CHAPTER 1
MAPPING THE HUMAN GENOME

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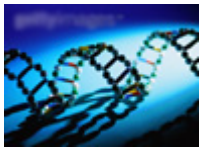

BACKGROUND

DNA, deoxyribonucleic acid, is the most important **molecule** ever discovered because it is the key to how all living things pass their **traits** on to the next generation. The search for DNA, specifically human DNA, has been selected as the topic of this first chapter because of what this discovery means for biology, medicine, law, and ethics.

Some of the questions this chapter explores include:

- What is DNA?
- What was the chain of discoveries that finally led scientists to their current understanding of DNA?
- How advanced is our current understanding of human DNA?
- What is the **human genome project?**
- What effects on society will the mapping of the human genome have?

Rather than focus on the scientific details of this discovery, this chapter gives an overview of the important concepts related to DNA's initial discovery and later research conducted in this field. Teachers can use the lesson plans and materials to help students understand these fundamental concepts and gain a command of the vocabulary necessary to discuss them.

BACKGROUND INFORMATION

It is a well-known fact that we inherit physical traits from our parents. If a child is left-handed, it is very likely that one of her parents writes with his or her left hand. If a boy's father is bald, he will have a greater chance of going bald when he gets older. While everyone knew that physical traits were inherited, it wasn't until the twentieth century that scientists discovered the biological key to this process: DNA, or deoxyribonucleic acid. Finding DNA took almost 100 years.

Now scientists are working to decode the DNA molecule. To do this, they must "read" each of the over 3 billion DNA letters in the human body. This decoding of human DNA, also known as the **human genome**

project, will result in a map of the DNA in the human body. As former U.S. President Clinton said, "Without a doubt, this is the most important, most wondrous map ever produced by humankind."

From 1866 to the present, many scientists have learned important things about how genes work. Below is a review of the important developments in DNA research.

1866	Gregor Mendel, an Austrian monk, described basic elements of **heredity** (these are now called genes).
1860's	Friedrich Miescher, a Swiss chemist, did research on the chemical composition of **white blood cells.** He discovered two types of molecules in the **nucleus** of the blood cells—ribonucleic acid (RNA) and deoxyribonucleic acid (DNA).
1870-1900	There were no major findings during this period. Scientists knew that DNA played some part in heredity, but its structure seemed too simple to play a major role. During this period, scientists thought that **proteins** (with their much more complex structure) played the most important role in heredity.
1902	At Columbia University in New York City, a medical student began to study whether **chromosomes** were made up of genes and if all cells in the body contained these genes.
1920's	Frederick Griffith, an English physician, accidentally discovered a **transforming factor** while doing experiments with **bacteria**. When this factor was taken from one bacteria and put into another, it caused changes in the second bacteria.
1934	Griffith's colleague, Oswald Avery, conducted a 10-year study to identify the transforming factor. His experiments showed that neither proteins nor RNA carry genetic information. He then wondered if DNA was the transforming agent. To answer this question, he conducted an experiment. In it, he destroyed the DNA in the first bacteria. When the DNA was destroyed, no hereditary information was transmitted to the second bacteria. Avery then concluded that DNA causes changes in the second bacteria by transmitting traits from the first bacteria.
1953	James Watson, an American geneticist, and Francis Crick, a British biophysicist, discovered the structure of DNA. To do this, they used X-ray photographs of DNA taken by New Zealand biophysicist Maurice Wilkins. Until this time, it was not known how DNA made a copy of itself in order to transmit genetic information to other cells. In 1962, the three men won the Nobel Prize for their discovery.
1960's	Marshall Nirenberg, an American biochemist, and Har Gobind Khorana, an American biochemist born in India, decoded DNA and discovered the building blocks of DNA. This code consists of four chemical units, represented by the letters A (adenine), T (thymine), C (cytosine), and G (guanine). Each string of letters produces a specific **amino acid**. When these amino acids are combined, they create human traits such as eye color and genetic diseases.
1977	Frederick Sanger, a British **biologist**, developed a method to decode all of the DNA strings in one bacteria. This was the first living organism to be totally decoded.
1990	Agencies of the U.S. government funded a 15-year project to sequence the human genome. This is a map of the cell's inner workings and of all the chemicals produced by DNA that determine human characteristics and behavior.
1999	The Human Genome Project finishes sequencing the first human chromosome.

June 2000 Both the U.S. government-sponsored Human Genome Project and a privately funded research group announced that they have a draft of the first human genome.

Just like many scientific discoveries in the past, this decoding of human DNA will undoubtedly have an impact on our future in ways that are almost unimaginable today. Understanding DNA promises to give us a better understanding of human biology, new diagnostic tests for certain hereditary diseases such as cerebral palsy and breast cancer, and possibly treatments or cures for diseases such as Alzheimer's, diabetes, and heart disease. It will also help companies create drugs that match a person's genetic profile. However, there are many things that still will not be known once the 3.4 billion or so chemical units in human DNA are coded. We won't fully know how many human genes there are in the human organism, and we won't fully know how genes interact with one another.

While there is much work to be done mapping the human genome, scientific findings in the past decade have ensured that a more complete knowledge of human DNA and the contributions it will make to our lives will not occur in the distant future but in our own lifetimes.

CLASSROOM APPLICATIONS

Given the amazing advances in biological research and the new knowledge that has become available to human beings about their own biological makeup, it is important for students to know basic concepts related to DNA research and the human genome project. This following lesson provides a basic introduction to this topic in an interactive fashion.

PRELIMINARY LESSON PLANNING

Materials:

- Prepare enough copies of the student handouts in Appendix **A**, **B**, and **C** for each student in the class.
- If available, bring relevant illustrations (see below) to assist student comprehension.
  If not available, these illustrations can also be found at the Internet sites listed below.
  - **Site with diagrams of the DNA double helix**
  - **Gene Hunters Teaching Guide (PBS)**
  - **Illustrations of human cells (NIH)**

Student Grouping:

- Decide on procedures for grouping students for each activity (see suggestions below). Groups should have no more than six participants.
- For most activities, you should group students either by language proficiency level or, for those activities where language is less of an issue, by expertise (that is, how much group members know about the topic). For some activities, you may wish to group students randomly. In class contexts where students have varied first languages, it is important to put students from different backgrounds into each group.

Vocabulary:

- Before teaching the lesson, preview the glossary items and select those items that need to be taught before you begin the lesson (that is, those that are absolutely essential for introducing and understanding the topic). These will probably include key concept words such as gene, chromosome, heredity, gene mapping, etc.

WARM UP ACTIVITY (APPROXIMATELY 5 MINUTES)

Purpose:

- To find out what students already know about heredity
- To activate vocabulary related to traits, characteristics, and heredity

Procedures:

1. On the blackboard, draw a table with two columns. Label one column “Positive Traits from your Parents” and the other column “Negative Traits from your Parents.”
2. Have students fold a piece of notebook paper in half to make two columns and copy the headings from the blackboard, one at the top of each column.
3. Ask students to brainstorm the positive and negative traits (hair color, eye color, personality, height, weight) that they inherited from their parents and write these in the appropriate columns on their paper. (There is no right or wrong answer).
4. Ask for student volunteers to explain their responses. Write these on the blackboard.
5. On the blackboard, write the words “learned vs. inherited traits.” Ask students to explain the difference. (Inherited traits include intelligence, hair color, eye color, and height; learned traits include academic success, and athletic and musical abilities.)
6. Divide the class into groups of from two to five students. The total number of groups will depend on the number of students in the class. Ask them to look over the traits they generated as a class and discuss which ones are truly inherited.
7. Have students discuss their answers. Put a check mark next to the traits on the blackboard that students agree are inherited.

Transition from Warm Up to Activities

Tell students that the class will be devoted to learning some of the history of DNA research and its role in heredity and to discussing the implications of the project to map the human genome.

ACTIVITY 1 (APPROXIMATELY 30–35 MINUTES)

Purpose:

- To present facts about DNA and the human genome project
- To introduce content-specific vocabulary
- To activate students' speaking and listening skills while collaboratively generating a timeline of DNA research

Procedures:

1. Across the top of the blackboard, draw a timeline containing the dates 1860, 1866, 1902, 1920, 1934, 1953, 1960, 1977, 1990, 1999, and 2000. Leave room beneath and between the dates for you to record information (see Step 4 below).

2. Divide the class into 6 groups. The total number of members in each group will depend on the number of students in the class. Assign each group a letter (A–F). With a large class, you can create multiple groups of A, B, etc. With a small class, this can be done as a whole class activity.

3. Give each group (or, in small classes, each student) a copy of Handout 1 (**Appendix B**) that corresponds to the their letter designation. Instruct students to do the following:

   a. Work with other group members to read and understand your two pieces of information about DNA research.
   b. Turn over your papers. With your group, write a summary in your own words.
   c. Choose a spokesperson who will report your group's summary to the whole class.

4. Call on each group in chronological order, asking the spokesperson to provide a summary of the DNA research that occurred during that period. Write the information under the corresponding date in note form.

5. Ask students if they have clarification questions for the reporting group and allow the group time to answer these questions.

6. Have students copy the completed timeline into their notebooks.

ACTIVITY 2 (APPROXIMATELY 20 MINUTES)

Purpose:

- To provide students with surprising facts about DNA research
- To give students an opportunity to respond to these facts

Procedures:

1. Divide the class into groups of from two to five students. The total number of groups will depend upon the number of students in the class (**Appendix C**).

2. Give students a copy of Handout 2 and ask them to do the following (write instructions on the blackboard if necessary):

   a. Work as a group. Find the information in Column 2 that you think best answers the questions in Column 1. Use key words in the questions to help you guess the answers.
   b. Find a partner from another group and compare your answers.

3. Have the class vote on the answer they believe to be correct for each question.

4. For those answers that the students were not able to figure out, provide the correct answer. See the answer key in **Appendix C**.

5. Have students rejoin their original groups and choose the most surprising answer. Tell them to be prepared to explain their choice.

6. Have groups report and explain their choices to the whole class.

ACTIVITY 3 (APPROXIMATELY 25–30 MINUTES)

Purpose:

- To acquaint students with the primary benefits of DNA research and the human genome project
- To provide students an opportunity to critically assess these benefits
- To provide a context for students to use key vocabulary about this topic

Procedures:

1. On the blackboard, create two columns. Label Column 1 “Benefits of DNA research” and Column 2 “Tally of Class Opinions.” Under Column 1, write the letters A-G. See **Appendix D** for blackboard layout.

2. Divide the class into groups of from two to five students. The total number of groups will depend on the number of students in the class.

3. Give students a copy of Handout 3 and ask them to do the following (write instructions on the blackboard if necessary):

   a. Work as a group to determine the three most important benefits of DNA research.
   b. Discuss the reasons for your group’s selection.

4. Ask each group to report its choices. Tally the responses on the blackboard (see **Appendix D** for blackboard layout).

5. Ask students to defend the reasons for their choices.

6. As a class, consider the two benefits that were ranked the lowest. Discuss why these benefits seemed least important. Write student responses on the blackboard.

COOL DOWN ACTIVITY (APPROXIMATELY 15 MINUTES)

Purpose:

- To wrap up the lesson
- To provide students with an opportunity to consider the possible negative consequences of DNA research
- To provide additional speaking and writing practice

Procedures:

1. Have students individually write two or three questions related to DNA research and the human genome project. These should be questions about what this research means for individuals and society.
2. Divide the class into groups of from two to five students. The total number of groups will depend on the number of students in the class.
3. Ask students to discuss each other's questions and pick the best one to explain to the class. Have each group send one student to write this question on the board.
4. Choose the question that will generate the most interesting discussion. Ask students to discuss their opinions.

POSSIBLE EXTENSIONS TO LESSON

1. Have students visit this **website** to review simple explanations of the basic elements of DNA. This will help them complete all activities in the chapter.
2. Have students visit one of the web sites listed in this chapter (see **Internet Resources**). Have them find one interesting piece of new information to tell the class about genes, DNA, or the human genome project.
3. For Activity 2, have students work in new groups that have one member from each original group (Groups A–F). Have them make their own quiz that draws on the information they have learned. Then allow them to give their quiz to the other groups.
4. For the Cool Down Activity, have students choose one of the questions written on the blackboard and write a journal entry in which they answer the question from their own point of view.

Refer to the websites listed in the next section of this chapter for more information and lesson planning ideas.

INTERNET APPLICATIONS

(Websites with prepared lesson plans and activities)

Quia: Biology Games
http://www.quia.com/shared/biology/

DNA: Genes and Chromosomes
http://www.quia.com/jg/65756.html
The structure of DNA, replication, transcription, translation, and mutations are covered.

Genetics
http://www.quia.com/jg/65753.html
This site reviews basic vocabulary associated with the study of genetics.

Think Quest: Genetics
http://www.thinkquest.org/library/cat_show.html?cat_id=21&cid=1
This site provides a description of and links to several excellent lessons on the subject of genes. It also contains easy-to-understand readings about genetics, both at the basic and advanced levels, often with ready-made quizzes.

DNA and the Mystery of Life (A complete Webquest lesson on DNA designed for high school students)
http://projects.edtech.sandi.net/miramesa/DNAproject/dnaindex.html
By clicking on "a set of questions to guide your study…", students are provided with a worksheet that asks them to answer questions about pictures of DNA.

Biotech, Inc. (A Webquest for high school biology)
http://projects.edtech.sandi.net/kearny/biotech/
In this simulation, students become employees at a biotechnology company whose president challenges them to research local biotechnology companies and design and market a new genetics product.

EXPLOITABLE CONTENT

(Websites with information about the topic, but without any prepared lessons)

Mad Sci Network
http://www.madsci.org/
A staff of science experts are available to answer nearly any question you may have. At this link, simply enter a "key term" for a question you have and you will see what answers have already been provided.
http://www.madsci.org/info/class.html This link gives instructions for how this site can be most effectively used in your classroom and provides links to lessons..

DNA Learning Center
http://vector.cshl.org/
This site by the Cold Spring Harbor Laboratory has an especially useful link to DNA from the Beginning http://vector.cshl.org/dnaftb/ , which is organized around key concepts and is explained by animation, an image gallery, video interviews, problems, biographies, and links.

OFFICIAL DOCUMENTS

National Center for Biotechnology Information (NCBI)

http://www.ncbi.nlm.nih.gov/

This organization, a division of the United States National Institute of Health, stores and analyzes recent discoveries in the field of biotechnology. The link below shows the most recent information on Human genome Sequencing.

The International Centre for Genetic Engineering and Biotechnology (ICGEB)

http://www.icgeb.trieste.it/

This organization is dedicated to the safe use of biotechnology.

The National Human Genome Research Institute

http://www.genome.gov/glossary.cfm

This institute has compiled an excellent audio glossary of genetic terms.

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Jacobs, P., and Zitner, A. (2000, June 27). *Scientists reach milestone in mapping of human genome*. Los Angeles Times, A1, pp. 12–13.

Lemonick, M. D. (2000, July 3). *The genome is mapped. Now what?* Time, pp. 24–29.

Park, A. (2000, October 16). *Designer baby*. Time, p. 102.

APPENDIX A

Glossary

Alzheimer's: A mental disorder that gradually destroys vital nerve cells in the brain. Symptoms include loss of memory, judgment, and reasoning, as well as changes in mood and behavior. It is not a normal part of aging.

Amino Acid: Any of 20 basic elements that make proteins.

Bacteria: A tiny one-cell organism that reproduces by cell division.

Biologist: A person who studies plants, animals, and humans. Many biologists now do research on genetics.

Cell (human): In biology, a structure surrounded by a membrane and containing genetic material (DNA) on the inside. Considered by most biologists to be the basic unit of life.

Chromosome: In organisms without a nucleus (such as bacteria), this is a circular DNA molecule used in genetic engineering. In organisms with a nucleus (including plants and humans), this is one of the threadlike structures within the nucleus that contains DNA.

Diabetes: A disorder caused by the body's decreased production or use of insulin (a hormone produced by the pancreas cells need to be able to use blood sugar).

Diagnostic test: Tests used to identify a particular disease or characteristic.

DNA: (deoxyribonucleic acid) A molecule in the form of a double helix , found within a structure known as a chromosome, within the nucleus of every living cell. First discovered in the 19th century, it controls the daily operation of a cell, and provides the genetic "blueprint" for the physical characteristics of all living organisms.

Gene: A small stretch of DNA that directs the production of proteins. A hereditary unit that occupies a specific position (locus) on the chromosome. This unit has a specific effect on the physical characteristics of the organism.

Gene mapping: Finding the relative positions of genes on a DNA molecule (chromosome) and of the distance between them.

Genetics: The study of heredity.

Hereditary disease: A physical disorder that is inherited from parents or grandparents rather than caused by environmental factors.

Heredity: The passing of certain traits from parents to their offspring through the genes.

Human genome: The complete set of genes in a cell that creates a living organism.

Human genome project: The scientific project to "read" the DNA of human chromosomes. Consists of not one project, but rather hundreds of separate research projects conducted throughout the world. The objective is to create a directory of the genes that can be used to answer questions such as what specific genes do and how they work.

Inherited trait: Ways of looking or being that are caused by the genetic make-up or environment of a living organism.

Learned traits: Ways of looking or being that are caused by the environment of a living organism.

Molecule: A chemical entity consisting of two or more atoms of the same or different elements chemically bonded together.

Nucleus (of a cell): The control center of the cell that contains chromosomes and controls the cell's actions.

Organism: Any living thing, either vegetable or animal.

Protein: The "building blocks" of our bodies that contain substances such as hormones and antibodies to regulate body functions.

RNA: (ribonucleic acid) Like DNA, this is another molecule that is important in genetics. It is different from DNA because it is single stranded (not double stranded).

Trait: Ways of looking or being. Traits that are genetic are passed down through the genes from parents to their children

Transforming factor: Something that causes a change.

White blood cells: Cells that circulate in the blood and work as part of the immune system to fight off "foreign bodies" that cause disease.

Handout 1 for Students in Group A

1866	Gregor Mendel, an Austrian monk, described basic elements of heredity (these are now called genes).
1860's	Friedrich Miescher, a Swiss chemist, did research on the chemical composition of white blood cellss. He discovered two types of molecules in the nucleus of the blood cells—ribonucleic acid (RNA) and deoxyribonucleic acid (DNA).

Handout 1 for Students in Group B

1870-1900	There were no major findings during this period. Scientists knew that DNA played some part in heredity but its structure seemed too simple to play a major role in heredity. During this period, scientists thought that proteins (with their much more complex structure) played the most important role in heredity.
1902	At Columbia University in New York City, a medical student began to study whether chromosomes were made up of genes and if all cells in the body contained these genes.

Handout 1 for Students in Group C

1920's	Frederick Griffith, an English physician, accidentally discovered a transforming factor while doing experiments with bacteria. When this factor was taken from one bacteria and put into another, it caused changes in the second bacteria.
1934	Griffith's colleague, Oswald Avery, conducted a 10-year study to identify the transforming factor. His experiments found that neither proteins nor RNA carry genetic information. He then wondered if DNA was the "transforming agent." To answer this question, he conducted an experiment. In it, he destroyed the DNA in the first bacteria. When the DNA was destroyed, no hereditary information was transmitted to the second bacteria. Avery then concluded that DNA causes changes in the second bacteria by transmitting traits from the first bacteria.

Handout 1 for Students in Group D

1953	James Watson, an American geneticist, and Francis Crick, a British biophysicist, discovered the structure of DNA. To do this, they used X-

	ray photographs of DNA taken by New Zealand biophysicist Maurice Wilkins. Until this time, it was not known how DNA made a copy of itself in order to transmit genetic information to other cells. The three men won the Nobel Prize for their discovery in 1962.
1960's	Marshall Nirenberg, an American biochemist, and Har Gobind Khorana, an American biochemist born in India, decoded DNA and discovered the building blocks of DNA. This code consists of four chemical units, represented by the letters A (adenine), T (thymine), C (cytosine), and G (guanine). Each string of letters produces a specific Amino Acid. When these Amino Acids are combined, they create human traits such as eye color and genetic diseases.

Handout 1 for Students in Group E

1977	Frederick Sanger, a British biologist, developed a method to decode the entire DNA strings in one bacteria. This was the first living organism to be totally decoded.
1990	Agencies of the U.S. government fund a 15-year project to sequence the human genome. This is a map of the cell's inner workings and of all the chemicals produced by DNA that determine human characteristics and behavior.

Handout 1 for Students in Group F

1999	The Human Genome Project finishes sequencing the first human chromosome.
June 2000	Both the U.S. government-sponsored Human Genome Project and a privately funded research group announce that they have a draft of the first human genome.

APPENDIX C

Handout 2 for Activity 2

1. How many pages of newspaper would it take to write out all the DNA in one human cell?	**a.** 10%
2. How many DNA letters are contained in the genome of a mouse?	**b.** 99.9%
3. How many DNA letters are contained in the human genome?	**c.** between 30,000 and 140,000
4. How many human genes do scientists estimate there are?	**d.** less than 5%
5. What percentage of human genes cannot be understood with current technology?	**e.** 3,100,000,000
6. What percentage of all human DNA creates the genes in our body?	**f.** 2,300,000,000
7. What percentage of each human's DNA is identical to that of every other human being?	**g.** 195
8. How many scientists did it take to decode the entire genome of the common fruit fly?	**h.** 100,000

Answer Key for Activity 2

h.	**1.** How many pages of newspaper would it take to write out all the DNA in one human cell?
f.	**2.** How many DNA letters are contained in the genome of a mouse?
e.	**3.** How many DNA letters are contained in the human genome?
c.	**4.** How many human genes do scientists estimate there are?
a.	**5.** What percentage of the genetic code cannot be deciphered with current technology?
d.	**6.** What percentage of all human DNA creates the genes in our body?
b.	**7.** What percentage of each human's DNA is identical to that of every other human being?
g.	**8.** How many scientists did it take to decode the entire genome of the common fruit fly?

APPENDIX D

Blackboard Layout for Activity 3

Benefits of DNA Research	Tally of Class Opinions
a.	XXX
b.	XXXX
c.	etc.
d.	
e.	
f.	
g.	

Handout 3 for Activity 3

_____	a. Pharmaceutical companies will be able to create drugs tailored to a person's genetic profile.
_____	b. Doctors will be able to describe diseases more clearly (for example, they will be able to determine what type of breast cancer a patient has).
_____	c. Doctors will be able to predict a person's chances of getting serious diseases such as heart disease or Alzheimer's.
_____	d. Scientists will be able to predict new technologies such as DNA chips and micro-arrays that show which genes are active in a tissue sample.
_____	e. Scientists will be able to find genes that trigger certain diseases (for example, it will find the genes responsible for diabetes or obesity).
_____	f. Scientists will be able to manipulate the genes for memory, hormones, learning, and human growth and development.
_____	g. Scientists will be able to produce comprehensive databases of DNA sequences in genes that will assist in genetic research.

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