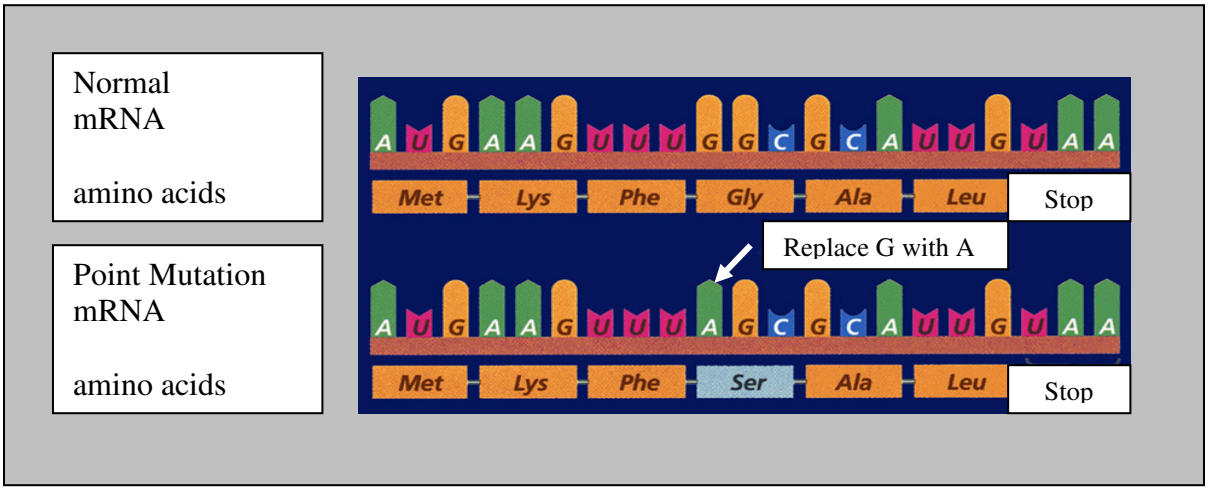
Mutations

A section of DNA on a chromosome that directs the making of a specific protein is called a **gene**. Genes control the traits inherited by an organism. If a change occurs in the sequence of nitrogen bases, the trait may be changed. Human DNA is made up of about 6 billion base pairs, so it is not surprising that sometimes errors occur when DNA is copied or when cells divide.

Any permanent change in the sequence of nucleotides is called a **mutation**. Some mutations, like flower color in plants, are not harmful, but some mutations can cause disease. For example, sickle cell anemia and cystic fibrosis are diseases caused by mutations.

Although many mutations are harmful, they also add genetic diversity to a species. Also, if a mutation causes a change that is beneficial (helpful), the individuals with the helpful mutation will have an advantage over organisms without the mutation.

Example of a mutation:



If a different nucleotide replaces one that was originally present, it is called a **substitution** mutation. If a base is added, it is called an **insertion** mutation. If a base is deleted, it is called a **deletion** mutation.

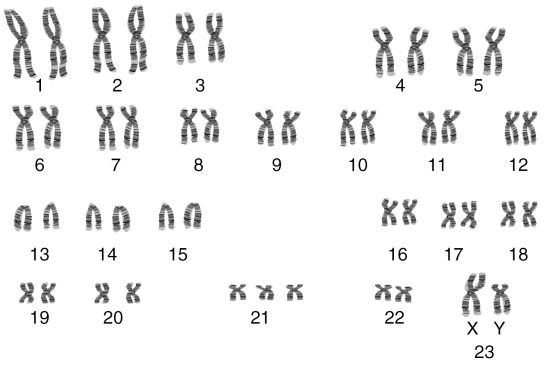
Mutations can also occur at the chromosomal level. For example, a piece of chromosome may break off.

******Any change in DNA or RNA can change which proteins are made, which can change how our cells look and how our cells work******

Causes of mutations

Mutations are usually random events. Some mutations are caused by **mutagens**- substances in the environment like ultraviolet light, X rays, radioactive substances, cigarette smoke, and pollutants. If mutations occur in body (somatic) cells, they can cause cancer. Only changes in gametes (eggs and sperm) are passed on to offspring.

The diagram to the right is a **karyotype**- a picture of chromosomes. Humans normally have 23 pairs of chromosome, but the individual whose chromosomes are shown here has an extra chromosome 21. This condition is called Down's Syndrome, and it is caused by **nondisjuction** (chromosomes separating incorrectly) during meiosis.



Practice

1. What is nondisjunction?
   A. Failure of genes to be passed on to future generations
   B. A mutation caused by the environment
   C. Failure of chromosomes to separate properly
   D. A duplication of genes on a chromosome

2. Mutations in DNA molecules can occur when —
   A. replication of DNA is exact
   B. a DNA enzyme attaches to an RNA codon
   C. RNA codons are replaced by DNA nucleotides
   D. a change occurs in DNA nucleotide bases

Codon Chart

First Position (5′)	Second Position: U	Second Position: C	Second Position: A	Second Position: G	Third Position (3′)
U	Phenylalanine	Serine	Tyrosine	Cysteine	U
	Phenylalanine	Serine	Tyrosine	Cysteine	C
	Leucine	Serine	Stop	Stop	A
	Leucine	Serine	Stop	Tryptophan	G
C	Leucine	Proline	Histidine	Arginine	U
	Leucine	Proline	Histidine	Arginine	C
	Leucine	Proline	Glutamine	Arginine	A
	Leucine	Proline	Glutamine	Arginine	G
A	Isoleucine	Threonine	Asparagine	Serine	U
	Isoleucine	Threonine	Asparagine	Serine	C
	Isoleucine	Threonine	Lysine	Arginine	A
	Methionine	Threonine	Lysine	Arginine	G
G	Valine	Alanine	Aspartic acid	Glycine	U
	Valine	Alanine	Aspartic acid	Glycine	C
	Valine	Alanine	Glutamic acid	Glycine	A
	Valine	Alanine	Glutamic acid	Glycine	G

3. Which of the following changes would be expected in the amino acid chain if the following mutation occurred? *(use the codon chart above)*

ACG GUC CCG ⟶ ACG GCC CCG

A. The amino acid sequence would be shorter than expected.
B. The identity of one amino acid would change.
C. The amino acid sequence would remain unchanged.
D. The identities of more than one amino acid would change.

4. Which part of DNA provides instructions for how to produce a protein?
   A. The sequence of nitrogen bases
   B. The sugar molecules
   C. The phosphate groups
   D. The bonds that hold the sugar and bases together

5. Substitutions, insertions, and deletions are known as "point" mutations because they
   A. change the genetic information of DNA
   B. affect the structure of a chromosome
   C. involve only a single nucleotide
   D. change the amino acid that is inserted into a protein

6. Which of the following can be a mutagen?
   A. an inversion
   B. a toxic chemical
   C. a nucleotide
   D. a random mutation

7. The DNA sequence TTT codes for the amino acid lysine. The DNA sequence TTC also codes for lysine. What can you conclude from this information?
   A. a deletion in the genetic instructions may not be significant
   B. adding a nucleotide to a genetic sequence has no effect
   C. any substitution that changes the genetic code affects a trait
   D. this kind of substitution in the genetic code may go unnoticed

8. Which two types of biomolecules are directly affected by a mutation?
   A. nucleic acids and lipids
   B. nucleic acids and carbohydrates
   C. proteins and nucleic acids
   D. proteins and carbohydrates

9. Any permanent change in a gene is called a _____.

10. When can a mutation be passed on to offspring?