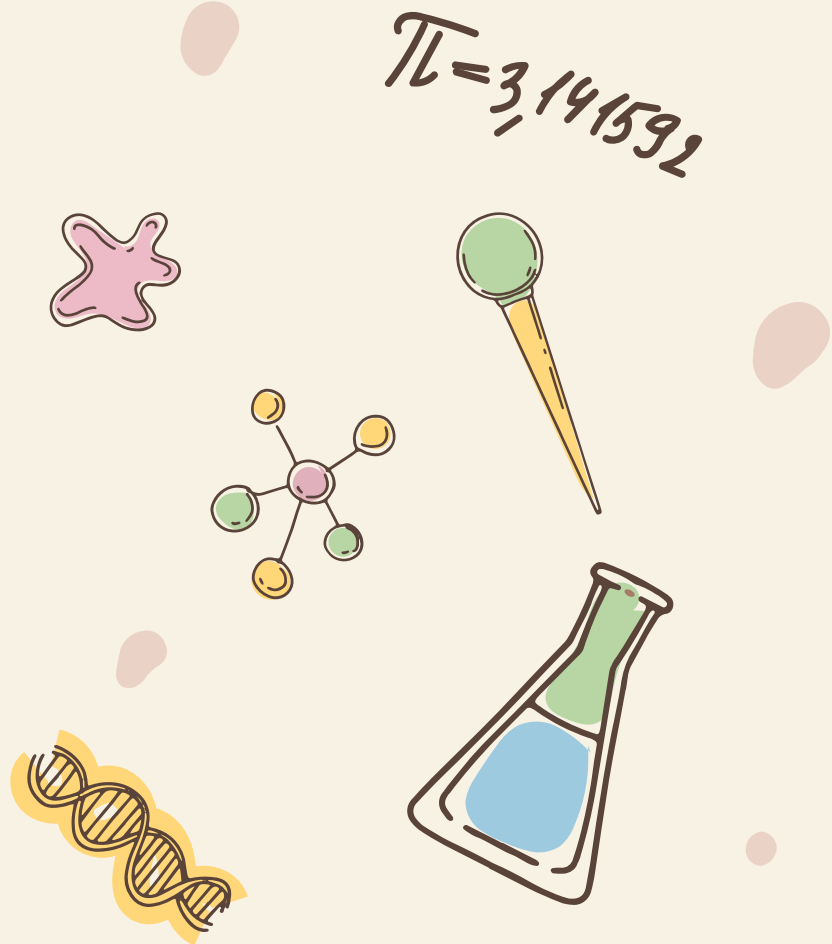# Biology - DNA Mutations
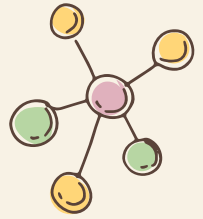
Science 10

Lesson Overview

01
Warm Up Activity

02
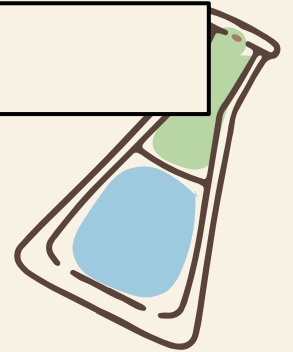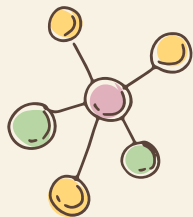Introduction to Genetic Variations

03
Mutations & Types

04
Review & Practice

Warm Up Activity: Telephone

Going down each row we are going to play a game of "telephone".
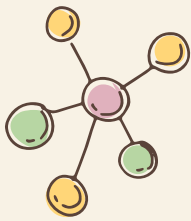
1. The person at the start of the row will write down a sentence and then say it to the next person.
2. Person 2 will then repeat what they heard to person 3 going down the line.
3. The last person in the group will write down what they heard.
4. REPEAT!

After everyone is done, will discuss the results!

How did the messages change?
Does the new message make sense?

DNA Variations

- DNA replicates with an error rate of about 1 in every 1 billion base pairs (1×10^9).

- The human genome is about 3 billion base pairs long. In theory, that means that there could be three errors per DNA replication cycle.

- These errors lead to **DNA mutations** which can cause significant problems, diseases or changes to the organism (or not!).
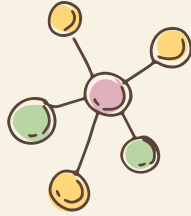
DNA Mutations

A mutation is a permanent alteration or change in the DNA sequence that makes up a gene.

Mutations can range in size; they can affect anywhere from a single base pair to an entire chromosome that includes multiple genes.

Types of Mutations

There are many different ways that DNA can be changed, resulting in different types of mutations. Here is a quick summary of these types.
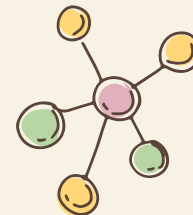
1. **Substitution**
   a. **Missense**
   b. **Silent**
   c. **Nonsense**
2. **Insertion**
3. **Deletion**
4. **Frameshift**

Normal DNA:	ATG	TGT	GTT	TAT	GGA…
Normal Protein:	Met	Cys	Val	Tyr	Gly…
Silent:	ATG	TGC	GTT	TAT	GGA…
New Protein:	Met	Cys	Val	Tyr	Gly…
Missense:	ATG	TGG	GTT	TAT	GGA…
New Protein:	Met	Trp	Val	Tyr	Gly…
Insertion:	ATG	TGG	TGT	TTA	TGG A…
New Protein:	Met	Trp	Cys	Leu	Trp…
Deletion:	ATG	TG-G	TTT	ATG	GA…
New Protein:	Met	Trp	Phe	Met…	
Nonsense:	ATG	TGA	GTT	TAT	GGA…
New Protein:	Met	STOP			

Substitution Mutations

Substitution is a mutation that exchanges one base for another (i.e., a change in a single "chemical letter" such as switching an A to a G).

1. **Substitution**
   a. **Missense**
   b. **Silent**
   c. **Nonsense**
2. **Insertion**
3. **Deletion**
4. **Frameshift**

Example:

Normal RNA: GACCUC
Mutated RNA: GACCCC

CTGGAG
CTGGGG

Substitution Mutations

Substitution is a mutation that exchanges one base for another (i.e., a change in a single "chemical letter" such as switching an A to a G).
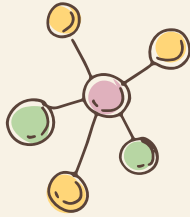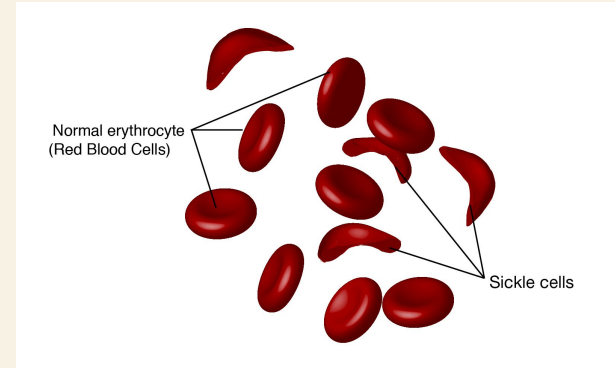
1. **Substitution**
   a. **Missense**
   b. **Silent**
   c. **Nonsense**
2. **Insertion**
3. **Deletion**
4. **Frameshift**

NORMAL

DNA   G A G
       C T C

MUTATION →

SICKLE CELL

       G T G
       C A C

RNA   G A G   →   G U G

PROTEIN GLU   VAL

NORMAL PROTEIN   MUTANT PROTEIN

Normal erythrocyte (Red Blood Cells)
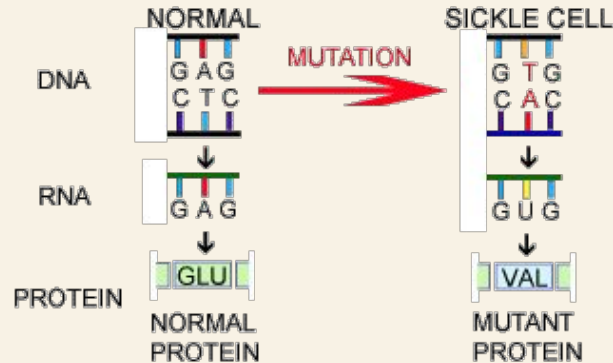
Sickle cells

Sickle Cell Anemia: Is cause by a substitution in the beta-hemoglobin gene, which alters Adenine to Thymine.
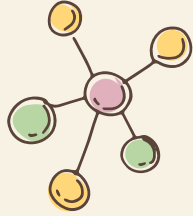
Insertion Mutations

Insertions are mutations in which extra base pairs are **inserted** into a new place in the DNA.

1. **Substitution**
   a. **Missense**
   b. **Silent**
   c. **Nonsense**
2. **Insertion**
3. **Deletion**
4. **Frameshift**

Example:

Normal RNA: GACCUC
Mutated RNA: GACCACCUC

Normal Protein: Aspartic Acid—Leucine
Mutated Protein: Aspartic Acid—Histidine—Leucine

CTGGAG
CTGGTGGAG

Deletion Mutations

Deletions are mutations in which a section of DNA is **lost**, or **deleted**.

1. **Substitution**
   a. **Missense**
   b. **Silent**
   c. **Nonsense**
2. **Insertion**
3. **Deletion**
4. **Frameshift**

Example:

Normal RNA: GACCUC
Mutated RNA: GAUC

Normal Protein: Aspartic Acid—Leucine
Mutated Protein: Aspartic Acid—...

CTGGAG
CTAG

Frameshift Mutations

Since protein-coding DNA is dividing into codons of three bases long, insertions and deletions can alter a gene so that its message is no longer read correctly. These mutations are called **frameshifts.**
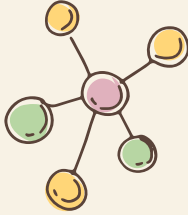
1. **Substitution**
   a. **Missense**
   b. **Silent**
   c. **Nonsense**
2. **Insertion**
3. **Deletion**
4. **Frameshift**

Frameshift Mutations

For example, consider the sentence: "The fat cat sat." Each word is three letters long similar to how we would read a set of codons. If we delete the first letter and decode the sentence the same way it does not make sense.

1. **Substitution**
   a. **Missense**
   b. **Silent**
   c. **Nonsense**
2. **Insertion**
3. **Deletion**
4. **Frameshift**

Example:

~~T~~he fat cat sat
hef atc ats at...

~~T~~he fat cat sat
hef atc ats at

Missense Mutations

A substitution mutation is called a **missense mutation** if the substitution **changes** the amino acid.

1. **Substitution**
   a. **Missense**
   b. **Silent**
   c. **Nonsense**
2. **Insertion**
3. **Deletion**
4. **Frameshift**

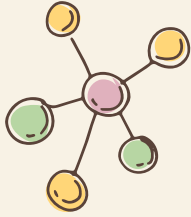**Example:**

Normal RNA: GACCUC
Mutated RNA: GACCCC

Normal Protein: Aspartic Acid—Leucine
Mutated Protein: Aspartic Acid—Proline

Silent Mutations

A substitution mutation is called a **silent mutation** if the substitution **does not change** the amino acid.

1. **Substitution**
   a. **Missense**
   b. **Silent**
   c. **Nonsense**
2. **Insertion**
3. **Deletion**
4. **Frameshift**

Example:

Normal RNA: GACCUC
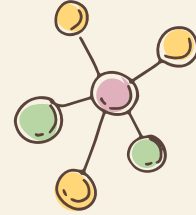**Mutated RNA:**   GACCUA

Normal Protein: Aspartic Acid—Leucine
Mutated Protein: Aspartic Acid—Leucine

Nonsense Mutations

A substitution mutation is called a **nonsense mutation** if the substitution **changes** the amino acid into a **"stop"**.

1. **Substitution**
   a. **Missense**
   b. **Silent**
   c. **Nonsense**
2. **Insertion**
3. **Deletion**
4. **Frameshift**

Example:

Normal RNA: GAC CUC
Mutated RNA: GAC UGA

Normal Protein: Aspartic Acid—Leucine
Mutated Protein: Aspartic Acid—STOP

DNA Mutations