DNA MUTATION

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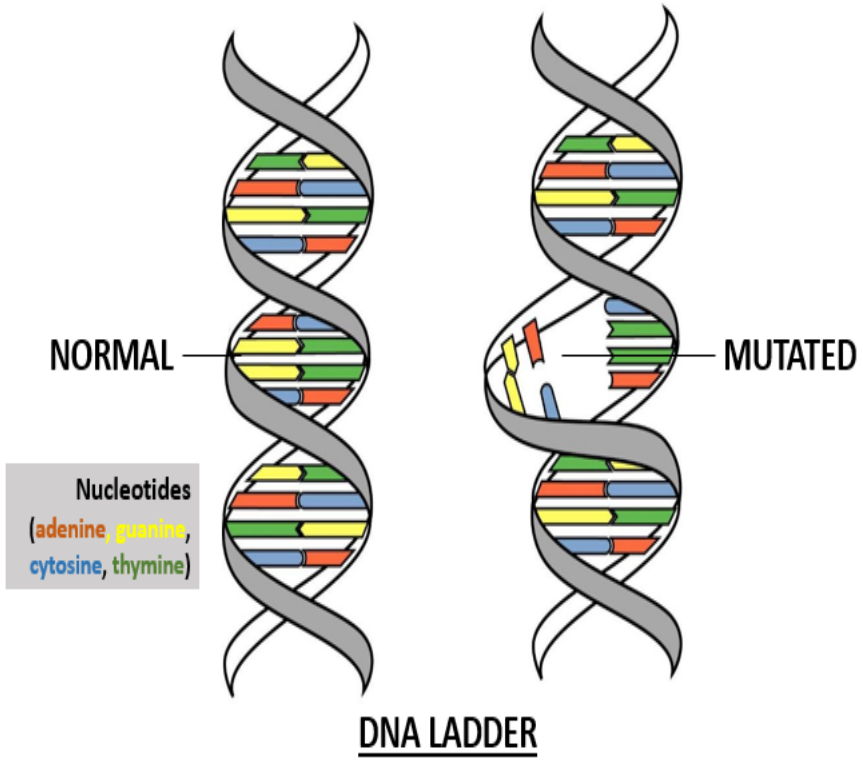
OBJECTIVES

- ❑ Define Mutation.
- ❑ Differentiate Between Somatic & Germline Mutations.
- ❑ Classify Causes of Mutation.
- ❑ Classify Types of Mutation.
- ❑ Describe Examples of Diseases Caused by Mutations.
- ❑ Conclusion

DEFINE MUTATION



A mutation is a change in DNA, the hereditary material of life.

An organism's DNA affects how it looks, how it behaves, and its physiology.

So a change in an organism's DNA can cause changes in all aspects of its life.

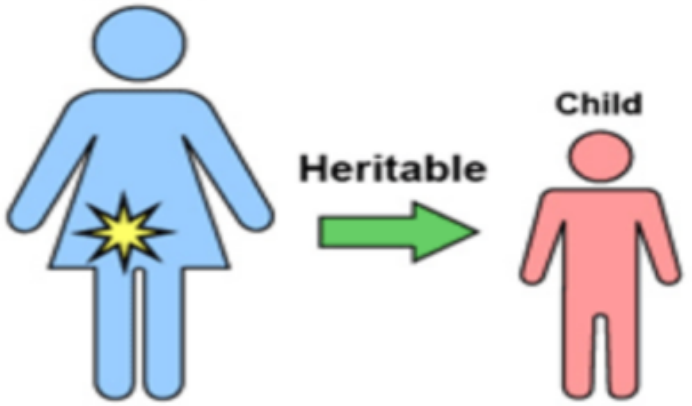
DIFFERENTIATE BETWEEN SOMATIC & GERMLINE MUTATIONS

Somatic mutations
- Occur in ***nongermline*** tissues
- Cannot be inherited

Nonheritable

Mutation in tumor only
(for example, breast)

Germline mutations
- Present in egg or sperm
- Can be inherited
- Cause cancer family syndrome



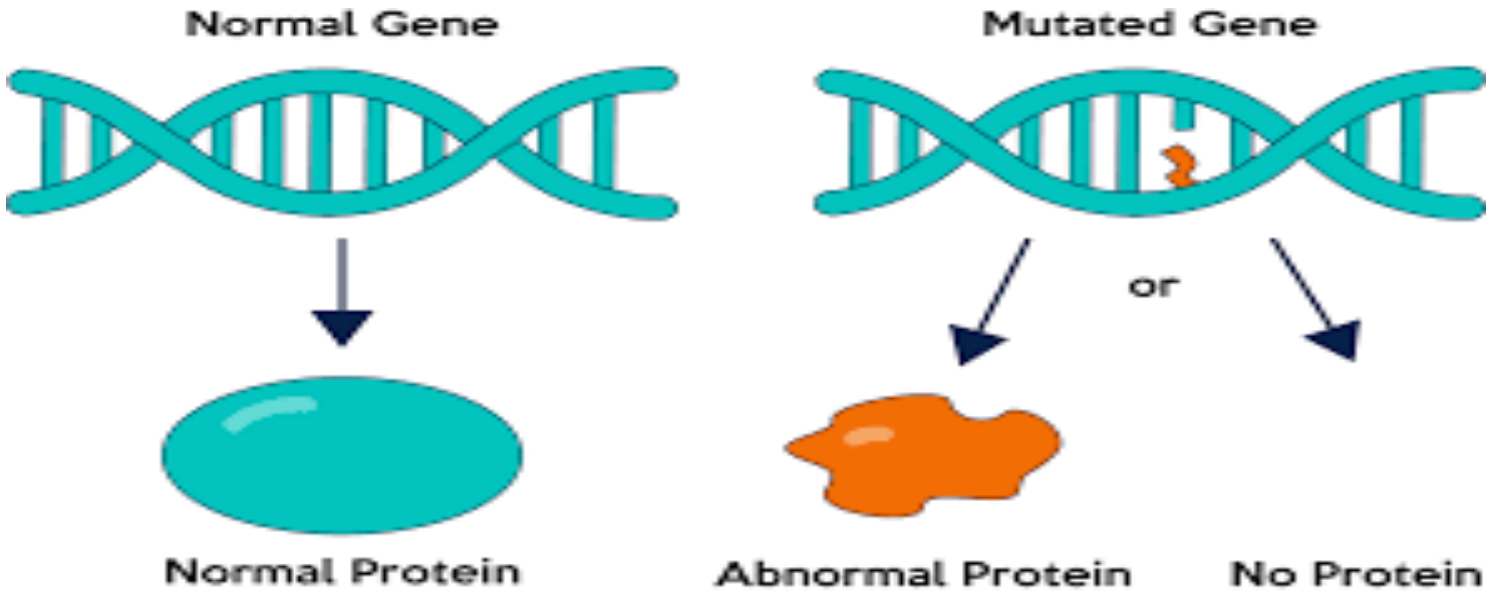
Mutation in
egg or sperm

All cells
affected in
offspring

CLASSIFY CAUSES OF MUTATION

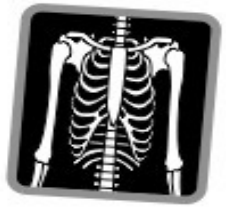
- **Spontaneous:**

  Spontaneous mutations are the result of errors in natural biological processes, DNA fails to copy accurately.



❑ Induced:

Induced mutations are due to agents in the environment that cause changes in DNA structure.

Radiation		Chemicals			Infectious Agents	
						
UV *(from sunlight)*	X-rays *(medical uses)*	Carcinogens *(e.g. cigarettes)*	Processed foods & preservatives	Cosmetics & cleaning products	Viruses *(e.g. HPV)*	Bacteria (e.g. *Helicobacter*)

CLASSIFY TYPES OF MUTATION

Substitution Mutation

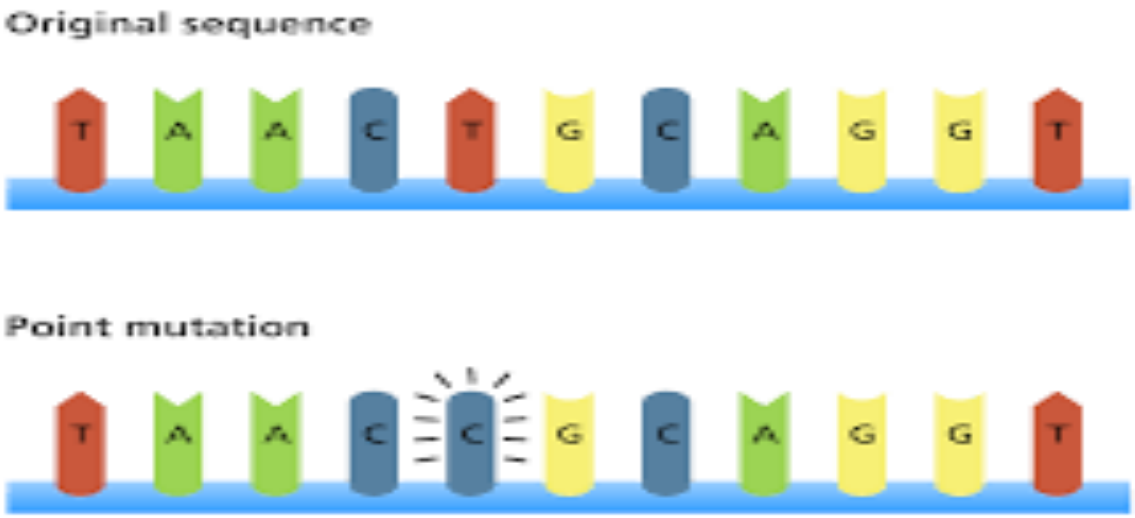
- **Silent Mutation**
- **Missense Mutation**
- **Nonsense Mutation**

Frameshift Mutation

- **Insertion Mutation**
- **Deletion Mutation**

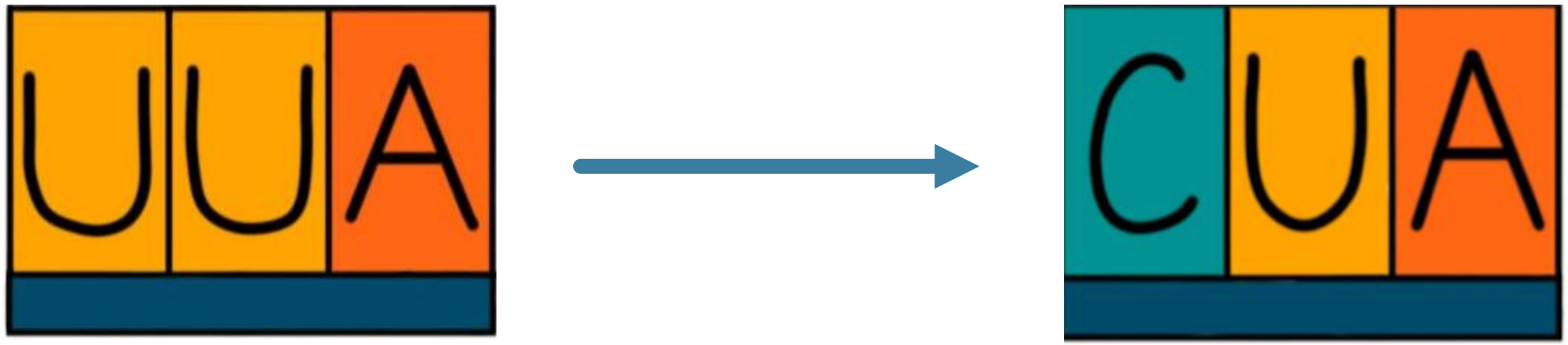
❑ Substitution Mutation

A substitution is a mutation that exchanges one base for another.



- **Types of Substitution Mutations:**
  - **Silent Mutation:**

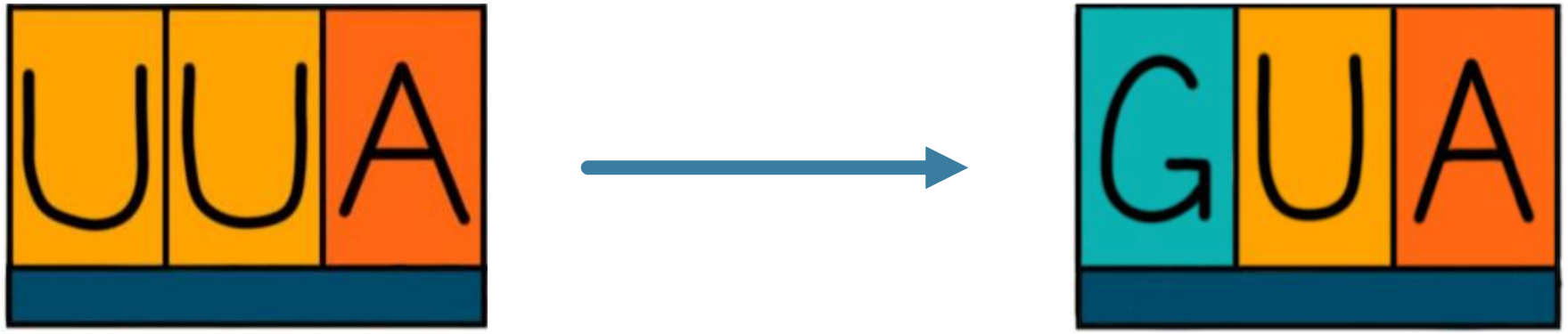
    Change a codon to one that encodes the same amino acid and causes no change in the protein produced.



Leucine **Leucine**

- **Missense Mutation:**

  Changes a codon to one that encodes a different amino acid and cause a small change in the protein produced.



Leucine **Valine**

- **Nonsense Mutation:**

  Changes an amino-acid-coding codon to a single "stop" codon and cause an incomplete protein.

  This can have serious effects since the incomplete protein probably won't function.

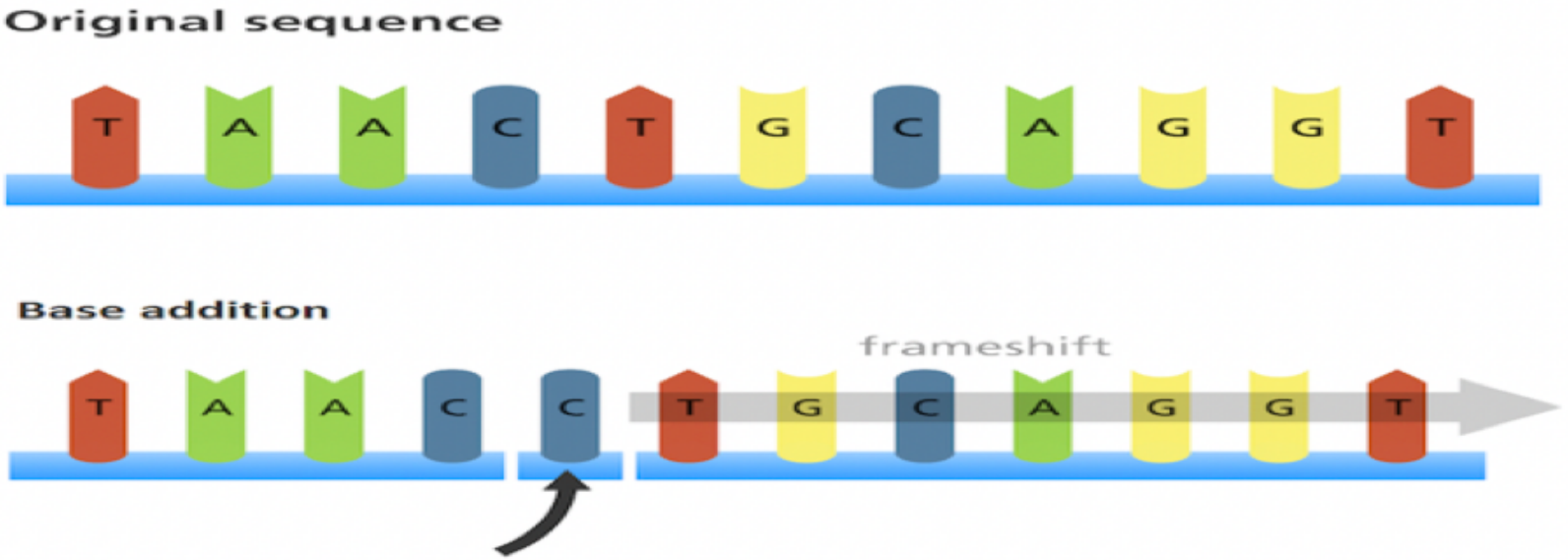
Leucine **Stop Codon**

❑ Frameshift Mutation:

Shifts the way the sequence is read by adding or removing a DNA sequence.

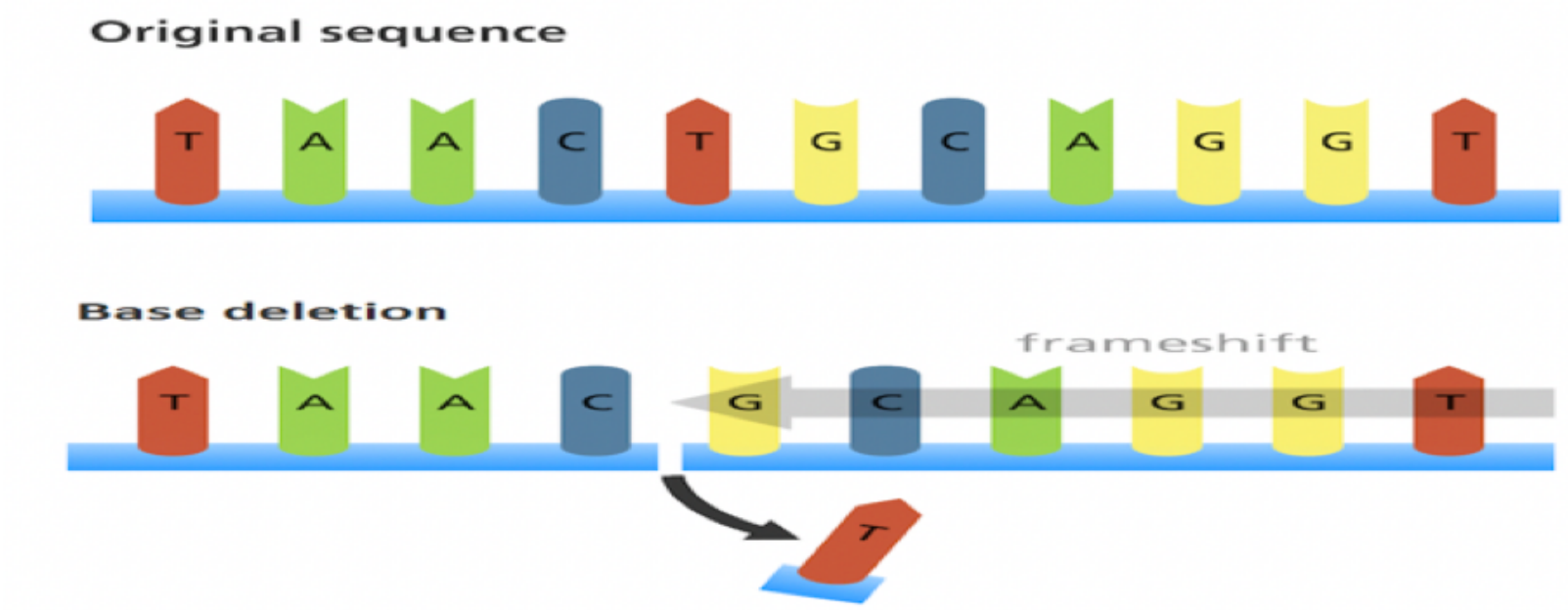
- **Types of Substitution Mutation:**
- **Insertion Mutations:**

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



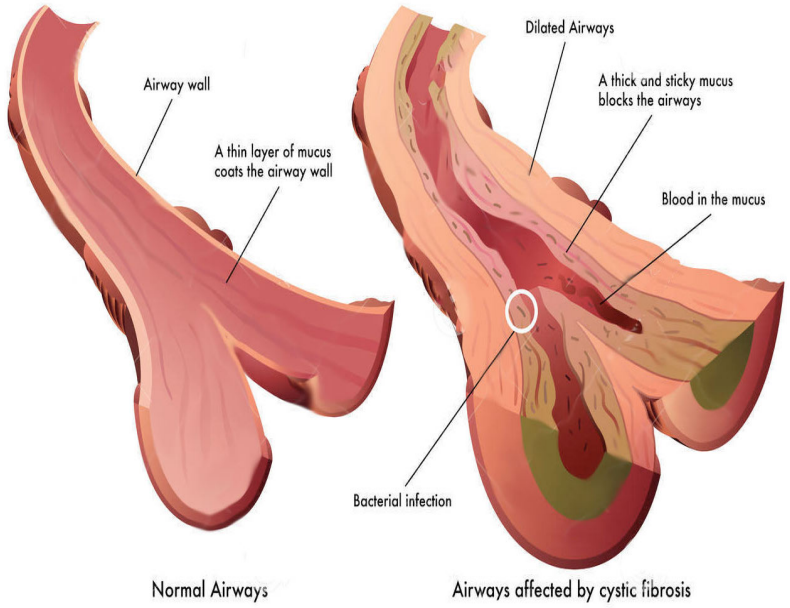
- **Deletion Mutations:**

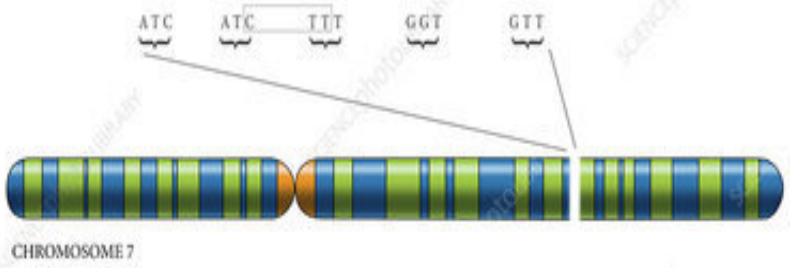
  Deletions are mutations in which a section of DNA is lost, or deleted.



DESCRIBE EXAMPLES OF DISEASES CAUSED BY MUTATIONS

- **Cystic Fibrosis**
  - **An inherited autosomal disease, result of deletion mutation of CFTR protein on chromosome 7.**
  - **CFTR is involved in the production of sweat, digestive fluids, and mucus. When the CFTR is not functional, secretions which are usually thin instead become thick.**



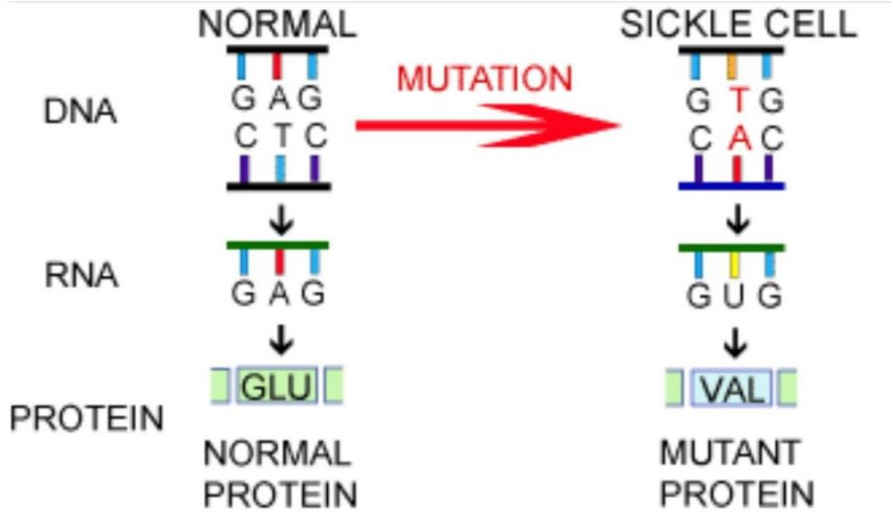


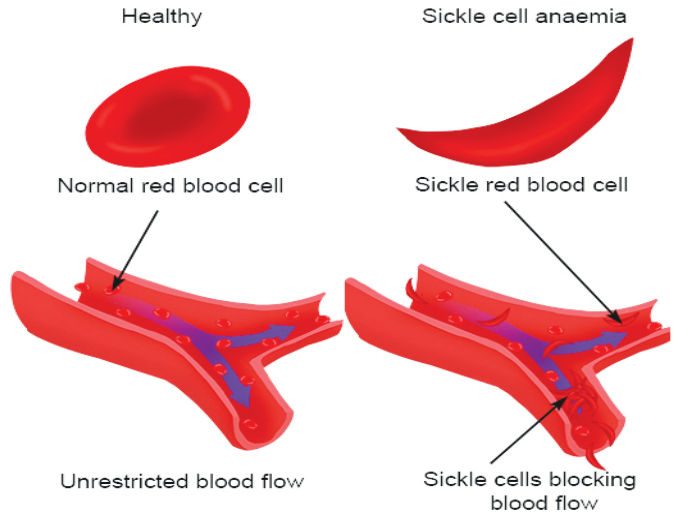
❑ Sickle Cell Anemia:

- **An inherited autosomal disease, result of substitution mutation on .**

- **Change in just one nucleotide in hemoglobin-Beta gene found on chromosome 11.**

- **This mutation causes the hemoglobin in red blood cells to distort to a sickle shape when deoxygenated.**





CONCLUSION

- Mutation is a change in DNA, it can occur in either somatic or germline cells
- It may be spontaneous a result of errors in natural biological processes, or induced caused by agents like radiation and chemicals.
- Mutations are classified into substitutions and frameshift mutations
- Cystic Fibrosis is caused by deletion mutation.
- Sickle Cell Anemia is caused be substitution mutation.

REFERENCES

- Francisco, A. R. L. (2013) *iGenetics*, *Journal of Chemical Information and Modeling*. doi: 10.1017/CBO9781107415324.004.
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THANK
YOU
ROMANS 1:8