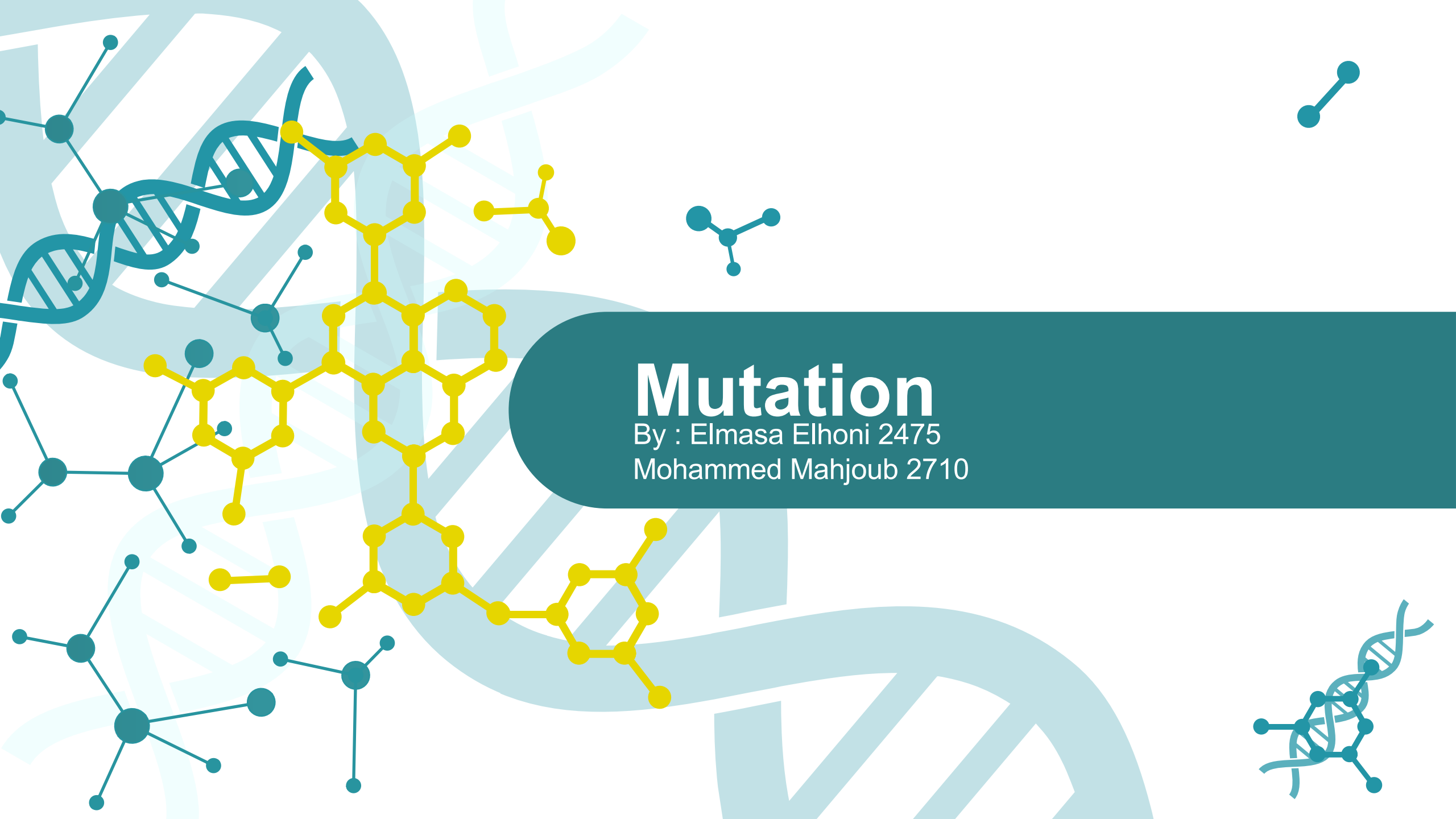

Mutation

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Objectives

1 Define DNA mutation ?

2 Describe causes of DNA mutation

3 Identify mutation analogy and types

4 List diseases liked to DNA mutation

5 Summary

6 Reference

What is DNA MUTATION ?

DNA mutation is a permanent alteration in the DNA sequence that makes up a **gene**, such that the sequence differs from what is found in most people. **Mutations** range in size; they can affect anywhere from a single DNA building block (base pair) to a large segment of a chromosome that includes multiple **genes**

What are the causes of DNA mutation ?

mutation can be spontaneous or acquired :

Spontaneous mutation :

- Heritable mutation are passed down from parents to offspring
- Spontaneous chemical changes like :

Tautomerism : a base is changed by the repositioning of a hydrogen atom

Depurination : loss of a purine base (A or G)

Acquired mutation :

is a result from exposure to an environmental factors like drugs , radiation , ultra violate light from the sun , chemicals and smoking

Acquired causes

Sunlight

Increased exposure to sunlight leading to thymine dimers in DNA

X-ray radiation

- Multiple X-rays
- Close proximity to X-rays

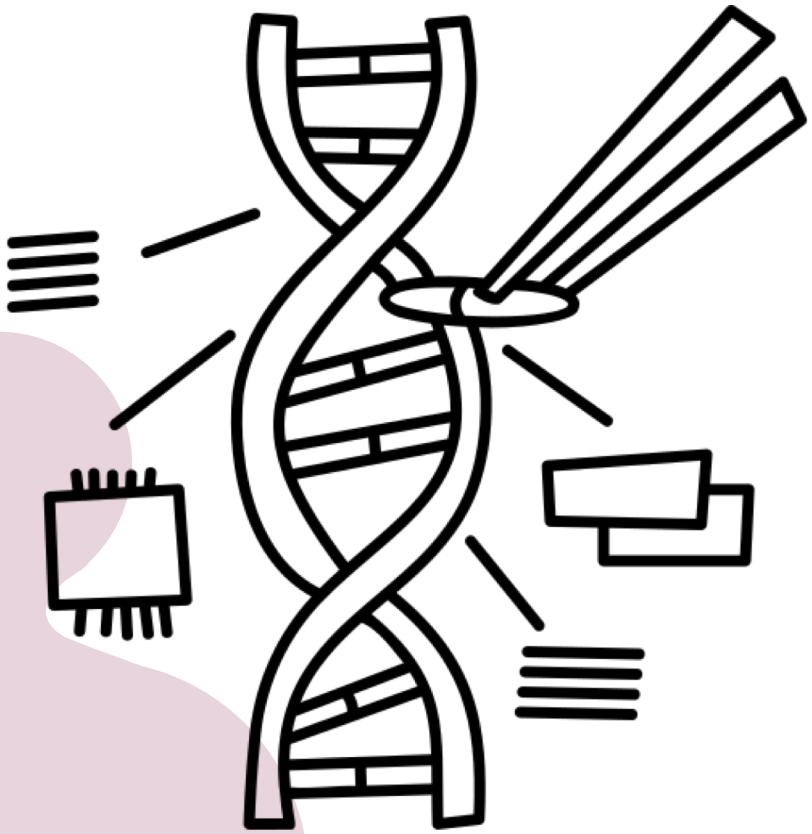
Tobacco products

- Smoking
- Chewing tobacco
- Second-hand smoke

Chemicals

Increased exposure to carcinogenic/mutagenic chemicals

Mutation Analogy and types of mutation

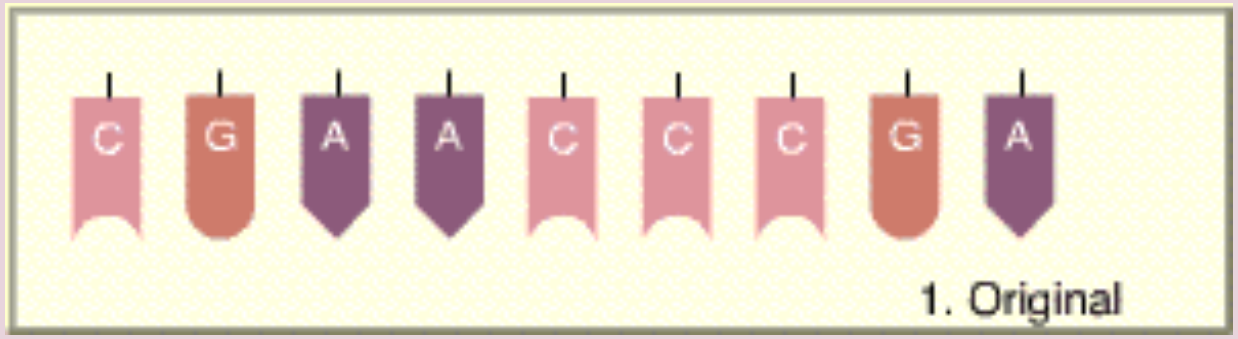


Analogy

Much like reading text, the DNA sequence is "read" by messenger RNA to produce a "story" or an amino acid chain that will be used to make a protein. Since each codon is three letters long, let's see what happens when a "mutation" occurs in a sentence that uses only three-letter words.

“THE RED CAT ATE THE RAT “

TYPES



base-pair substitution :

base pair substitutions A type of mutation involving replacement or substitution of a single nucleotide base with another in DNA or RNA molecule.

Base-pair deletion :

occurs when a nitrogen base is taken out of the sequence. Again, this causes the entire reading frame to change.

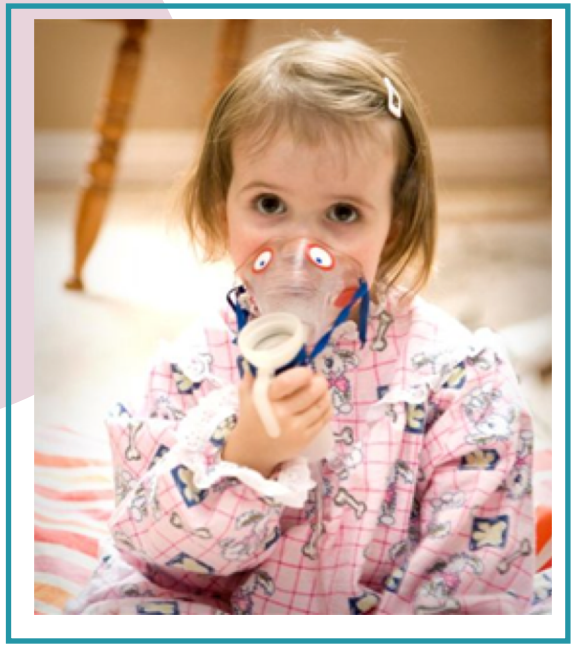
Base-Pair Insertions :

As the name implies, an insertion occurs when a single nitrogen base is accidentally added in the middle of a sequence.

Base-pair inversion :

in this type a chromosome rearrangement in which a segment of a chromosome is reversed end to end

Disease Linked to D N A mutation

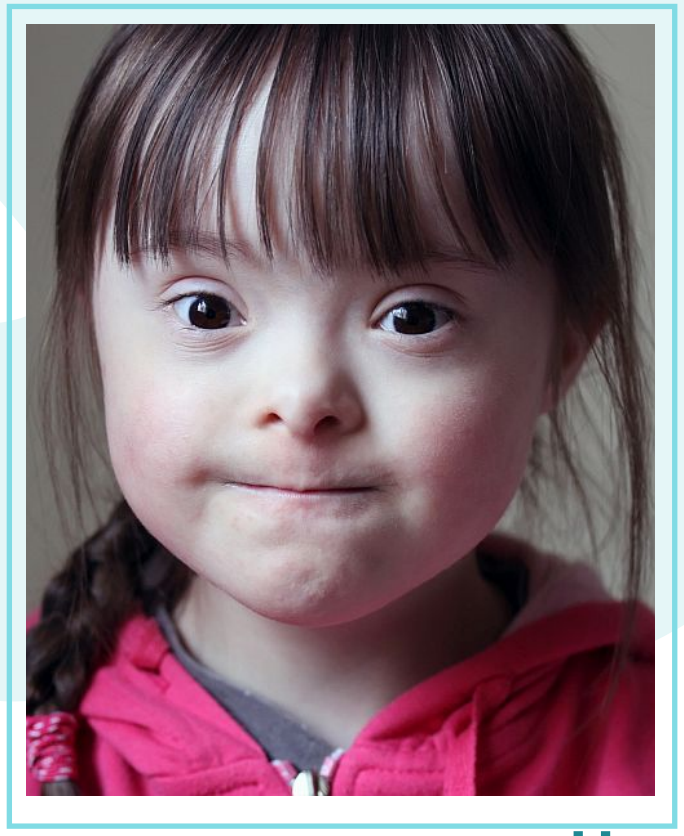


Cystic fibrosis

A genetic disorder, in which the lungs and the digestive system get clogged with mucus This is the most common mutation, ΔF508, is a deletion (Δ signifying deletion)

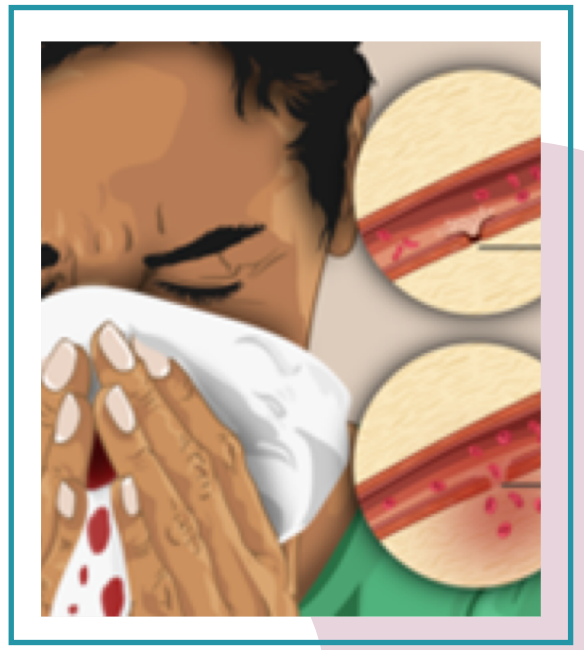
down syndrome

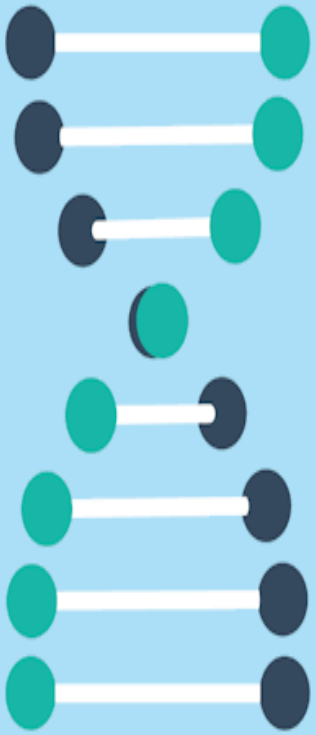
A genetic condition in which a person has an extra chromosome cused by a random error in cell division that results in the presence of an extra copy of chromosome 21



Hemophilia

An inherited disorder in which the blood does not clot due to insufficient clotting factors.





Summary

1. Mutations describe changes to a DNA sequence and are associated with both disease and evolution
2. Mutations can occur spontaneously in all cell types like inheritance or induced by the exposure to chemicals, environmental factors, or mistakes in DNA replication machinery
3. Mutations that can happen are base-pair substitution , base-pair addition , base pair-deletion
4. Mutations can cause a lot of disease like sickle cell disease and down syndrome

Reference

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Thank
you