KEY CONCEPT
DNA was identified as the genetic material through a series of experiments.

MAIN IDEA: Griffith finds a “transforming principle.”
Write the results of Griffith’s experiments in the boxes below.

Experiments | Results
--- | ---
1. Injected mice with R bacteria |  
2. Injected mice with S bacteria |  
3. Killed S bacteria and injected them into mice |  
4. Mixed killed S bacteria with R bacteria and injected them into mice | Found live S bacteria in the mice’s blood

5. Which type of bacteria caused disease, the S form or the R form? 

6. What conclusions did Griffith make based on his experimental results?

VOCABULARY
bacteriophage
MAIN IDEA: Avery identifies DNA as the transforming principle.

7. Avery and his team isolated Griffith’s transforming principle and performed three tests to learn if it was DNA or protein. In the table below, summarize Avery’s work by writing the question he was asking or the results of his experiment.

<table>
<thead>
<tr>
<th>Avery’s Question</th>
<th>Results</th>
</tr>
</thead>
<tbody>
<tr>
<td>What type of molecule does the transforming principle contain?</td>
<td>The ratio of nitrogen to phosphorus in the transforming principle is similar to the ratio found in DNA.</td>
</tr>
<tr>
<td>Which type of enzyme destroys the ability of the transforming principle to function?</td>
<td></td>
</tr>
</tbody>
</table>

MAIN IDEA: Hershey and Chase confirm that DNA is the genetic material.

8. Proteins contain ______________ but very little ______________.

9. DNA contains ______________ but no ______________.

10. Summarize the two experiments performed by Hershey and Chase by completing the table below. Identify what type of radioactive label was used in the bacteriophage and whether radioactivity was found in the bacteria.

<table>
<thead>
<tr>
<th>Experiment</th>
<th>Bacteriophage</th>
<th>Bacteria</th>
</tr>
</thead>
<tbody>
<tr>
<td>Experiment 1</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Experiment 2</td>
<td></td>
<td></td>
</tr>
</tbody>
</table>

Vocabulary Check

11. Explain what a bacteriophage is and describe or sketch its structure.
8.1 IDENTIFYING DNA AS THE GENETIC MATERIAL

Griffith’s experiments:

Conclusion:

Avery’s experiments:

- 
- 
- 

Conclusion:

Hershey and Chase’s experiments:

- 
- 

Conclusion:
SECTION 8.1 IDENTIFYING DNA AS THE GENETIC MATERIAL

**Reinforcement**

**KEY CONCEPT** DNA was identified as the genetic material through a series of experiments.

A series of experiments helped scientists recognize that DNA is the genetic material. One of the earliest was done by Frederick Griffith who was studying two forms of the bacterium that causes pneumonia. The S form was surrounded by a coating that made them look smooth. The R form did not have a coating, and the colonies looked rough. Griffith injected these bacteria into mice and found that only the S type killed the mice. When the S bacteria were killed, they did not cause the mice to die. However, when killed S bacteria were mixed with live R bacteria, the mice died and Griffith found live S bacteria in their blood. This led Griffith to conclude that there was a transforming principle that could change R bacteria into S bacteria.

Oswald Avery, another scientist, developed a way to purify this transforming principle. He then conducted a series of chemical tests to find out what it was. Many scientists thought that DNA was too simple of a molecule to be the genetic material and that proteins, being more complex, were a better candidate. However, Avery made three key discoveries about his samples of transforming principle that showed otherwise:

- DNA was present, not protein.
- The chemical composition matched that of DNA, not protein.
- The addition of enzymes that break down DNA made the transforming principle inactive. The addition of enzymes that break down proteins or RNA had no effect.

Alfred Hershey and Martha Chase carried out the final, conclusive experiments in 1952. **Bacteriophages** are viruses that infect bacteria and take over bacteria’s genetic machinery to make more viruses. They consist of a protein coat surrounding DNA. Hershey and Chase grew these viruses in cultures containing radioactively labeled sulfur, a component of proteins, or phosphorus, a component of DNA. Bacteria were then infected with viruses that either had radioactively labeled sulfur or phosphorous. Hershey and Chase next separated the viruses from the bacteria with a blender. The bacteria had radioactive phosphorus but no radioactive sulfur. Hershey and Chase concluded that the viruses’ DNA, but not the protein coat, had entered the bacteria.

1. What was “transformed” in Griffith’s experiment?

2. Which molecule had entered the bacterium in the Hershey-Chase experiments, sulfur or phosphorus? Which molecule is a major component of DNA?
SECTION 8.2

STRUCTURE OF DNA

Study Guide

KEY CONCEPT
DNA structure is the same in all organisms.

VOCABULARY
- nucleotide
- base pairing rules
- double helix

MAIN IDEA: DNA is composed of four types of nucleotides.

In the space below, draw a nucleotide and label its three parts using words and arrows.

1. How many types of nucleotides are present in DNA?

2. Which parts are the same in all nucleotides? Which part is different?

MAIN IDEA: Watson and Crick developed an accurate model of DNA’s three-dimensional structure.

3. What did Franklin’s data reveal about the structure of DNA?

4. How did Watson and Crick determine the three-dimensional shape of DNA?
5. How does DNA base pairing result in a molecule that has a uniform width?

**MAIN IDEA:** Nucleotides always pair in the same way.

6. What nucleotide pairs with T? with C?

In the space below, draw a DNA double helix. Label the sugar-phosphate backbone, the nitrogen-containing bases, and the hydrogen bonds.

**Vocabulary Check**

7. Explain how the DNA double helix is similar to a spiral staircase.

8. How do the base pairing rules relate to Chargaff’s rules?
SECTION 8.2 STRUCTURE OF DNA

Power Notes

Parts of a DNA molecule

Overall shape:

Nitrogen-containing bases

Backbone

1.

2.

Base pairing rules:

Bonding

1.

2.

Chargaff’s rules:

Pyrimidines

Purines

Nitrogen-containing bases

Backbone

OVERALL SHAPE:

A

G

T

C

G

C

A

T

Pyrimidines

Purines

Base pairing rules:

Bonding

1.

2.

Chargaff’s rules:
DNA structure is the same in all organisms.

DNA is a chain of nucleotides. In DNA, each nucleotide is made of a phosphate group, a sugar called deoxyribose, and one of four nitrogen-containing bases. These four bases are cytosine (C), thymine (T), adenine (A), and guanine (G). Two of the bases, C and T, have a single-ring structure. The other two bases, A and G, have a double-ring structure.

Although scientists had a good understanding of the chemical structure of DNA by the 1950s, they did not understand its three-dimensional structure. The contributions of several scientists helped lead to this important discovery.

- Erwin Chargaff analyzed the DNA from many different organisms and realized that the amount of A is equal to the amount of T, and the amount of C is equal to the amount of G. This A = T and C = G relationship became known as Chargaff’s rules.

- Rosalind Franklin and Maurice Wilkins studied DNA structure using x-ray crystallography. Franklin’s data suggested that DNA is a helix consisting of two strands that are a regular, consistent width apart.

James Watson and Francis Crick applied Franklin’s and Chargaff’s data in building a three-dimensional model of DNA. They confirmed that DNA is a double helix in which two strands of DNA wind around each other like a twisted ladder. The sugar and phosphate molecules form the outside strands of the helix, and the bases pair together in the middle, forming hydrogen bonds that hold the two sides of the helix together. A base with a double ring pairs with a base with a single ring. Thus, in accordance with Chargaff’s rules, they realized that A pairs with T, and C pairs with G. The bases always pair this way, which is called the base pairing rules.

1. What did Chargaff’s rules state?
2. What did Franklin’s data show about the three-dimensional structure of DNA?
3. What forms the backbone strands of the DNA double helix? What connects these strands in the middle?
### SECTION 8.3 DNA REPLICATION Study Guide

#### KEY CONCEPT
DNA replication copies the genetic information of a cell.

<table>
<thead>
<tr>
<th>VOCABULARY</th>
</tr>
</thead>
<tbody>
<tr>
<td>replication</td>
</tr>
</tbody>
</table>

#### MAIN IDEA: Replication copies the genetic information.

1. What is DNA replication?

2. Where does DNA replication take place in a eukaryotic cell?

3. When is DNA replicated during the cell cycle?

4. Why does DNA replication need to occur?

5. What is a template?

6. If one strand of DNA had the sequence TAGGTAC, what would be the sequence of the complementary DNA strand?

#### MAIN IDEA: Proteins carry out the process of replication.

7. What roles do proteins play in DNA replication?

8. What must be broken for the DNA strand to separate?

9. Why is DNA replication called semiconservative?
STUDY GUIDE, CONTINUED

Use words and diagrams to summarize the steps of replication, in order, in the boxes below.

10. ____________________  11. ____________________  12. ____________________

13. Human chromosomes have hundreds of ________________, where the DNA is unzipped so replication can begin.

14. DNA polymerase has a ________________ function that enables it to detect errors and correct them.

**MAIN IDEA:** Replication is fast and accurate.

**Vocabulary Check**

15. Explain what DNA polymerase is by breaking the word into its parts.

16. Write a short analogy to explain what replication is.

**Be Creative**

17. People sometimes like to display bumper stickers that relate to their trade or field of study. For example, a chemist may have a bumper sticker that says “It takes alkynes to make the world.” Then, chemists or other people who know that an alkyne is a molecule that contains carbon atoms joined by a triple bond get a nice little chuckle out of the play on words. Use your knowledge of DNA replication to write a bumper sticker.

18. ____________________
SECTION 8.3 | DNA REPLICATION

Power Notes

General description:

1. Process
2. Identify the structures.
3. End result

1. 
2. 
3. 
4. 

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DNA replication copies the genetic information of a cell.

Every cell needs its own complete set of DNA, and the discovery of the three-dimensional structure of DNA immediately suggested a mechanism by which the copying of DNA, or DNA replication, could occur. Because the DNA bases pair in only one way, both strands of DNA act as templates that direct the production of a new, complementary strand. DNA replication takes place during the S stage of the cell cycle.

The process of DNA replication is very similar in both eukaryotes and prokaryotes, but we will focus on eukaryotes.

- During the S stage of the cell cycle, the DNA is loosely organized in the nucleus. Certain enzymes start to unzip the double helix at places called origins of replication. The double helix unzips in both directions along the strand. Eukaryotic chromosomes are very long, so they have many origins of replication to help speed the process. Other proteins hold the two strands apart.

- The unzipping exposes the bases on the DNA strands and enables free-floating nucleotides to pair up with their complementary bases. **DNA polymerases** bond the nucleotides together to form new strands that are complementary to the original template strands.

- The result is two identical strands of DNA. DNA replication is described as semiconservative because each DNA molecule has one new strand and one original strand.

DNA polymerase not only bonds nucleotides together. It also has a proofreading function. It can detect incorrectly paired nucleotides, clip them out, and replace them with the correct nucleotides. Uncorrected errors are limited to about one per 1 billion nucleotides.

1. Why is DNA replication described as semiconservative?

2. What are two major functions that DNA polymerase performs?
KEY CONCEPT
Transcription converts a gene into a single-stranded RNA molecule.

VOCABULARY
- central dogma
- messenger RNA (mRNA)
- RNA
- ribosomal RNA (rRNA)
- transcription
- transfer RNA (tRNA)
- RNA polymerase

MAIN IDEA: RNA carries DNA’s instructions.
Label each of the processes represented by the arrows in the diagram below. Write where each of these processes takes place in a eukaryotic cell in parentheses.

1. Replication (Nucleus)
2. Transcription (Nucleus)
3. Translation (Ribosomes)

Fill in the table below to contrast DNA and RNA.

<table>
<thead>
<tr>
<th>DNA</th>
<th>RNA</th>
</tr>
</thead>
<tbody>
<tr>
<td>4. Contains the sugar deoxyribose</td>
<td>Contains the sugar ribose</td>
</tr>
<tr>
<td>6. Typically double-stranded</td>
<td>Typically single-stranded</td>
</tr>
</tbody>
</table>

MAIN IDEA: Transcription makes three types of RNA.
7. What enzyme helps a cell to make a strand of RNA?
   RNA Polymerase
8. Summarize the three key steps of transcription.

1. RNA Polymerase unwinds DNA.

2. Using one strand of the DNA as a template, RNA Polymerase strings together complementary strand of RNA.

3. The RNA strand detaches from the DNA as it is transcribed and the DNA zips back together.

9. Write the basic function of each type of RNA in the chart below.

<table>
<thead>
<tr>
<th>Type of RNA</th>
<th>Function</th>
</tr>
</thead>
<tbody>
<tr>
<td>mRNA</td>
<td>Intermediate message from DNA that is translated to form a protein</td>
</tr>
<tr>
<td>rRNA</td>
<td>forms part of ribosomes</td>
</tr>
<tr>
<td>tRNA</td>
<td>brings amino acids from cytoplasm to ribosome to help make the growing protein</td>
</tr>
</tbody>
</table>

**MAIN IDEA:** The transcription process is similar to replication.

10. List two ways that the processes of transcription and replication are similar.

Both occur in nucleus of eukaryotic cells, both are catalyzed by large enzymes, both involve unwinding DNA, involve complementary base pairing of DNA stranded, both are highly regulated.

11. List two ways that the end results of transcription and replication differ.

Replication only occurs once during each round of the cell cycle and makes a double-stranded copy of all DNA.

Transcription occurs repeatedly through cell cycle to make proteins, rRNA and tRNA. It makes a single-stranded complement to a specific DNA sequence.

**Vocabulary Check**

12. How does the name of each type of RNA tell what it does?

m=form of DNA message about type of protein to make

r=part of ribosome

T=transfers amino acids from cytoplasm to ribosome

13. What is transcription?

Process of coping a sequence of DNA to produce a complementary strand of RNA.
SECTION 8.4 | TRANSCRIPTION

**Power Notes**

Central Dogma

1. DNA  
   2.  
   3.  
   4.  
   5.  

DNA:
-  
-  
-  

RNA:
-  
-  
-  

Transcription

Label the parts on the lines below. Summarize the steps of transcription in the boxes.

1.  
2.  
3.  
4.  
5.  
6.  
7.  

<table>
<thead>
<tr>
<th>RNA Type</th>
<th>Function</th>
</tr>
</thead>
<tbody>
<tr>
<td>1. Messenger RNA (mRNA)</td>
<td></td>
</tr>
<tr>
<td>2.</td>
<td></td>
</tr>
<tr>
<td>3.</td>
<td></td>
</tr>
</tbody>
</table>
DNA provides the instructions needed by a cell to make proteins. But the instructions are not made directly into proteins. First, a DNA message is converted into RNA in a process called transcription. Then, the RNA message is converted into proteins in a process called translation. The relationship between these molecules and processes is summed up in the central dogma, which states that information flows in one direction, from DNA to RNA to proteins.

Like DNA, RNA is a nucleic acid. It is made of nucleotides that consist of a phosphate group, a sugar, and a nitrogen-containing base. However, RNA differs in important ways from DNA: (1) RNA contains the sugar ribose, not deoxyribose; (2) RNA is made up of the nucleotides A, C, G, and uracil, U, which forms base pairs with A; (3) RNA is usually single-stranded. This single-stranded structure enables RNA to fold back on itself into specific structures that can catalyze reactions, much like an enzyme.

During transcription, a gene is transferred into RNA. Specific DNA sequences and a combination of accessory proteins help RNA polymerase recognize the start of a gene. RNA polymerase is a large enzyme that bonds nucleotides together to make RNA. RNA polymerase, in combination with the other proteins, forms a large transcription complex that unwinds a segment of the DNA molecule. Using only one strand of DNA as a template, RNA polymerase strings together a complementary RNA strand that has U in place of T. The DNA strand zips back together as the transcription complex moves forward along the gene.

Transcription makes three main types of RNA.

- **Messenger RNA (mRNA)** is the intermediate message between DNA and proteins. It is the only type of RNA that will be translated to form a protein.

- **Ribosomal RNA (rRNA)** forms a significant part of ribosomes.

- **Transfer RNA (tRNA)** carries amino acids from the cytoplasm to the ribosome during translation.

The DNA of a cell therefore has genes that code for proteins, as well as genes that code for rRNA and tRNA.

1. What is stated in the central dogma?

2. What are the three main types of RNA? Which is translated into a protein?
KEY CONCEPT
Translation converts an mRNA message into a polypeptide, or protein.

VOCABULARY
<table>
<thead>
<tr>
<th>translation</th>
<th>stop codon</th>
<th>anticodon</th>
</tr>
</thead>
<tbody>
<tr>
<td>codon</td>
<td>start codon</td>
<td></td>
</tr>
</tbody>
</table>

MAIN IDEA: Amino acids are coded by mRNA base sequences.

1. What is translation?
   Process that converts mRNA message into a polypeptide (protein)

2. What is a codon?
   Sequence of 3 nucleotide that code for an amino acid

3. Would the codons in Figure 8.13 be found in a strand of DNA or RNA?
   RNA

4. What is a reading frame?
   Order in which nucleotides are read, they are read as a series of 3 nonoverlapping nucleotides

Refer to Figure 8.13 to complete the table below.

<table>
<thead>
<tr>
<th>Codon</th>
<th>Amino Acid or Function</th>
</tr>
</thead>
<tbody>
<tr>
<td>5. AGA</td>
<td>Arginine (Arg)</td>
</tr>
<tr>
<td>6. UAG</td>
<td>Stop Codon</td>
</tr>
<tr>
<td>7. UGG</td>
<td>tryptophan (Trp)</td>
</tr>
<tr>
<td>8. GGA</td>
<td>Glycine (Gly)</td>
</tr>
</tbody>
</table>

MAIN IDEA: Amino acids are linked to become a protein.

9. Ribosomes and tRNA are the tools that help a cell translate an mRNA message into a polypeptide.

10. The small subunit of a ribosome holds onto the mRNA strand.

11. The large subunit of a ribosome has binding sites for tRNA.
12. A tRNA molecule is attached to an **amino acid** at one end and has an **anticodon** at the other end.

Fill in the cycle diagram below to outline the steps of translation.

**Vocabulary Check**

13. What are AGG, GCA, and GUU examples of?

   **Codons**

14. What is a set of three nucleotides on a tRNA molecule that is complementary to an mRNA codon?

   **Anticodon**

15. What do codons code for in addition to amino acids?

   **Stop codons indicate where translation is to stop.**
SECTION 8.5
TRANSLATION

Power Notes

Reading frame:

Triplet Code
Codon

Common language:

Start codon:

Stop codon:

Ribosome

Anticodon

Transfer RNA (tRNA)

Translation
Parts

1.
2.
3.
4.
5.
6.
7.
8.

Process

1.
2.
3.

From DNA to Proteins
KEY CONCEPT  Translation converts an mRNA message into a polypeptide, or protein.

**Translation** is the process that converts an mRNA message into a polypeptide, or protein. An mRNA message is made up of combinations of four nucleotides, whereas proteins are made up of twenty types of amino acids. The mRNA message is read as a series of non-overlapping **codons**, a sequence of three nucleotides that code for an amino acid. Many amino acids are coded for by more than one codon. In general, codons that code for the same amino acid share the same first two nucleotides. Three codons, called **stop codons**, signal the end of the polypeptide. There is also a **start codon**, which both signals the start of translation and codes for the amino acid methionine. This genetic code is the same in almost all organisms, so it is sometimes called the universal genetic code.

Although tRNA and rRNA are not translated into proteins, they play key roles in helping cells translate mRNA into proteins. Each tRNA molecule folds up into a characteristic L shape. One end has three nucleotides called an **anticodon**, which recognize and bind to a codon on the mRNA strand. The other end of the tRNA molecule carries a specific amino acid. A combination of rRNA and proteins make up the ribosome. Ribosomes consist of a large and small subunit. The large subunit has binding sites for tRNA. The small subunit binds to the mRNA strand.

At the start of translation, a small subunit binds to an mRNA strand. Then the large subunit joins. A tRNA molecule binds to the start codon. Another tRNA molecule binds to the next codon. The ribosome forms a bond between the two amino acids carried by the tRNA molecules and pulls the mRNA strand by the length of one codon. This causes the first tRNA molecule to be released and opens up a new codon for binding. This process continues to be repeated until a stop codon is reached and the ribosome falls apart.

1. What is a codon?
   -

2. What role does tRNA play in translation?
   -

3. What forms the bond between neighboring amino acids?
   -
KEY CONCEPT
Gene expression is carefully regulated in both prokaryotic and eukaryotic cells.

VOCABULARY

<table>
<thead>
<tr>
<th>promoter</th>
<th>exon</th>
</tr>
</thead>
<tbody>
<tr>
<td>operon</td>
<td>intron</td>
</tr>
</tbody>
</table>

MAIN IDEA: Prokaryotic cells turn genes on and off by controlling transcription.

1. Why is gene expression regulated in prokaryotic cells?

2. In prokaryotic cells, gene expression is typically regulated at the start of ____________.

3. A ____________ is a segment of DNA that helps RNA polymerase recognize the start of a gene.

4. An ____________ is a region of DNA that includes a ____________, an ____________, and one or more ____________ that code for proteins needed to carry out a task.

Complete the cause-and-effect diagram below about the lac operon.

- **Bacteria growing in culture**
  - medium without lactose added
  - medium with lactose added

- **The repressor continues to bind to the operator.**

5. ____________

6. ____________

7. ____________

8. ____________

9. The resulting transcript is translated into 3 enzymes.
MAIN IDEA: Eukaryotic cells regulate gene expression at many points.

10. Why do the cells in your body differ from each other?

11. What role do transcription factors play in a cell?

12. What is a TATA box?

13. What is “sonic hedgehog” an example of?

MAIN IDEA: The diagrams below represent unprocessed and processed mRNA in a eukaryotic cell. Using the diagrams as a reference, fill in the legend with the corresponding element: cap, exon, intron, tail.

Legend

Unprocessed MRNA

Processed MRNA

Vocabulary Check

14. What is the difference between an exon and an intron?

15. Make an analogy to help you remember what a promoter is.
Promoter:

Operon:

lac operon:

Without lactose:

With lactose:

Controlling transcription in eukaryotic cells:

mRNA processing:
  •
  •
  •
KEY CONCEPT  Gene expression is carefully regulated in both prokaryotic and eukaryotic cells.

The regulation of gene expression better allows cells to respond to their environment and to interact in a coordinated manner. Controlling the start of transcription is important in both prokaryotic cells and eukaryotic cells. It is especially important in prokaryotic cells because there is no separation between DNA and the cytoplasm.

In prokaryotic cells, genes are often organized into **operons**, which are sets of genes that code for all of the proteins needed to carry out a particular task. These genes are transcribed as a unit, and they are often controlled by a DNA sequence called a promoter. **Promoters** help RNA polymerase know where a gene starts. One of the first operons to be discovered was the **lac** operon, which is involved in the breakdown of the sugar lactose. The **lac** promoter acts like a switch. When lactose is absent, a repressor protein binds to the promoter and blocks RNA polymerase from transcribing the **lac** genes. When lactose is present, it binds to the repressor protein. This action blocks the repressor from binding to the promoter. As a result, RNA polymerase can transcribe the **lac** genes, and lactose is broken down.

The start of transcription is still a very important point of regulation in eukaryotic cells as well. Eukaryotes also have DNA sequences that help regulate transcription. These include promoters, enhancers, and silencers. Some sequences are found in almost all eukaryotic cells, such as the TATA box. Others are more specific. Each gene has a unique combination of sequences and transcription factors, proteins that recognize DNA sequences and help RNA polymerase recognize the start of a gene. Regulating the expression of genes that control the expression of other genes is critical to the normal development of an organism.

The mRNA in eukaryotic cells undergoes processing. An mRNA strand is a patchwork of sequences that are either expressed in the protein or are cut out. The expressed sequences are called **exons**; the sequences removed during processing are called **introns**. In addition, a cap is added that helps prevent break down and directs the mRNA to a ribosome. A tail is added that helps the mRNA strand exit the nucleus.

1. How does the presence of lactose enable RNA polymerase to transcribe the **lac** genes?

2. What types of DNA sequences help eukaryotic cells regulate gene expression?

3. What happens during mRNA processing?
KEY CONCEPT
Mutations are changes in DNA that may or may not affect phenotype.

VOCABULARY
<table>
<thead>
<tr>
<th>mutation</th>
<th>frameshift mutation</th>
</tr>
</thead>
<tbody>
<tr>
<td>point mutation</td>
<td>mutagen</td>
</tr>
</tbody>
</table>

MAIN IDEA: Some mutations affect a single gene, while others affect an entire chromosome.

1. List two types of gene mutations.
   - Point mutation/Substitution; frameshift

2. List two types of chromosomal mutations.
   - Gene duplication; translocation

3. Which type of mutation affects more genes, a gene mutation or a chromosomal mutation?
   - Chromosomal mutation

4. What leads to gene duplication?
   - Unequal crossing over

5. What is a translocation?
   - The attachment of a piece of one chromosome to a nonhomologous chromosome

Below is a string of nucleotides. (1) Use brackets to indicate the reading frame of the nucleotide sequence. (2) Copy the nucleotide sequence into the first box and make a point mutation. Circle the mutation. (3) Copy the nucleotide sequence into the second box and make a frameshift mutation. Use brackets to indicate how the reading frame would be altered by the mutation.

```
AGGCGTCCATGA
```

6. 

7. 
MAIN IDEA: Mutations may or may not affect phenotype.
Fill in the cause-and-effect diagram below to explain how a point mutation may or may not affect phenotype.

13. For a mutation to be passed to offspring, in what type of cell must it occur?

germ cells/gamete

MAIN IDEA: Mutations can be caused by several factors.

14. Can DNA polymerase catch and correct every replication error?

No

15. What is a mutagen?

an agent in the environment that can change DNA

16. How does UV light damage the DNA strand?

UV light can cause neighboring thymine nucleotides to break their hydrogen bonds to adenine and bond with each other instead.

Vocabulary Check

17. What is a mutation?

a change in an organism’s DNA

18. If a nucleotide is deleted from a strand of DNA, what type of mutation has occurred?

a frameshift mutation.
SECTION 8.7 MUTATIONS

Power Notes

Gene Mutations:

Chromosomal Mutations:

Potential impact:

Silent:

Mutagens:
KEY CONCEPT  Mutations are changes in DNA that may or may not affect phenotype.

A **mutation** is a change in an organism’s DNA. Although a cell has mechanisms to deal with mutations, exposure to mutagens may cause mutations to happen more quickly than the body can repair them. **Mutagens** are agents in the environment that can change DNA. Some occur naturally, such as UV light from the Sun. Many other mutagens are industrial chemicals.

Mutations may affect individual genes or an entire chromosome. Gene mutations include point mutations and frameshift mutations.

- A **point mutation** is a substitution in a single nucleotide.
- A **frameshift mutation** involves the insertion or deletion of a nucleotide or nucleotides. It throws off the reading frame of the codons that come after the mutation.

Chromosomal mutations include gene duplications and translocations. Gene duplication is the result of improper alignment during crossing over. It results in one chromosome having two copies of certain genes, and the other chromosome having no copies of those genes. Translocation is the movement of a piece of one chromosome to another, nonhomologous chromosome.

Mutations may or may not affect phenotype. Chromosomal mutations affect many genes and tend to have a large effect on an organism. They may also cause breaks in the middle of a gene, causing that gene to no longer function or to make a hybrid with a new function. The effect of a gene mutation can also vary widely. For example, a point mutation may occur in the third nucleotide of a codon and have no effect on the amino acid coded for. Or the mutation may occur in an intron and thus have no effect. However, the mutation might result in the incorporation of an incorrect amino acid that messes up protein folding and function. Or it might code for a premature stop codon. Even mutations that occur in noncoding regions of DNA can have significant effects if they disrupt a splice site or a DNA sequence involved in gene regulation. For a mutation to affect offspring, it must occur in an organism’s germ cells.

1. **What is a mutation?**

2. **In a frameshift mutation, what is the “frame” that is being shifted?**

3. **How might a point mutation in a gene affect the resulting protein?**
A histogram is a type of bar graph used to show the frequency distribution of data. The independent variable is usually shown on the x-axis and the dependent variable is shown on the y-axis.

In the example below, a scientist determined the number of base pairs in different species including E. coli, baker’s yeast, an RNA retrovirus, a lily plant, a fruit fly, a frog, and a shark. She decided to compile the data into categories based on the number of organisms that had a certain number of base pairs. The histogram shows the frequency distribution of the data.

**GRAPH 1. NUMBER OF BASE PAIRS IN VARIOUS ORGANISMS**

1. **Identify**  How many species in the study had base pairs that numbered in the hundreds of millions?

2. **Synthesize**  Suppose more data have been collected since the study above was completed. There are two more species with base pairs of $10^5$ and one more species with base pairs of $10^{10}$. Construct a graph that includes the new data.
In Chapter 8, you have learned about the three-dimensional structure of DNA. In the early 1950s, several groups of researchers raced to be the first to determine the details of DNA structure. At the time, consensus was growing that the DNA molecule consisted of a fiber that was spiral shaped, and formed either a double or triple helix.

Scientists already knew DNA was a nucleic acid made up of nitrogen-containing bases, phosphates, and sugars. They just weren’t sure which of these components made up the outer strands or “chains” of the helix and which components formed the bonds connecting one chain to another. You know that James Watson and Francis Crick won the race. Their paper was published in *Nature* on April 25, 1953. Several excerpts from their historic paper follow.

**PAULING-COREY MODEL**

**Excerpt** “A structure for nucleic acid has already been proposed by Pauling and Corey. . . . Their model consists of three intertwined chains, with the phosphates near the fibre axis, and the bases on the outside.”

**Your Interpretation** On a separate piece of paper, draw a diagram showing how the different components of the Pauling-Corey model would match up between just two of the three strands (chains). Assume an equal number of phosphates and bases. Use lines to represent bonds, circles to represent phosphates, and squares to represent bases. Draw just enough to give a sense of what the internal structure might look like. For the purpose of this activity, do not worry about the placement of the sugars.

**FRASER MODEL**

**Excerpt** “Another three-chain structure has also been suggested by Fraser . . . . In his model the phosphates are on the outside and the bases on the inside, linked together by hydrogen bonds.”

**Your Interpretation** On a separate piece of paper, draw a diagram showing how the different components of the Fraser model would match up. This time use dotted or dashed lines for hydrogen bonds, otherwise use the same style you used for the Pauling-Corey Model.

**WATSON-CRICK MODEL**

The Watson and Crick model took shape after seeing Rosalind Franklin’s x-ray image of DNA and having its details interpreted for them.

**Excerpt** “We wish to put forward a radically different structure for the salt of deoxyribonucleic acid. This structure has two helical chains each coiled round the same axis . . . . the bases are on the inside of the helix and the phosphates on the outside. . . . The novel feature of the structure is the manner in which the two chains are held together by the purine and pyrimidine bases . . . . One of the pair must be a purine and the other a pyrimidine for bonding to occur. . . . In other words, if an adenine forms one member of a pair, on either chain, then on these assumptions the other member must be thymine; similarly for guanine and cytosine.

**Your Interpretation** As before use a diagram to show how the different components of the Watson-Crick model match up. Use a dotted or dashed line for the hydrogen bonds that form between the bases. This time include the detail of the base pairs.
ROSALIND FRANKLIN’S CONTRIBUTION

Rosalind Franklin’s x-ray image of DNA and her explanation of it appeared in the same issue of Nature on April 25, 1953. An excerpt is given below. What she had contributed were the physical dimensions: DNA’s density and size, and its water content. She determined that each turn of the helix is 34 angstroms long and contains 10 base pairs that are 3.4 angstroms apart and pitched at a certain angle. She calculated the diameter of the helix. Also critical to the Watson-Crick model was her conviction that, given all the physical evidence, the phosphates had to be on the outside.

**Excerpt**

“Thus, if the structure is helical, we find that the phosphate groups . . . lie on a helix of diameter about 20 A., and the sugar and base groups must accordingly be turned inwards towards the helical axis.”

Interestingly enough, Watson and Crick had been thinking along the same lines as Pauling and Corey, that the bases were on the outside, exposed and available to pass along genetic information. What Franklin understood, as a chemist, was that the hydrophilic (“water-loving”) sugar-phosphates would be on the outside of the molecule and the hydrophobic (“water-fearing”) base pairs on the inside.

1. Why does it make more sense for the hydrophilic sugar-phosphates to be on the outside of the DNA molecule and the hydrophobic nitrogenous bases on the inside? What other cellular structure do you know of that has a similar orientation?
In Chapter 2, you learned about the structure of proteins and enzymes. You learned in Chapter 6 that an allele is an alternative form of a gene and in Chapter 7 that some genes have multiple alleles. In Chapter 8 you have learned about the base sequences of DNA and how they can be altered by mutation. Now you will combine all this information to form a biochemical picture of inheritance.

ENZYMES
An enzyme is functional only when its three-dimensional structure is intact. This structure is necessary for normal function because the substrate must fit precisely into the enzyme’s active site to catalyze a reaction. Any change in an amino acid located at a critical position in the active site can make the enzyme nonfunctional.

Most metabolic processes involve a pathway of several chemical reactions, rather than a single reaction, meaning several enzymes are needed to produce the final product. Compound A may be broken down to form compound B, which forms compound C, and so on. If any one of the enzymes in the pathway becomes nonfunctional, that step is blocked and no final product can be made. In addition, the reactant in the blocked reaction accumulates in the body. Many of these intermediate compounds are toxic.

MUTATIONS
When a base in DNA undergoes a point mutation, a codon in mRNA usually changes. When a codon changes, an amino acid in a protein may change, and when the amino acid sequence of an enzyme changes, its three-dimensional structure may change. Whether the structure changes or not depends on where in the protein the altered amino acid is located and how similar the properties of the new amino acid are to those of the original amino acid. An amino acid change in a non-critical part of the enzyme may have no effect. However, the substitution of a polar amino acid for a nonpolar amino acid, or an acidic amino acid for a basic amino acid, can completely disrupt the structure of the enzyme.

ALLELES
Whenever there is a mutation that causes a change in a metabolic pathway, a new allele can result. Theoretically, any base in a gene can mutate. Why, then, doesn’t each gene have hundreds or thousands of alleles? Because only a very small minority of the bases in a gene actually code for amino acid sequence of the enzyme. For example, in one particular gene with about 90,000 bases, only 1,356 bases code for amino acids. Much of the remainder is involved in regulation or has no known function.

Most mutations result in a nonfunctional enzyme. A gene has only two alleles—one for a functioning enzyme and one for a nonfunctioning enzyme. Some genes have multiple alleles, although any individual can have only two. These alleles usually reflect amino acid changes in an enzyme that reduce the enzyme’s effectiveness but do not completely destroy its functionality.
Metabolic pathway for the breakdown of phenylalanine

Phenylalanine → 4-Hydroxyphenylpyruvic acid → Homogentisic acid
Phenylalanine hydroxylase

Tyrosine → 4-Hydroxyphenylpyruvic acid 1,2-dioxygenase
Tyrosine aminotransferase

4-Maleylacetoacetic acid → Further breakdown

ANALYSIS OF A BIOCHEMICAL PATHWAY

In 1909, the British physician Archibald Garrod coined the term *inborn error of metabolism* to describe an inherited disease called alkaptonuria. Alkaptonuria is a rare disease in which a person excretes homogentisic acid in his or her urine.

Homogentisic acid is formed from the breakdown of amino acids phenylalanine and tyrosine, which are found in most protein foods. When the enzyme that breaks down homogentisic acid is defective, the biochemical pathway is blocked. Homogentisic acid accumulates in the body until it is removed in the urine.

Examine the biochemical pathway shown below. Each step in the pathway requires a different enzyme, and each enzyme is coded by a different gene. Including alkaptonuria, there are genetic diseases caused by defective enzymes in each of the steps of this metabolic pathway.

- phenylketonuria (PKU)—phenylalanine hydroxylase is defective
- tyrosinemia type II—tyrosine aminotransferase is defective
- tyrosinemia type III—4-hydroxyphenylpyruvic acid dioxygenase is defective

These three diseases result in mental retardation and other serious health problems.

1. What compound accumulates in the blood of people who have PKU? __________
2. What compound accumulates in people who have tyrosinemia type II? __________
3. What compound accumulates in people who have tyrosinemia type III? __________
4. Which enzyme is defective in people with alkaptonuria? __________
5. Babies born in the United States are routinely tested for PKU after birth. How might the symptoms of this disease be prevented in a baby who tests positive for PKU? __________
6. Would a baby with PKU be damaged by being fed tyrosine? Explain. __________
FROM DNA TO PROTEINS

**Vocabulary Practice**

<table>
<thead>
<tr>
<th>bacteriophage</th>
<th>RNA polymerase</th>
<th>promoter</th>
</tr>
</thead>
<tbody>
<tr>
<td>nucleotide</td>
<td>messenger RNA (mRNA)</td>
<td>operon</td>
</tr>
<tr>
<td>double helix</td>
<td>ribosomal RNA (rRNA)</td>
<td>exon</td>
</tr>
<tr>
<td>base pairing rules</td>
<td>transfer RNA (tRNA)</td>
<td>intron</td>
</tr>
<tr>
<td>replication</td>
<td>translation</td>
<td>mutation</td>
</tr>
<tr>
<td>DNA polymerase</td>
<td>codon</td>
<td>point mutation</td>
</tr>
<tr>
<td>central dogma</td>
<td>stop codon</td>
<td>frameshift mutation</td>
</tr>
<tr>
<td>RNA</td>
<td>start codon</td>
<td>mutagen</td>
</tr>
<tr>
<td>transcription</td>
<td>anticodon</td>
<td></td>
</tr>
</tbody>
</table>

**A. Compound Word Puzzle**  
Read the phrase and write the word that it most closely describes. Then write another phrase that describes the same word in a different way.

<table>
<thead>
<tr>
<th>PHRASE 1</th>
<th>WORD</th>
<th>PHRASE 2</th>
</tr>
</thead>
<tbody>
<tr>
<td>error that throws off the reading frame of an mRNA sequence</td>
<td>Example</td>
<td>frameshift mutation caused by insertion or deletion of nucleotides</td>
</tr>
<tr>
<td>explains Chargaff’s rules</td>
<td>1.</td>
<td></td>
</tr>
<tr>
<td>states that genetic information flows in one direction</td>
<td>2.</td>
<td></td>
</tr>
<tr>
<td>pairs with an mRNA codon during translation</td>
<td>3.</td>
<td></td>
</tr>
<tr>
<td>an intervening sequence</td>
<td>4.</td>
<td></td>
</tr>
<tr>
<td>the type of RNA that is converted to a protein during translation</td>
<td>5.</td>
<td></td>
</tr>
</tbody>
</table>
### VOCABULARY PRACTICE, CONTINUED

<table>
<thead>
<tr>
<th>PHRASE 1</th>
<th>WORD</th>
<th>PHRASE 2</th>
</tr>
</thead>
<tbody>
<tr>
<td>a change in an organism’s DNA</td>
<td><strong>6.</strong></td>
<td></td>
</tr>
<tr>
<td>monomer that makes up nucleic acids</td>
<td><strong>7.</strong></td>
<td></td>
</tr>
<tr>
<td>a sequence of mRNA that is expressed after processing</td>
<td><strong>8.</strong></td>
<td></td>
</tr>
</tbody>
</table>

### B. Find the Odd Word
Put a checkmark next to the word that does not belong.

1. __________ mutagen
   - Explanation

2. __________ codon
   - Explanation

3. __________ central dogma
   - Explanation

4. __________ codon
   - Explanation

5. __________ rRNA
   - Explanation

6. __________ tRNA
   - Explanation

7. __________ mRNA
   - Explanation

8. __________ replication
   - Explanation

9. __________ mutagen
   - Explanation

10. __________ mutation
    - Explanation

11. __________ double helix
    - Explanation

12. __________ frameshift mutation
    - Explanation

13. __________ bacteriophage
    - Explanation

14. __________ RNA polymerase
    - Explanation

15. __________ transcription
    - Explanation
C. Secret Message  Next to each definition, fill in the blanks with the vocabulary word that best fits each description. When complete, write the boxed letters in order in the blanks at the bottom of the page to answer the clue.

1. large enzyme that initiates transcription
   ________________

2. caused by the insertion or deletion of nucleotides in DNA
   ________________

3. spliced together during mRNA processing
   ____________

4. part of a ribosome; catalyzes the formation of peptide bonds between amino acids
   ____________

5. a change in a single nucleotide in DNA
   ____________

6. examples include radiation and UV light
   ____________

7. made up of a sugar, a phosphate group, and a nitrogen-containing base
   ________________

8. the part of the central dogma that occurs in the cytoplasm of eukaryotic cells
   ________________

9. Fill in the blanks with the boxed letters from above to name a region of DNA where RNA polymerase binds:
   ____________
D. DNA Adventure! Solve the clues by filling in the words in the numbered squares.

**Across**
1. a change in a single nucleotide in DNA
2. the process that makes a polypeptide
3. the process of making a copy of DNA
5. component of ribosomes
6. process that involves RNA polymerase
7. sequence in mRNA that is not expressed as protein
8. three-dimensional model developed by Watson and Crick
9. the rules that explain how nucleotides interact with each other

**Down**
1. a change in a single nucleotide in DNA
2. the process of making a copy of DNA
3. the process of making a copy of DNA
4. complementary to an mRNA codon
5. carries an amino acid from the cytoplasm to a ribosome
6. major enzyme involved in replication
7. describes the flow of genetic information
8. used by Hershey and Chase in their experiments
9. may be induced by mutagens
10. halts translation
11. helps RNA polymerase recognize the start of a gene

---

**Clues:**
1. a change in a single nucleotide in DNA
2. the process that makes a polypeptide
5. component of ribosomes
12. process that involves RNA polymerase
15. sequence in mRNA that is not expressed as protein
16. three-dimensional model developed by Watson and Crick
17. the rules that explain how nucleotides interact with each other

---

**Grid:**

```
Across:
2. [ ] [ ] [ ] [ ]
4. [ ] [ ] [ ] [ ]
5. [ ] [ ] [ ]
12. [ ] [ ] [ ]
15. [ ] [ ] [ ]
16. [ ] [ ] [ ]
17. [ ] [ ] [ ]

Down:
1. [ ] [ ] [ ]
3. [ ] [ ] [ ]
6. [ ] [ ] [ ]
7. [ ] [ ] [ ]
8. [ ] [ ] [ ]
9. [ ] [ ] [ ]
10. [ ] [ ] [ ]
11. [ ] [ ] [ ]
14. [ ] [ ] [ ]
15. [ ] [ ] [ ]
16. [ ] [ ] [ ]
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