

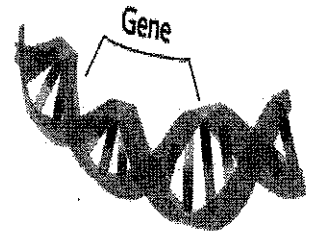
Genes and Gene Mutations

Reflect

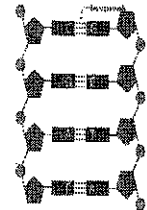
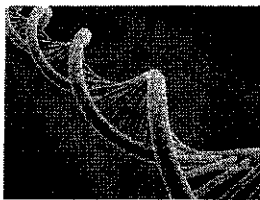
Genes carry the genetic information that determines the traits inherited from parent to offspring, such as height, eye color, and hair color. **Genes** are composed of the DNA located on chromosomes. Each person has two copies of each gene, one copy from the mother and the other copy from the father. DNA in the nucleus of the cell contains the coded instructions to make the proteins that play critical roles in the body. Proteins provide structure and support for cells; they work as enzymes and regulate the body's tissues and organs.

How Genes Produce Proteins

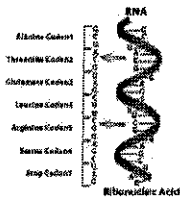
DNA is comprised of building blocks known as nucleotides, which are made up of a sugar, a phosphate, and a base group. The nucleotides are identified by the bases adenine (A), thymine (T), cytosine (C), and guanine (G). These bases pair together to form a double-stranded DNA molecule. Adenine pairs with thymine. Cytosine pairs with guanine.



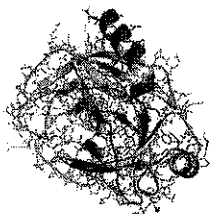
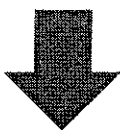
gene:
a piece of the genetic material that determines traits inherited from parent to offspring



Base pairs



The DNA in the nucleus of a cell is used to make RNA. The order of the nucleotides in DNA is the code that carries information about which proteins the cells should build. During a process known as **replication**, cells make copies of DNA molecules before they divide to form new cells. During normal cellular functioning, the information in DNA is copied to a molecule called ribonucleic acid, or RNA.



A type of RNA called messenger RNA carries the information copied from DNA in the nucleus to the ribosomes located in the cytoplasm of the cell. This process is known as **transcription**. At the ribosome, the RNA's message is translated into a protein. The process of making proteins from RNA is called **translation**. When RNA is used to build a protein, the specific sequence of bases encodes a specific protein. If the sequence of bases is transcribed or translated incorrectly, the protein may not function properly. The types of proteins your body makes determine your traits.

replication:
when a cell makes a copy of DNA

transcription:
when a cell makes a copy of RNA from DNA

translation:
when protein is made from RNA

Genes and Gene Mutations

Reflect

Controlling Genes

Cells are able to control genes by turning genes on and off. This process is known as **gene expression**. Genes are turned on and off in different arrangements to make a stomach cell function differently from a muscle cell or a brain cell.

Gene Mutations

A **gene mutation** is a permanent change in a gene or chromosome of a cell. The mutation can be a change in the order of bases, number of bases, or types of bases. Mutations may be harmful, beneficial, or have no effect on an organism at all. Mistakes can happen when DNA is copied during replication, causing proteins to not work properly. When this happens, it can affect the structure, function, and traits of organisms. Mutations that cause proteins to fail can lead to serious medical conditions or diseases.

Mutations can be caused by harmful chemical agents, x-rays, high temperatures, and viruses. They can result in altered genes and even extra or missing whole chromosomes. A physical or chemical agent that can cause a change in DNA is a **mutagen**.

Mutations that occur in genes can cause conditions known as **genetic disorders**. Some of the more common genetic disorders caused by gene mutations include cystic fibrosis, sickle-cell anemia, and Tay-Sachs disease.

On the other hand, some mutations can be beneficial.

protein:
provides structure and support for cells; works as enzymes and regulates the body's tissues and organs

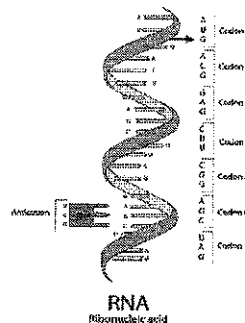
gene expression:
the ability of cells to turn genes on and off



mutation:
a permanent change in DNA

mutagen:
a physical or chemical agent that can cause a change in DNA

genetic disorders:
conditions caused by mutations that occur in genes



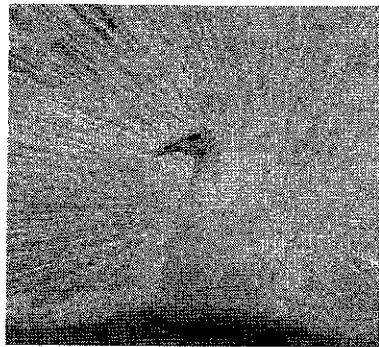
Genes and Gene Mutations

Look Out!

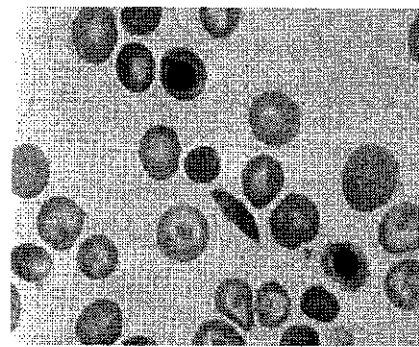
Type of Mutations

There are several different types of mutations. Recall that during translation, when RNA is used to build a protein, the specific sequence of bases encodes a specific protein. If the sequence of bases is incorrect, the protein may not function properly. A common type of mutation called a substitution is when one base replaces another. Another type of mutation called an insertion is when an extra base is added. When a base is left out, this type of mutation is called a deletion.

Below is a picture of an albino peacock and a picture of the red blood cells of a person with sickle-cell anemia. Both are examples of inherited genetic disorders caused by a mutation.



Albino peacock

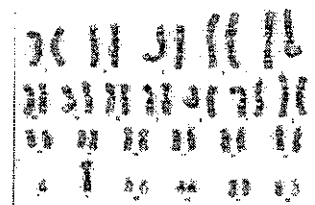
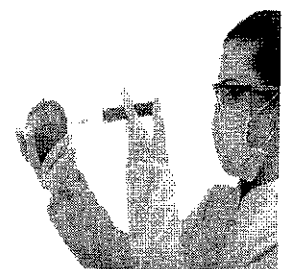


Sickled cells of a patient with the genetic disorder sickle-cell anemia

What Do You Think?

Career Corner: Research Geneticist

If you are interested in helping to find causes of diseases and looking for ways to treat and prevent diseases, you may be interested in becoming a research geneticist. Geneticists are biologists who study genes and heredity. They determine the causes of diseases, genetic variations, and chromosomal abnormalities. As a geneticist, you can have a successful career working in a wide range of industries, including agriculture and wildlife, environmental science, forensics, and medicine.



Genes and Gene Mutations

Try Now

What Do You Know?

Match the conditions described below to the inherited genetic disorder. Write your answers on the right side of the chart.

Inherited Genetic Conditions

Genetic Disorders	Function Affected
Sickle-cell anemia	
Cystic fibrosis	
Tay-Sachs disease	
Colorblindness	
Albinism	

- A condition where a person has partial or complete loss of pigmentation of the skin, eyes, and hair
- A condition where the lungs and digestive system become clogged with thick sticky mucus
- A condition that causes progressive damage to the nervous system
- A condition that affects the perception of color
- A condition where red blood cells, which carry oxygen around the body, develop abnormally

Genes and Gene Mutations

Connecting With Your Child

Take some time to explore and answer the questions below. Use technology to conduct some research if necessary.

1. Name the bases found in a strand of DNA.
2. When are mutations most likely to occur?
3. Is the stored genetic information used to build proteins from DNA or RNA?
4. How can a mutation in the DNA affect what proteins are made by cells?
5. Is cancer another example of a mutation that can occur in genetic material?
6. What is the Human Genome Project (HGP)? What advancements has the project made to modern-day science?