain them and decide for themselves whether to eat them.

Another concern about biotechnology is how it may affect the environment. Negative effects on the environment have already occurred because of some GMOs. For example, corn has been created that has a gene for a pesticide. The corn plants have accidentally cross-pollinated nearby milkweeds. Monarch butterfly larvae depend on milkweeds for food. When they eat milkweeds with the pesticide gene, they are poisoned. This may threaten the survival of the monarch species as well as other species that eat monarchs. Do the benefits of the genetically modified corn outweigh the risks? What do you think?

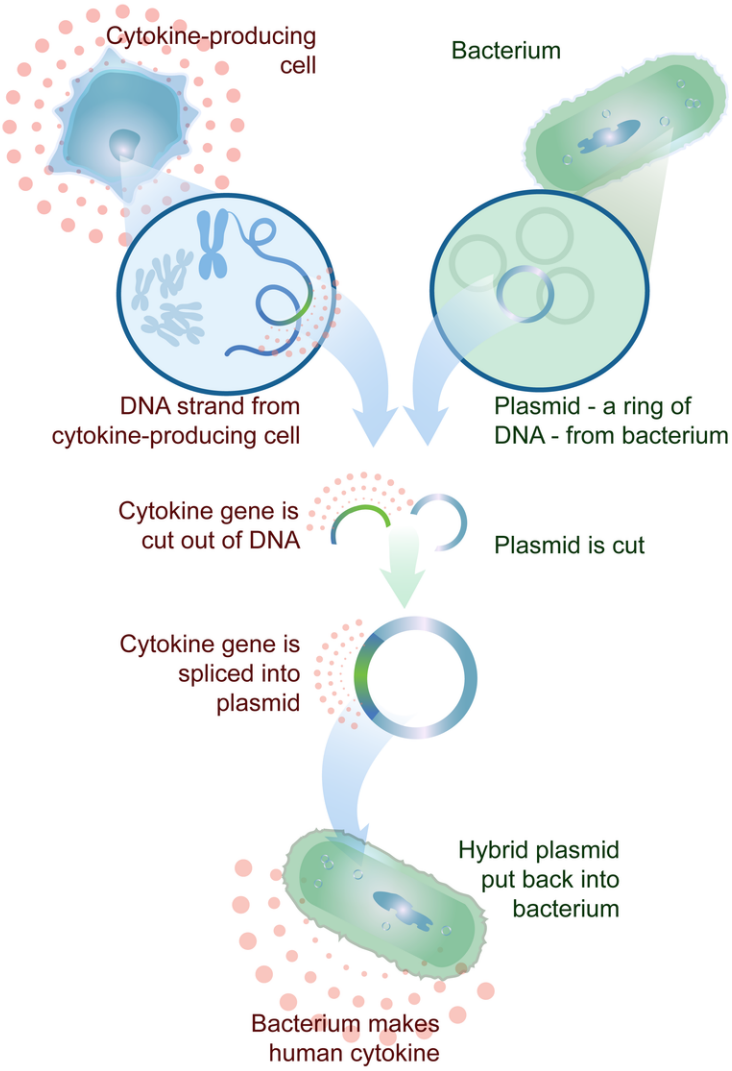

FIGURE 1.4

Bacteria are modified to produce the human protein cytokine. This is a protein that helps fight infections.

Lesson Summary

- A species' genome consists of all of its genetic information. One of the greatest advances in modern genetics was sequencing the human genome. This was achieved in 2003 by the Human Genome Project.
- Sequencing the human genome has increased our knowledge of genetic disorders. These are diseases caused by mutations. They may be caused by single gene mutations or the failure of chromosomes to separate correctly during meiosis.
- Biotechnology is the use of technology to treat genetic disorders or change organisms so they are more useful to people. Methods include gene cloning. Applications include gene therapy and genetically modified food crops.

FIGURE 1.5

Chances are that some of the foods you eat contain GMOs. However, they may not be labeled that way.

Lesson Review Questions

Recall

1. Define genome.
2. What was the Human Genome Project? What had it accomplished by 2003?
3. Identify and describe an autosomal recessive genetic disorder.

Apply Concepts

4. Pedigrees show that a certain genetic disorder passes from mothers to about half of their sons or from fathers to all of their daughters. Only males are actually affected by the disorder. What type of disorder is it?

Think Critically

5. Compare and contrast the polymerase chain reaction and gene cloning.
6. Weigh the pros and cons of using biotechnology to produce genetically modified organisms.

Points to Consider

Biotechnology can be used to artificially change the genetic makeup of organisms in a species.

1. How can the genetic makeup of a species change naturally?
2. What might be the outcome of this type of change?

References

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