Competency 2:

MUTATION

Content Standards:

The learners demonstrate an understanding of

1. the information stored in DNA is being used to make proteins.
2. how changes in DNA molecule may cause changes in its product.

Competency:

Explain how mutations may cause changes in the structure and function of a protein. **S10LT - IIIe -38.**

Objectives:

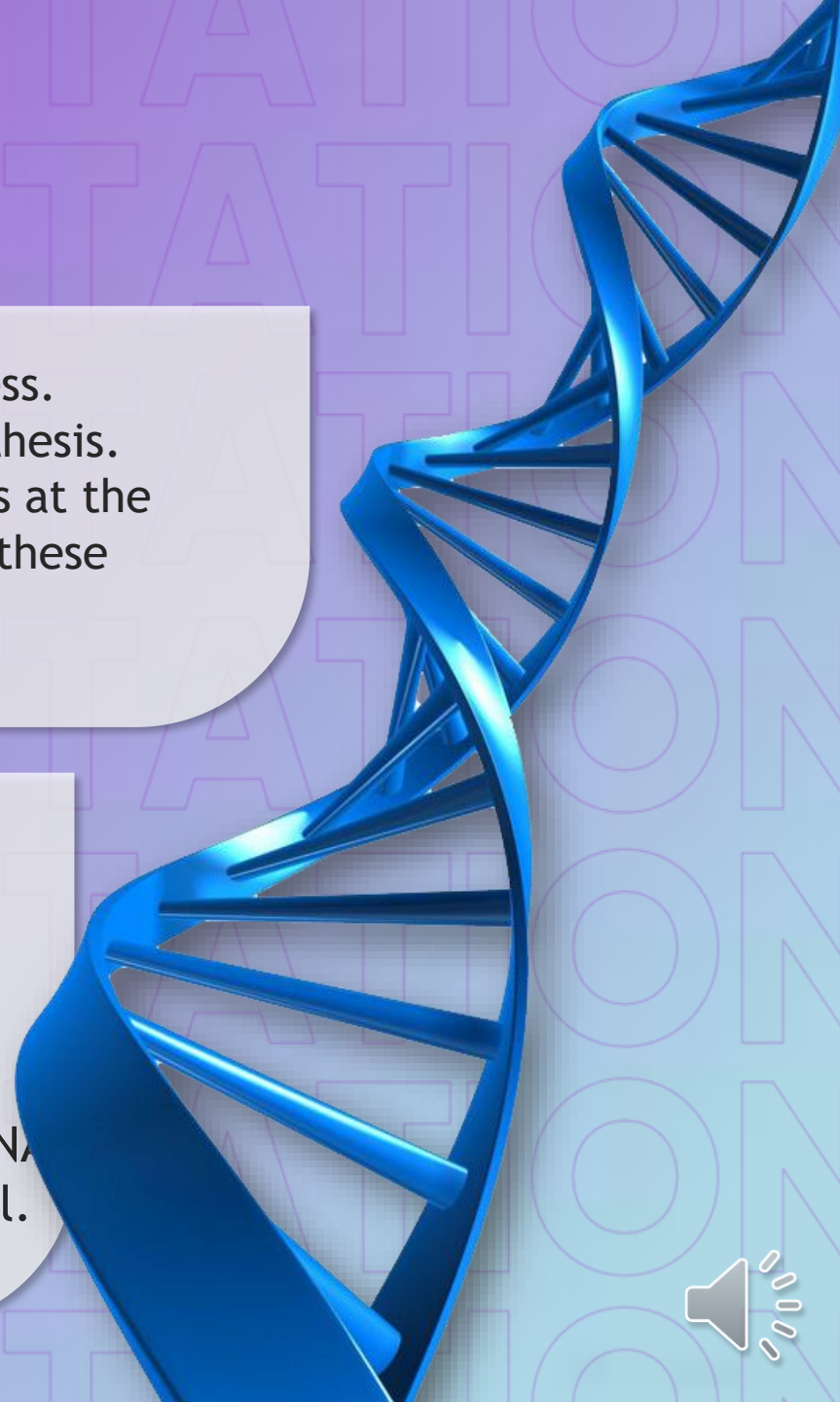
At the end of the material the student will be able to:

1. illustrate different types of chromosomal mutations;
2. differentiate the kinds of chromosomal mutations; and
3. value the significant understanding of genetic mutation and how it may affect one life.

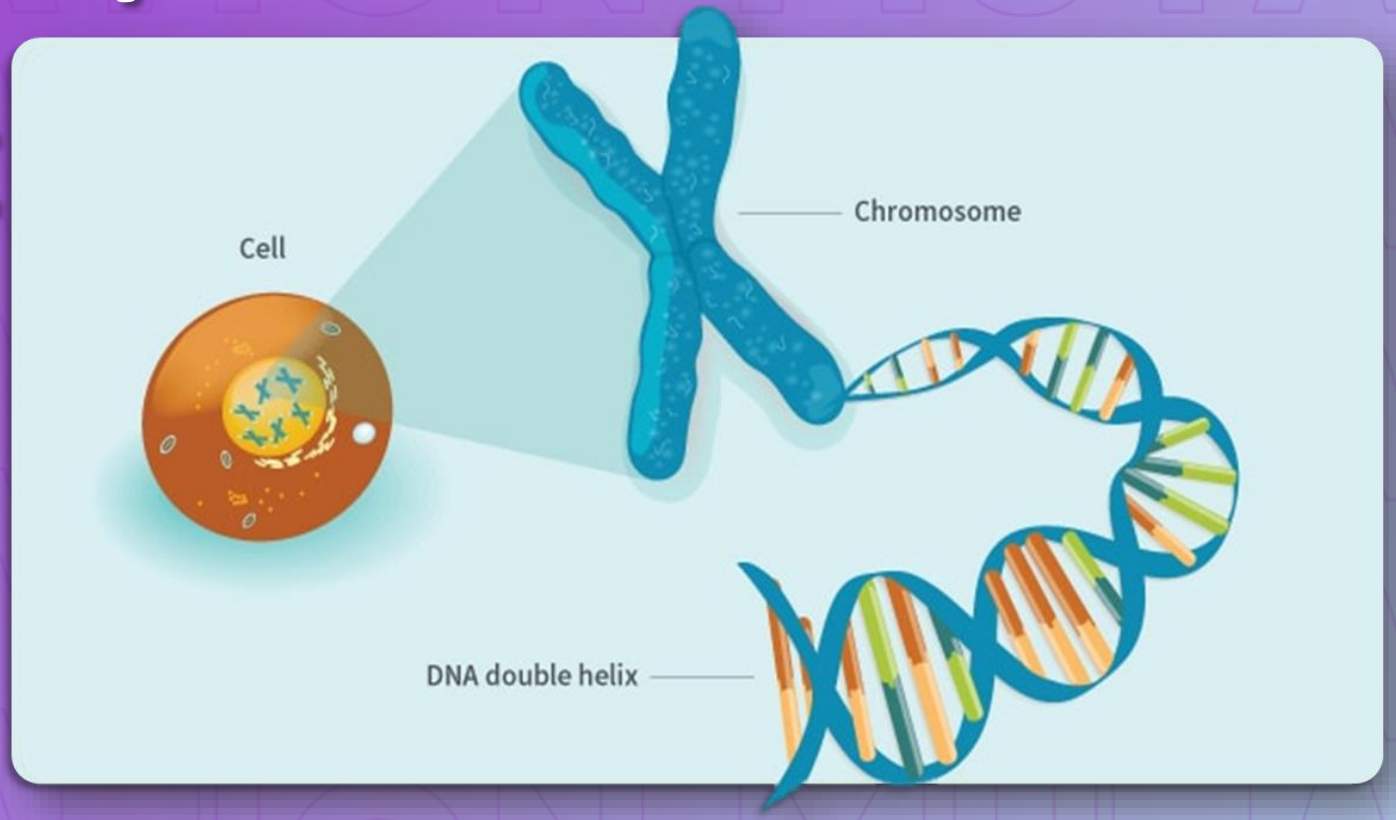
Introduction

The DNA is required to complete the protein production process. Transcription and translation are the two processes of protein synthesis. Messenger RNA sequences are read and translated into amino acids at the ribosome during protein production. Proteins will be formed from these amino acids.

Codons, which are carried by mRNA, specify these amino acids. The genes will be abnormal if the transcription process is incorrectly replicated. This is referred to as a mutation. A mutation is a change in our DNA sequence that happens as a consequence of errors during DNA copying or environmental influences such as UV radiation and cigarette smoke. Because DNA replication involves mutation, transcription into mRNA is unusual.

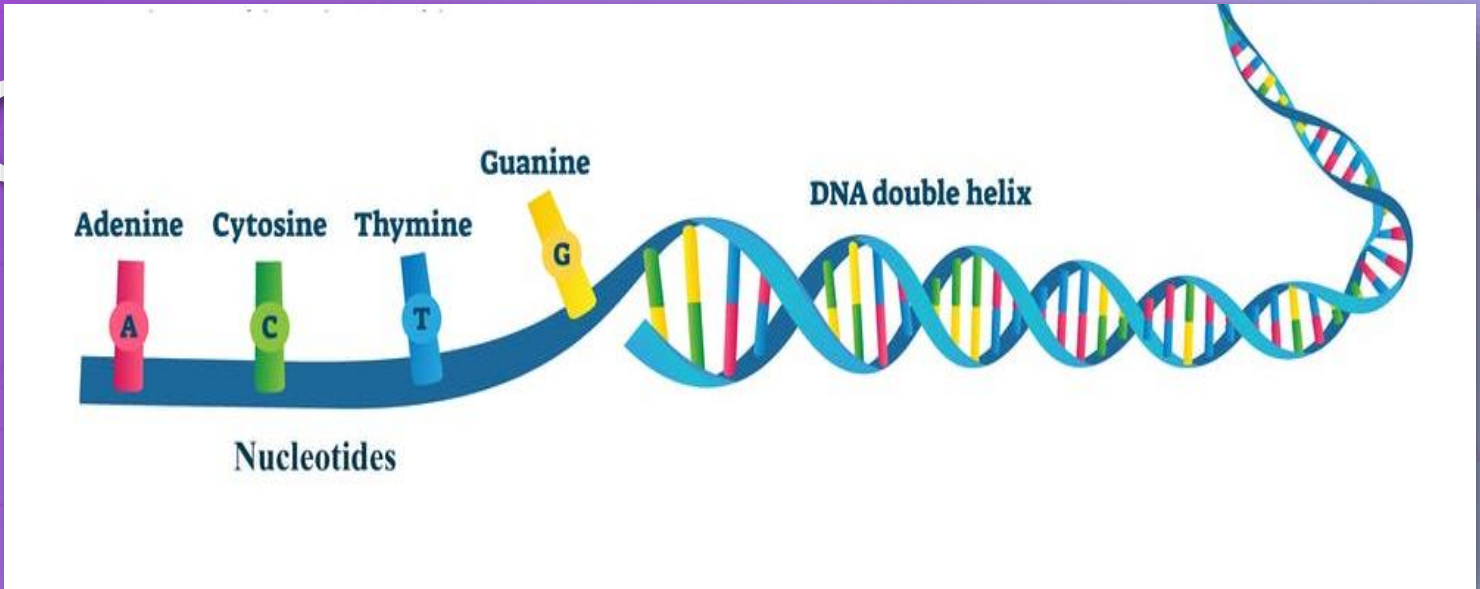


Deoxyribonucleic Ac



DNA (deoxyribonucleic acid) is the molecule that conveys genetic information for an organism's development and function. DNA is made up of two connected strands that loop around each other like a twisted ladder, giving it the form of a double helix

Deoxyribonuclei c Ac

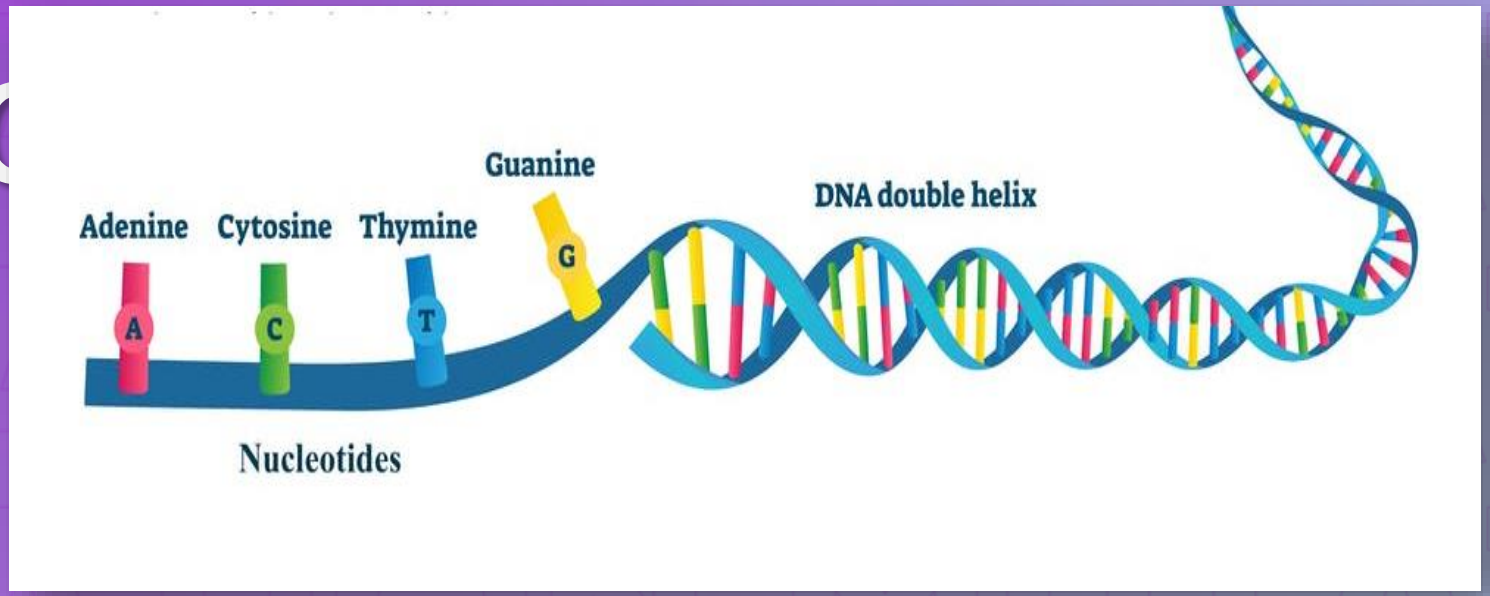


The backbone of each strand is made up of alternating sugar called ***deoxyribose*** and ***phosphate groups***. Each sugar has one of four bases attached to it: ***adenine (A)***, ***cytosine (C)***, ***guanine (G)***, or ***thymine (T)***. Chemical linkages between the bases connect the two strands.

Adenine (A) binds with thymine (T)

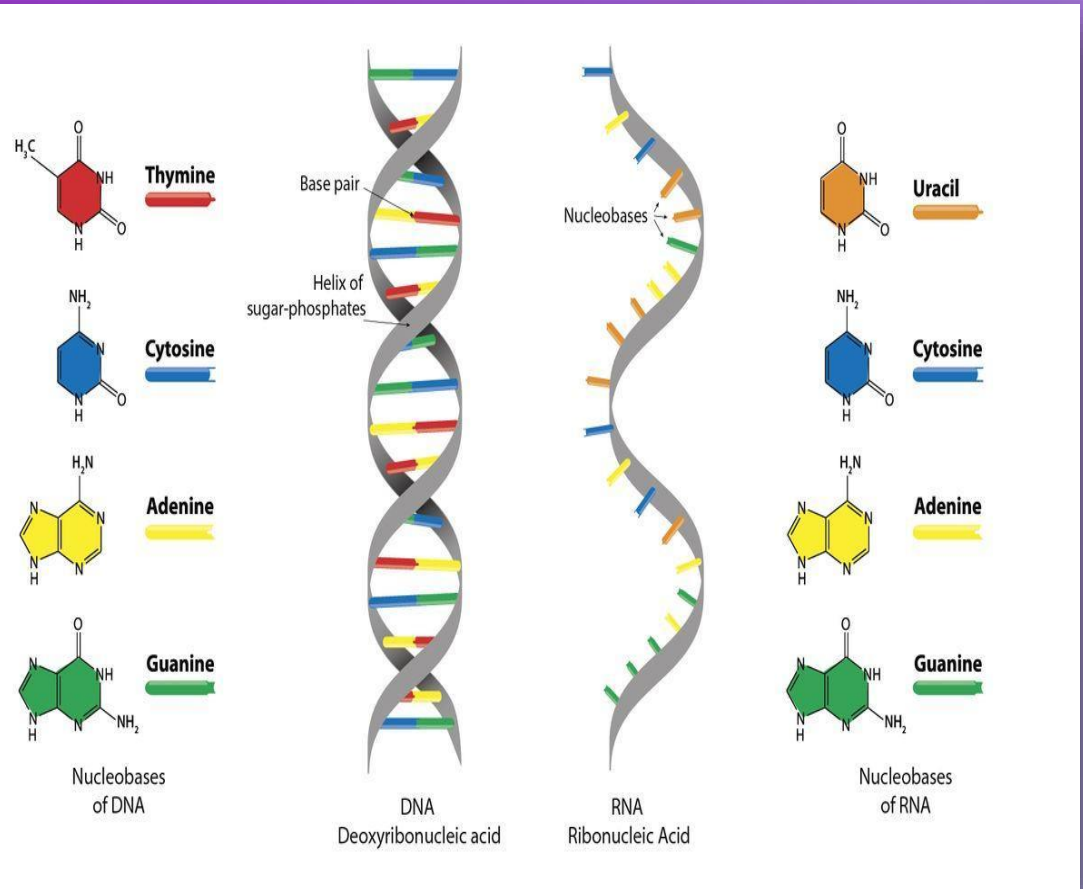
Cytosine (C) binds with guanine (G)

Deoxyribonuclei c Ac



The ***nucleotide sequence*** along DNA's backbone encodes biological information such as protein or RNA molecule instructions.

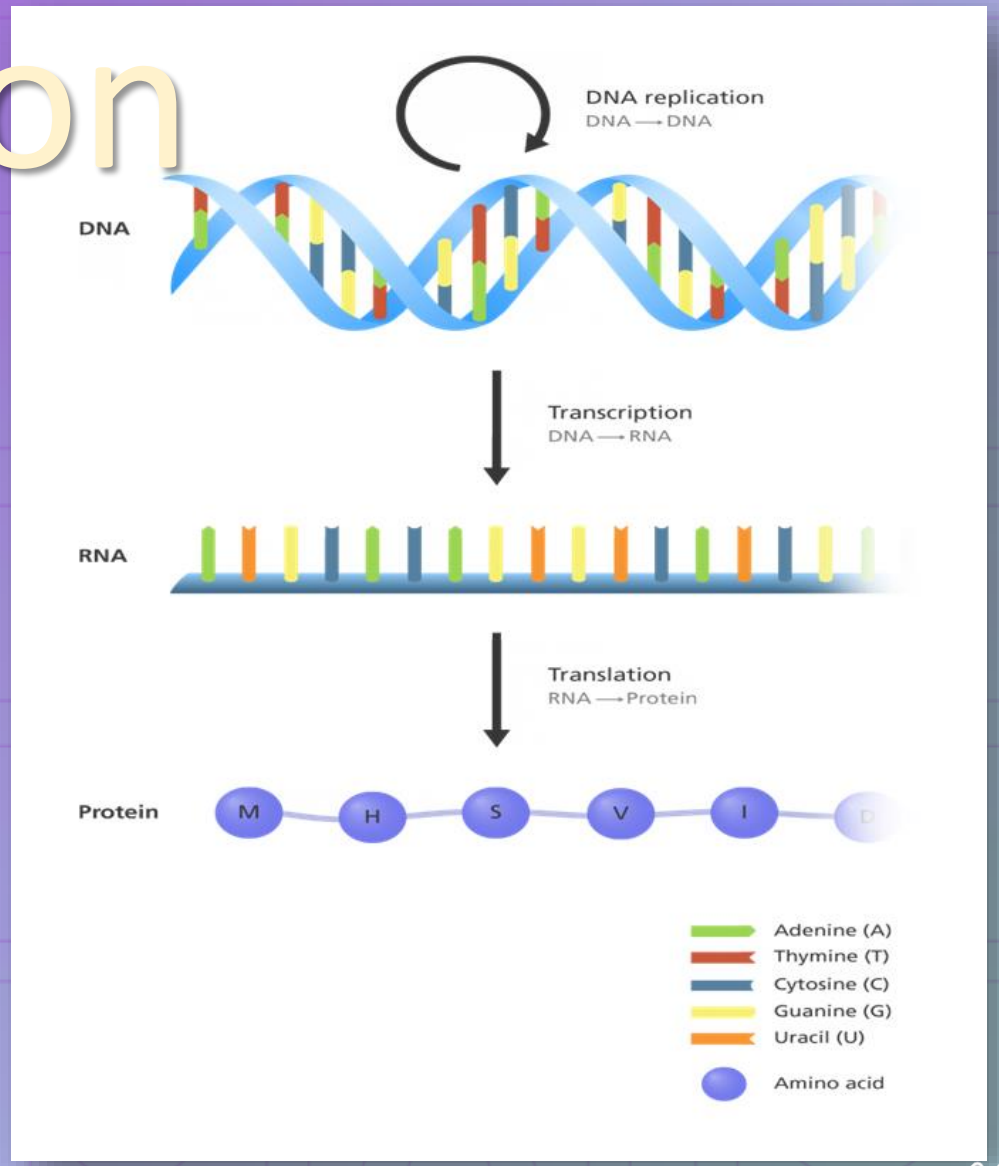
Ribonucleic Acid



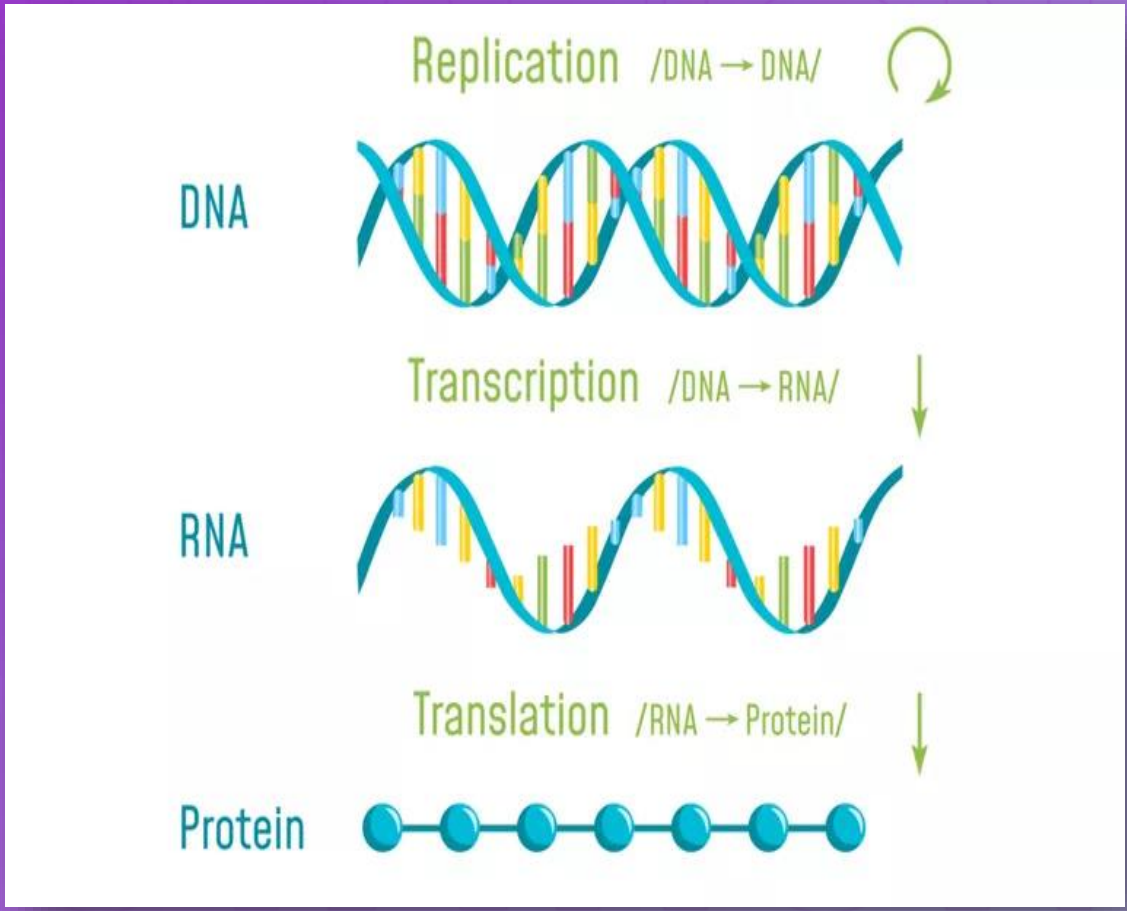
Ribonucleic acid *(RNA)* is a nucleic acid that has structural similarities to DNA and is found in all living organisms. RNA, unlike DNA, is almost always single-stranded. Instead of the deoxyribose present in DNA, an RNA molecule contains a backbone consisting of alternating phosphate groups and the sugar called ribose. Each sugar has one of four bases attached to it: ***adenine (A)***, ***uracil (U)***, ***cytosine (C)***, or ***guanine (G)***. In cells, there are three forms of RNA which are messenger RNA (mRNA), ribosomal RNA (rRNA), and transfer RNA (tRNA).

Gene Expression
(Central Dogma)

The ***Central dogma*** depicts the flow of genetic information in cells, DNA replication, and transcription, which codes for RNA, which then codes for proteins through translation



Transcription



DNA transcription is the process by which RNA polymerase rewrites the genetic information contained in DNA into messenger RNA (mRNA). The mRNA then leaves the nucleus and serves as the foundation for DNA translation. The cell regulates the pace of gene expression by managing the creation of mRNA within the nucleus.

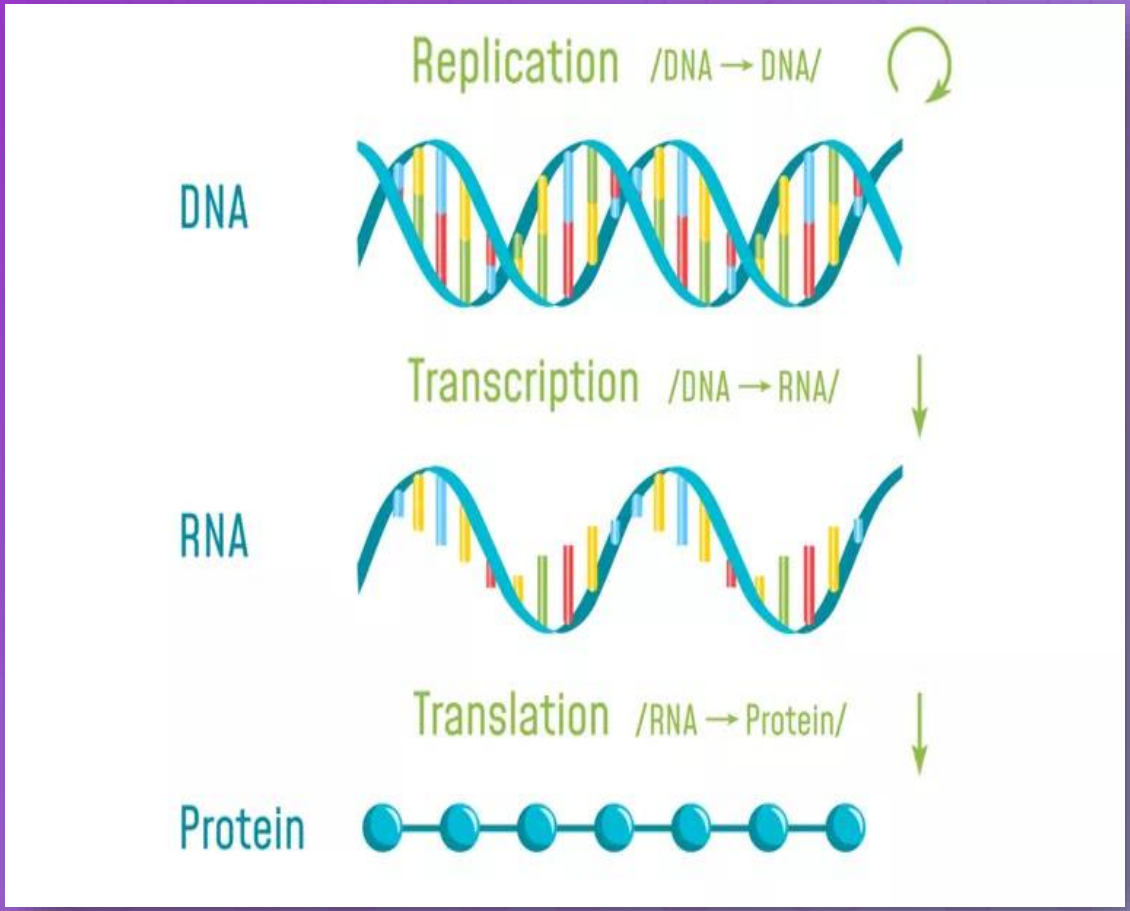
Example:

DNA Replicated sequence - ACGGGTAAGG

Transcribed mRNA - UGCCCAUUCC

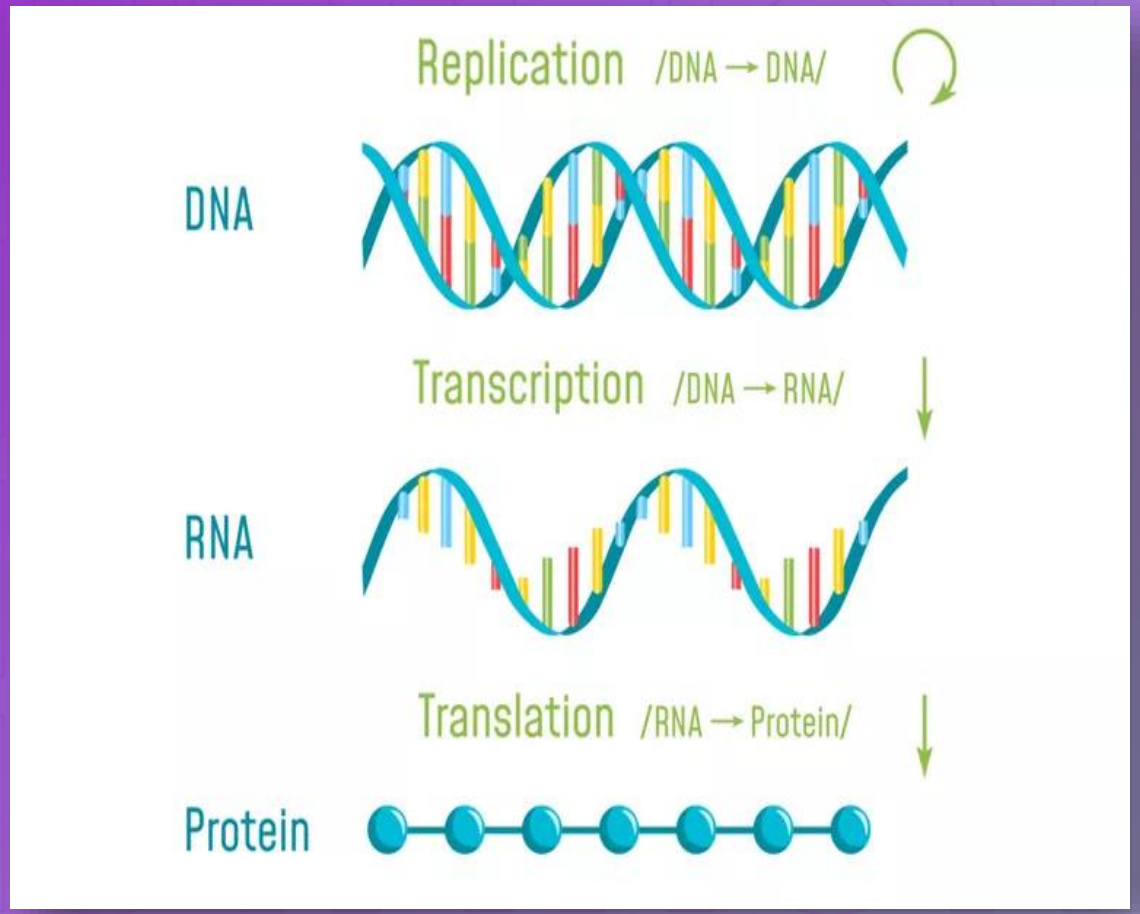
This code will be carried by mRNA to the ribosomes, instructing them on how to produce a protein.

Translation

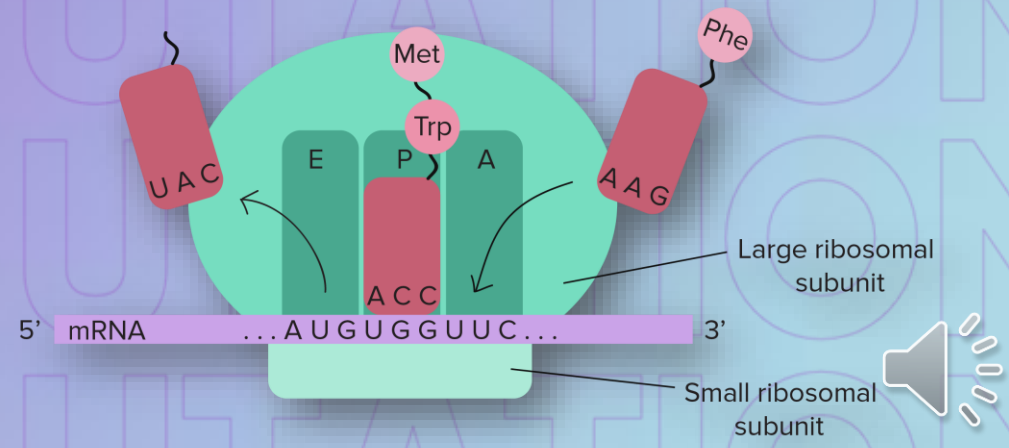


The translation is the process of converting information transmitted from DNA as messenger RNA into a sequence of amino acids linked by peptide bonds. It's a conversion from one code (nucleotide sequence) to another (amino acid sequence).

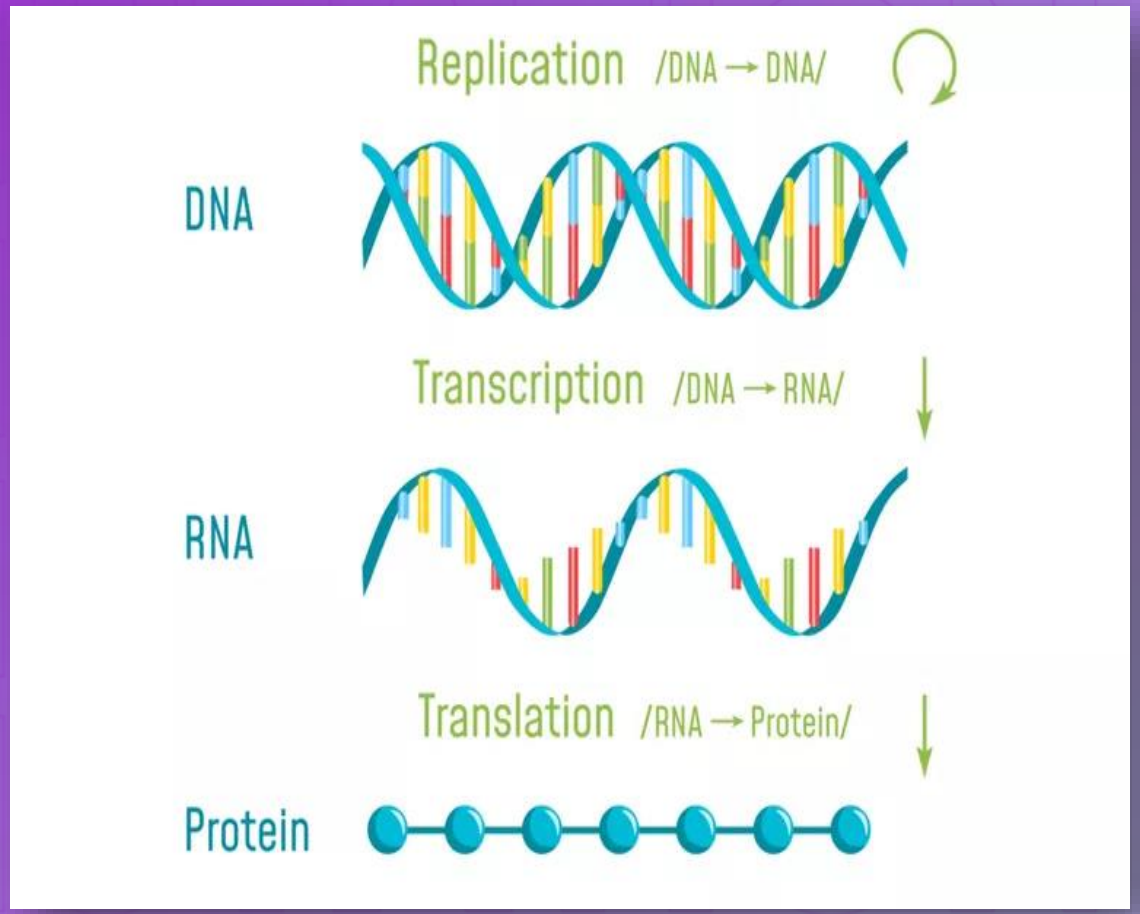
Translation



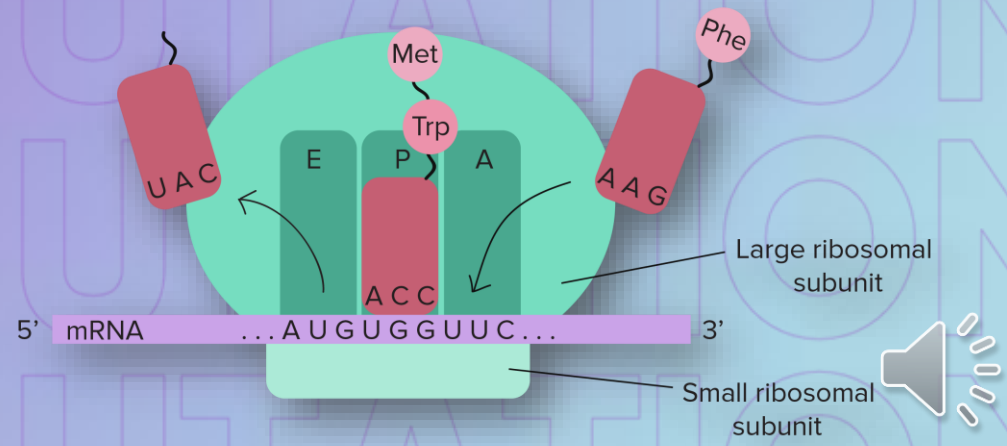
This process takes place on the ***ribosome***, much as mRNA synthesis takes place on the RNA polymerase. The ribosome matches the three complementary bases in the anticodon sections of tRNA molecules to the base sequence on the mRNA in sets of three bases called codons. The base-pairing rule is crucial in this identification once again Adenine (A) binds to Uracil (U) and Cytosine (C) binds to Guanine (G).



Translation



The ribosome travels along the mRNA chain, matching three base pairs at a time, and adds amino acids to the polypeptide chain. The ribosome releases both the polypeptide and the mRNA when it reaches one of the "stop" codes. This polypeptide will twist back into its original shape and begin to function as a protein.



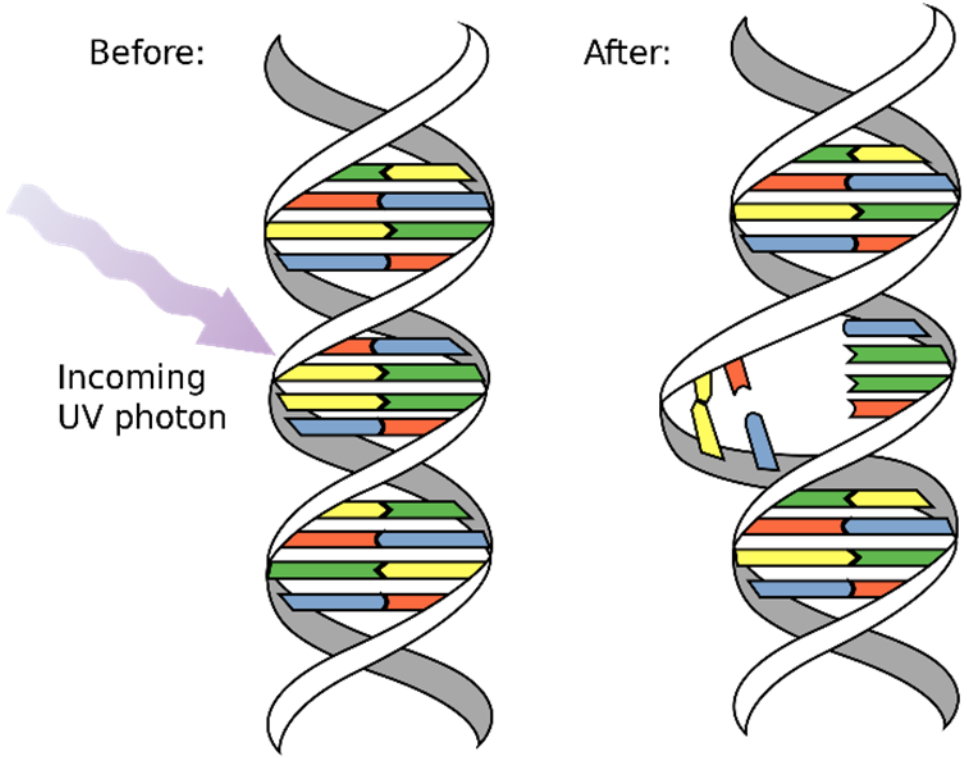
Translation

Example:

DNA	TAC	CGC	TCC	GCC	GTC	GAC	AAT	ACC	ACT
mRNA	AUG	GCG	AGG	CGG	CAG	CUG	UUA	UGG	UGA
tRNA	UAC	CGC	UCC	GCC	GUC	GAC	AAU	ACC	ACU
AA	Met	Ala	Arg	Arg	Gln	Leu	Leu	Trp	Stop

Mutation

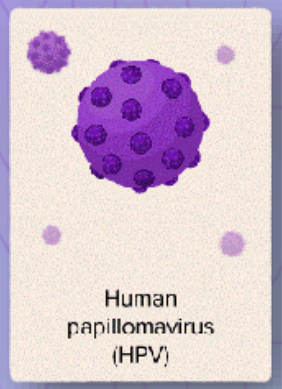
A mutation is a change in the sequence of DNA. In the same way that DNA contains information, Mutations are errors in the genetic coding of amino acids that usually happen in transcription. There are two types of mutation; gene mutation and chromosomal mutation.



Mutation

Mutagens

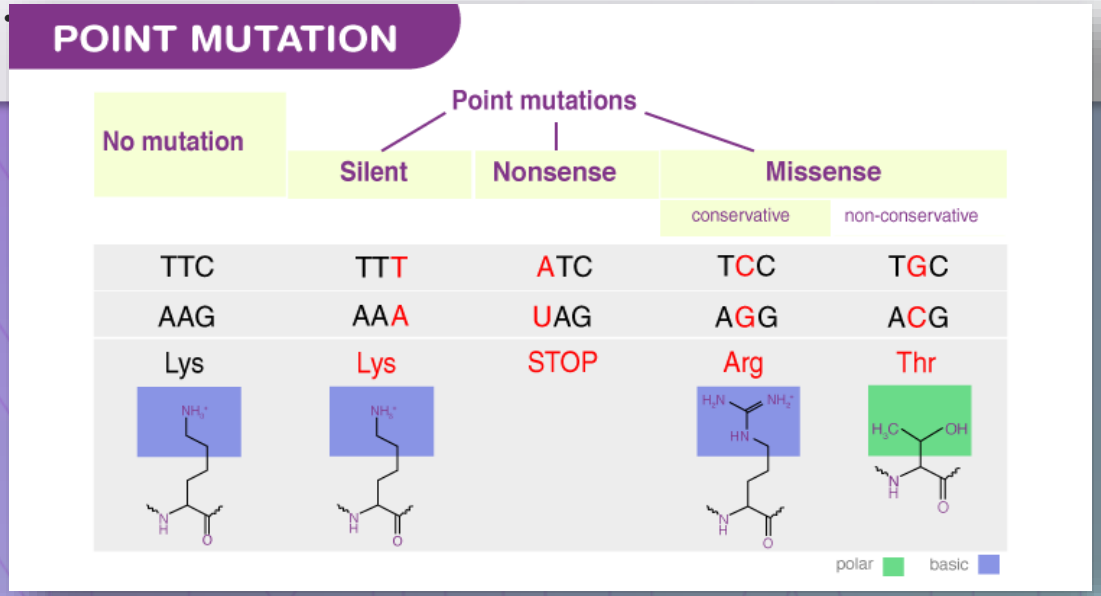
Mutagens are substances that change DNA and can cause cancer. Permanent changes in the DNA sequence are caused by an organism's capacity to repair the damage. Radioactive substances, x-rays, UV light, and some chemicals or medications are examples of mutagens.



Mutation

Point Mutation

It is a DNA or RNA mutation in which a single nucleotide base is deleted, inserted, or changed. Substitution mutation can exist as a result of this. Substitution mutation can have three forms. These are ***nonsense***, ***missense***, and ***silent mutation***.



Mutation

Point Mutation

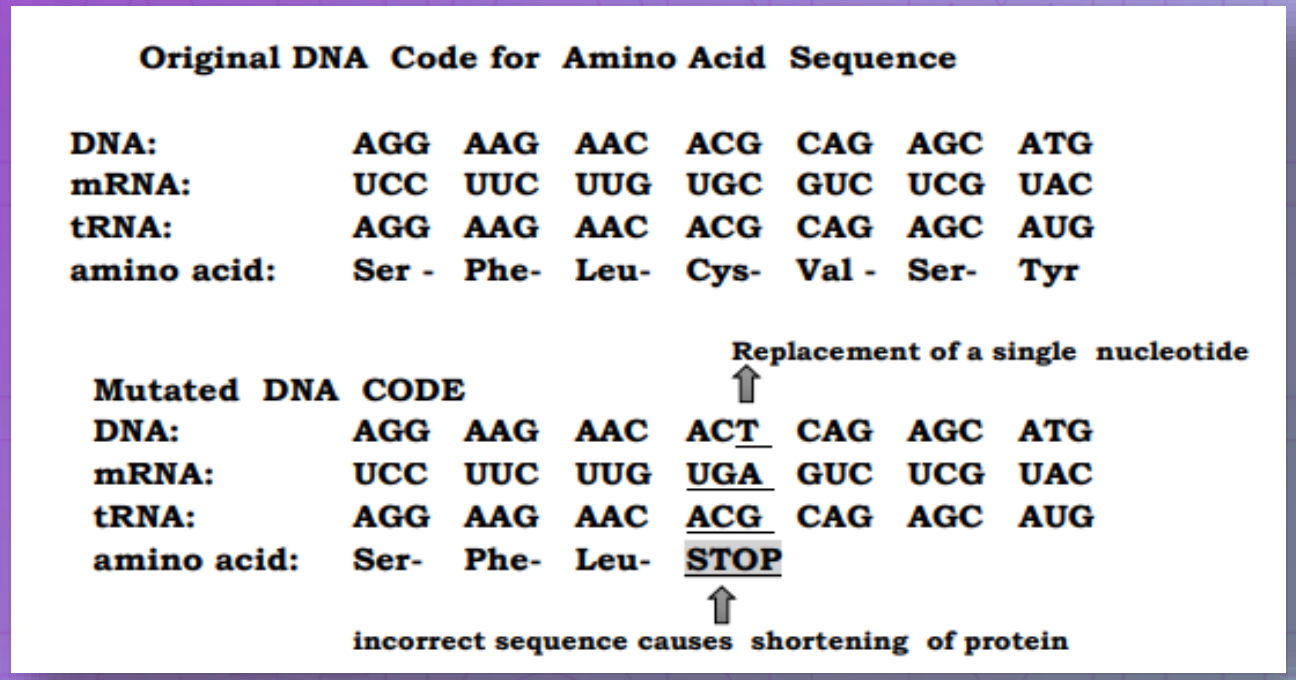
1. Nonsense Mutation

Due to the replacement of one nitrogenous nucleotide, nonsense mutation leads to the development of a stop codon. Stop codons are nitrogenous nucleotides that halt the translation step of protein synthesis. In DNA, they are ATC, ATT, or ACT, while in mRNA, they are UAG, UAA, or UGA. They are frequently found near the end of the nucleotide base sequence of messenger RNA. When a substitution mutation leads it to emerge in a different location, the translation process to amino acid is abruptly stopped and the right protein is not produced.

Mutation

Point Mutation

1. Nonsense Mutation



Mutation

Point Mutation

2. Missense Mutation

Missense mutation occurs when one nitrogenous base of the DNA is changed, resulting in an altered codon but without forming a stop codon.

This results in the formation of new amino acids during protein synthesis.

Example:

DNA: CAT to mRNA: GUA to tRNA CAU (Valine)

CAT is changed into CCT to mRNA: GGA to tRNA: CCU (Glycine)

Mutation

Point Mutation

2. Missense Mutation

Missense mutations are divided into two types: ***conservative*** and ***non-conservative***.

- ***Conservative mutation*** occurs when the newly created amino acid has the same qualities as the one that was intended to be produced.
- ***Non-conservative*** occurs when the newly generated amino acid differs from the one that was expected to be created.

Mutation

Point Mutation

2. Missense Mutation

Original DNA Code for Amino Acids

DNA:	GCA	ATG	CAT	CAT	CGT	CGC	CAA
mRNA:	CGU	UAC	GUA	GUA	GCA	GCG	GUU
tRNA:	GCA	ATG	CAU	CAU	CGU	CGC	CAA
amino acid:	ARG-	TYR	VAL-	VAL -	ALA-	ALA-	VAL

Mutated DNA CODE

Replacement of a single nucleotide

DNA:	GCA	ATG	CAT	CCT	CGT	CGC	CAA
mRNA:	CGU	UAC	GUA	GGA	GCA	GCG	GUU
tRNA:	GCA	ATG	CAU	CCU	CGU	CGC	CAA
amino acid:	ARG	TYR	VAL	GLY	ALA	ALA	VAL

This is an incorrect amino acid which may produce a malfunctioning protein

Mutation

Point Mutation

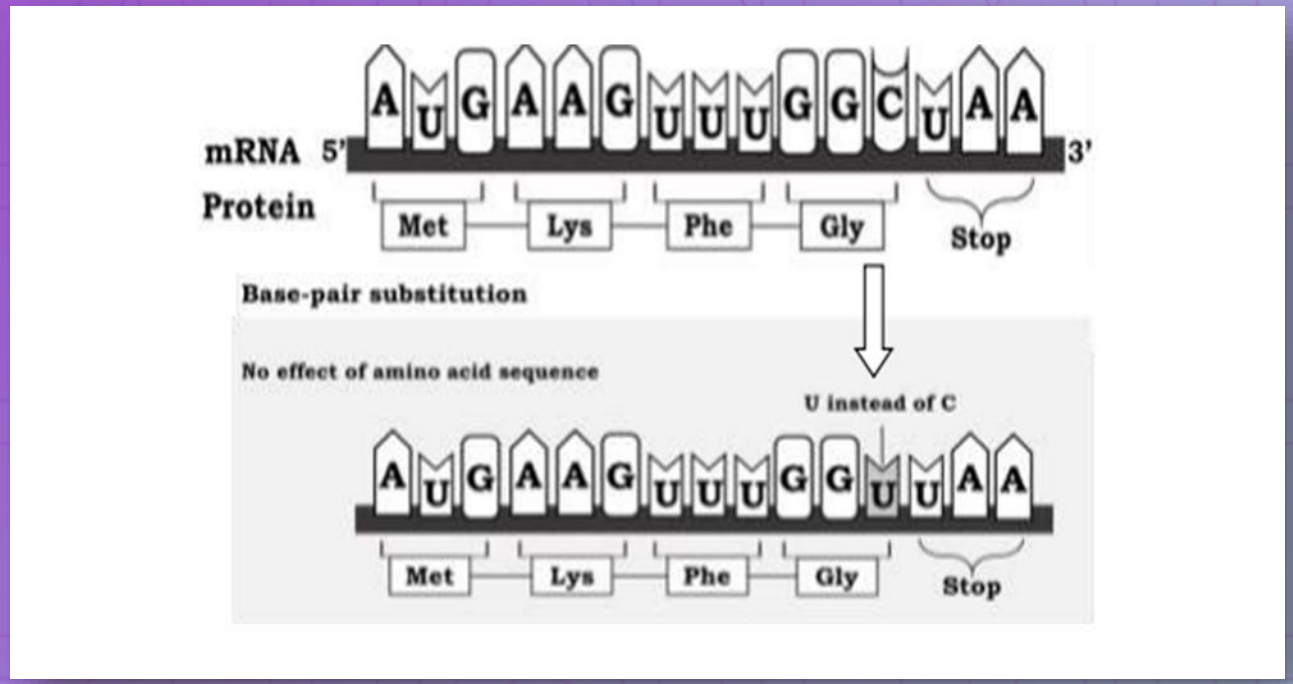
3. Silent Mutation

Silent mutation occurs when a nitrogenous base is mutated while producing the same amino acid. Keep in mind that many codons might code for the same amino acid. GGC and GGU, for example, can both code for glycine. If C is changed to U, the same amino acid is generated, and so the amino acid is not modified or altered

Mutation

Point Mutation

3. Silent Mutation



Mutation

Frameshift Mutation

Frameshift mutation occurs when the regular sequence of codons is disrupted by the insertion or deletion of one or more nitrogenous bases, where the number of nitrogenous bases inserted or deleted is not a multiple of three. For example, if only one nucleotide is removed, all codons after the mutation will have an altered reading frame. This can result in the integration of various modifications in amino acids into the protein, which can disrupt the amino acid chain. When three nitrogenous bases are deleted or inserted, there is no change in the codon reading frame, but there is either an additional or missing amino acid in the protein. As a result, frameshift mutations result in an aberrant protein with an incorrect amino acid sequence that might be longer or shorter than the normal protein.

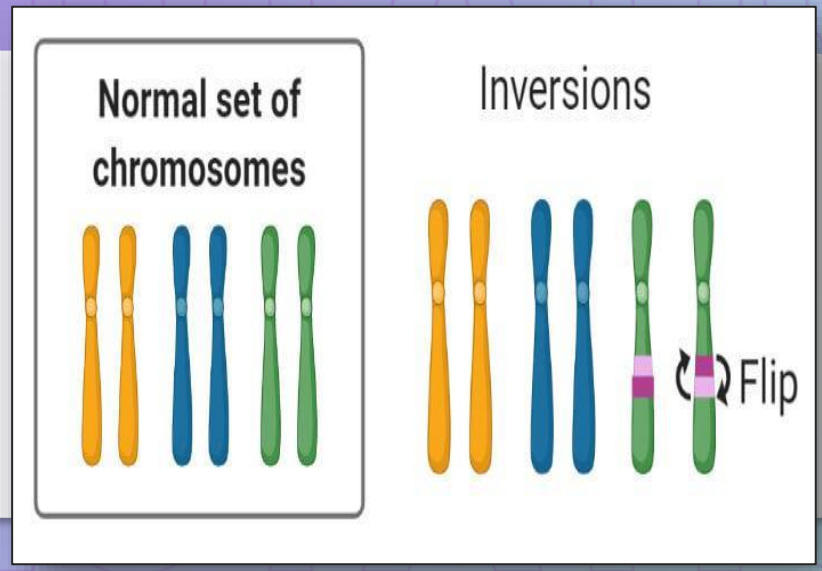
Mutation

Frameshift Mutation

Kinds of Frameshift Mutation:

1. **Inversion**

The chromosome is inverted, and its segments are reversed from end to end. A fragment of the chromosome is taken and then reattached, but oppositely as before.



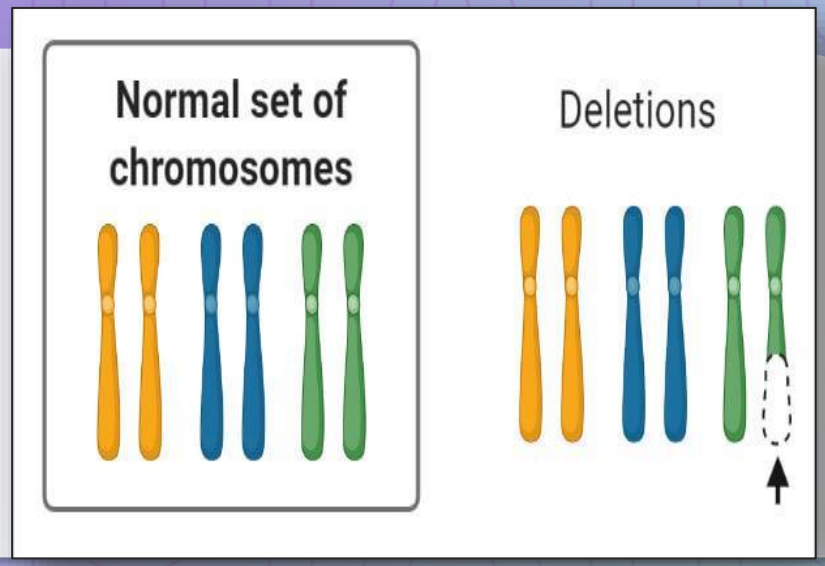
Mutation

Frameshift Mutation

Kinds of Frameshift Mutation:

2. Deletion

These arise when a chromosomal fragment is unintentionally removed or deleted. There are circumstances when one piece is removed at the end (terminal deletion), two deletions when one is within the chromosome, and one at the end (interstitial deletion).



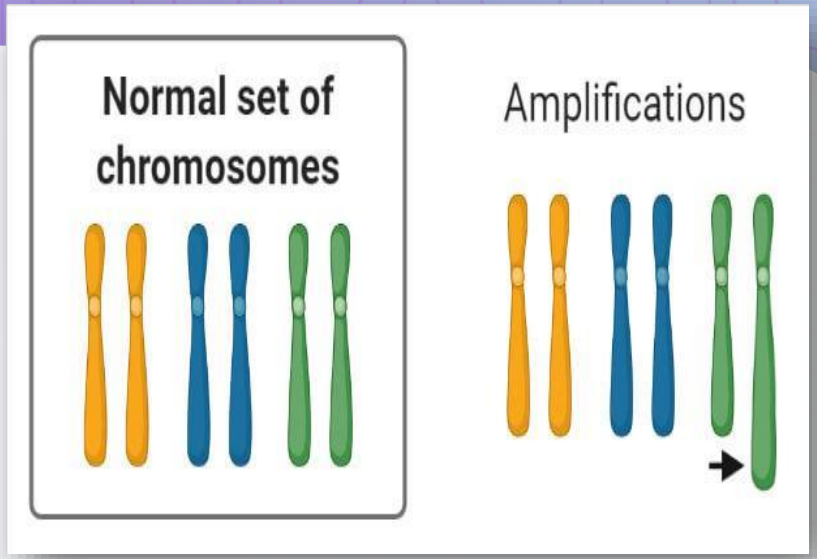
Mutation

Frameshift Mutation

Kinds of Frameshift Mutation:

3. **Duplication/Amplification**

Occurs when the nucleus has an extra copy of a segment or the complete chromosome. These are sometimes referred to as incomplete trisomies. In the case of duplication, an organism that normally has two copies of a chromosome will often have three. This can occur anywhere throughout the chromosome, including the middle and ends.



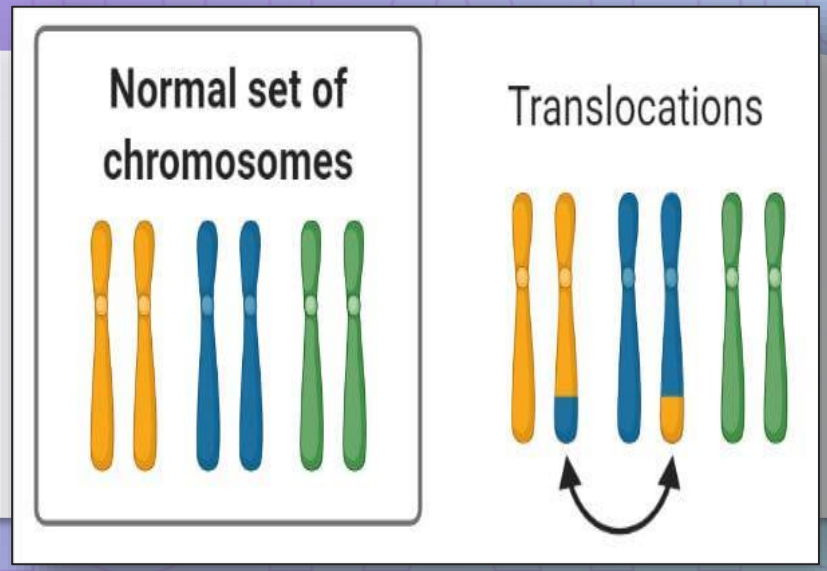
Mutation

Frameshift Mutation

Kinds of Frameshift Mutation:

4. **Translocation**

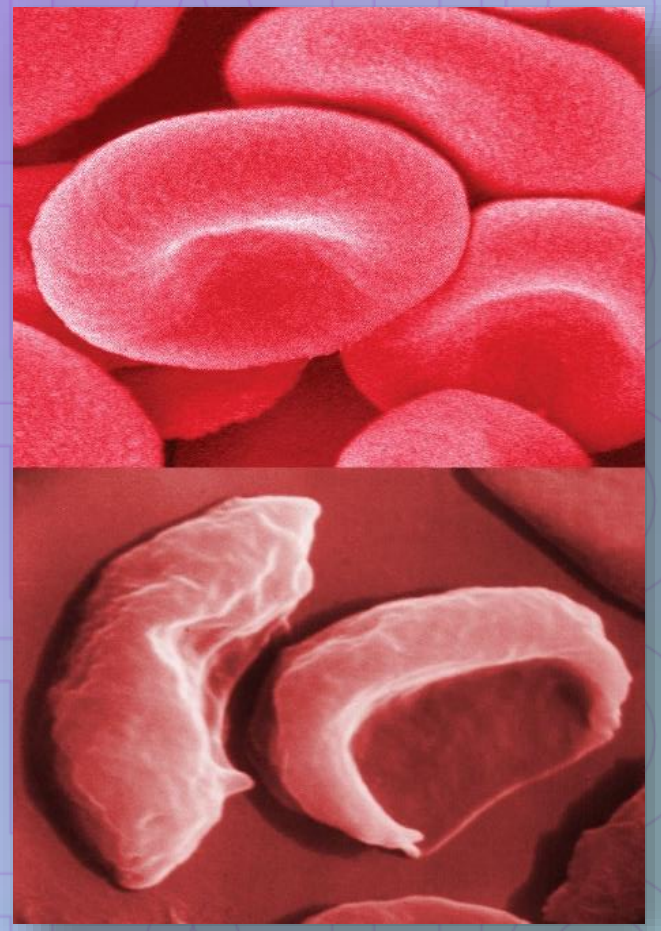
This occurs when a fragment of a chromosome breaks off and moves to a separate chromosome. As one kind of chromosome fuses with another, fusion chromosomes are formed.



Effects of Mutated Genes

Sickle Cell Anemia

This form of anemia is caused by a recessive condition induced by a single substitution mutation in the gene that produces hemoglobin. Hemoglobin is well-known for transporting oxygen throughout the body. Glutamic acid is generated in the chain of a normal gene. However, when the amino acid valine replaces glutamic acid, sickle-shaped blood cells are produced. These cells are unable to transport oxygen correctly. Anemia, pain crises, and recurrent infections are signs of sickle cell anemia. Prescription medicines, folic acid, bone marrow transplants, and blood transfusions can all help



Effects of Mutated

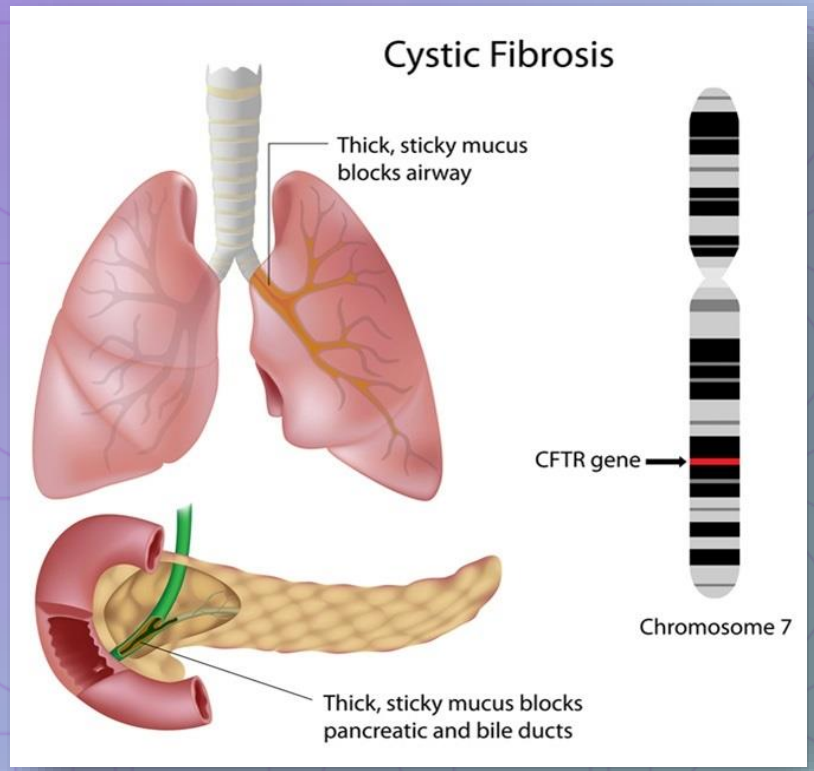
Albinism

It is an autosomal recessive condition caused by a deletion mutation in which melanin synthesis is decreased or nonexistent in skin, hair, and eyes due to tyrosinase inactivity. The loss of the tyrosinase gene causes this.

Effects of Mutated

Cystic Fibrosis

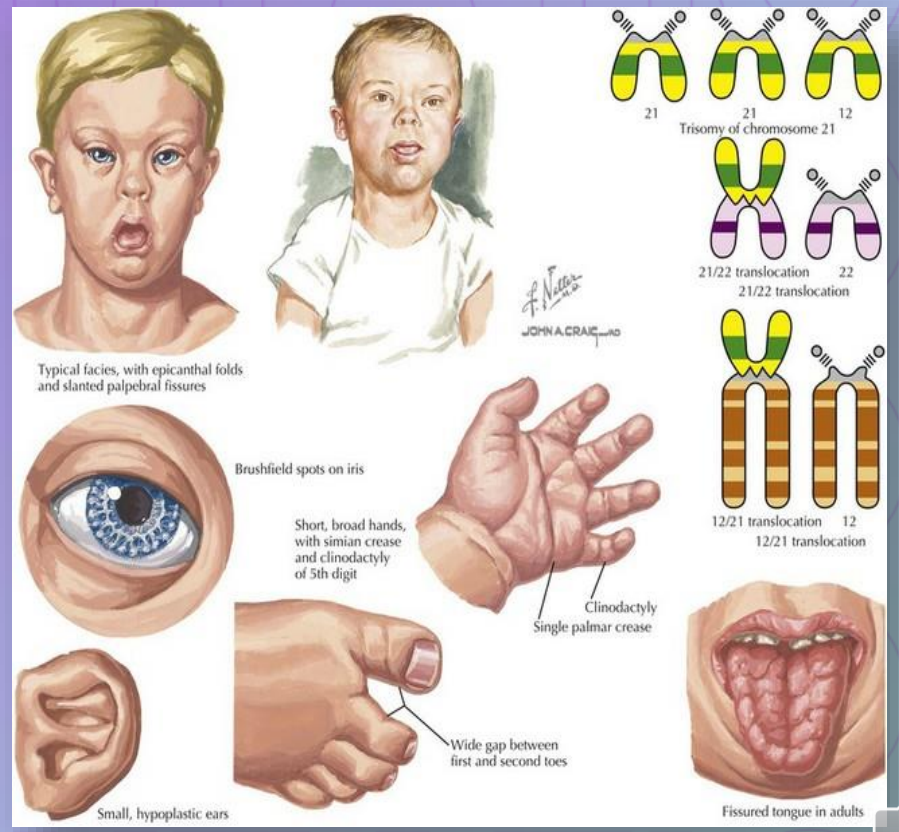
Is an inherited recessive condition. Although many distinct variants can cause cystic fibrosis, the deletion mutation is the most prevalent. It affects the cystic fibrosis transmembrane conductance regulator (CFTR) gene, causing the amino acid phenylalanine to be deleted. This results in an erroneous protein.



Effects of Mutated Genes

Down Syndrome

Is associated with mild cognitive impairment. It is also distinguished by physical development, body, and facial characteristics impairment. Down syndrome is caused by a meiotic translocation that transfers the majority of chromosome 21 onto chromosome 14.



Station 1

Modeling of Chromosomal Mutations!

(Mastery Modelling)

Materials:

i. Little bit pieces of paper
ii. Pencil
iii. Tape

Procedure:

a. Write numbers 1-8 on pieces of paper (one number for each piece). Tape the pieces together in numerical order to create a model of a chromosome with 8 genes.
b. Use the "chromosome" you created to model four changes in chromosome structure. For example, remove the number 3 and rejoin the remaining chromosome segments to represent a deletion.
c. Reconstruct the original chromosome before modeling inversion, deletion, duplication, and translocation. Use the extra pieces of paper to make the additional numbers you need.
d. You have 10 minutes to construct the model.
e. When time is up, prepare for the presentation of your model.

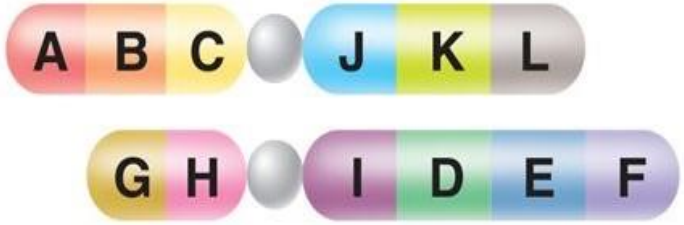
Guide Questions:

1. How long does a regular menstrual cycle last?
2. Describe what happens to an egg during the first 14 days of the cycle in Part A.
3. Describe what happens to the egg if fertilization occurs.
4. Explain what takes place in the uterus after fertilization.
5. Why is it important to study the menstrual cycle?

Station 2

Chromosomal Mutation

(Individual Activity)

A. 	➢
B. 	➢
C. 	➢
D. 	➢

SUM IT UP!

1. In _____, the chromosome is inverted, and its segments are reversed from end to end.
2. In _____, a chromosomal fragment is unintentionally removed or deleted.
3. In _____, are sometimes referred to as incomplete trisomies.

4. In _____, a fragment of a chromosome breaks off and moves to a separate chromosome.

Assessment

Competency 2
Describe the feedback mechanisms involved in regulating processes in the female reproductive system (e.g., menstrual cycle) S10LT – IIIc -35

Direction: Choose the letter of the correct answer.

12. What can occur in two different types of cells: the reproductive and body cells?
    a. Mutations
    b. RNA
    c. Gene mutation
    d. Chromosomal mutation

13. What is a permanent change in the DNA sequence that makes up a gene?
    a. Mutations
    b. RNA
    c. Gene mutation
    d. Chromosomal mutation

14. _____ occurs at the chromosome level resulting in gene deletion, duplication, or rearrangement.
    a. Mutations
    b. RNA
    c. Gene mutation
    d. Chromosomal mutation

15. It occurs when a piece of one chromosome breaks off and attaches to another chromosome. What kind of chromosomal mutation is this?
    a. Chromosomal Deletion
    b. Chromosomal Inversion
    c. Chromosomal Translocation
    d. Chromosomal Duplication

16. When a piece or section of chromosomal material is missing, this is referred to as partial monosomy. What kind of chromosomal mutation is this?
    a. Chromosomal Deletion
    b. Chromosomal Inversion
    c. Chromosomal Translocation
    d. Chromosomal Duplication

17. A chromosomal rearrangement in which a chromosome segment is reversed end-to-end. What kind of chromosomal mutation is this?
    a. Chromosomal Deletion
    b. Chromosomal Inversion
    c. Chromosomal Translocation
    d. Chromosomal Duplication

18. Involves the production of one or more copies of a gene or chromosomal region. What kind of chromosomal mutation is this?
    a. Chromosomal Deletion
    b. Chromosomal Inversion
    c. Chromosomal Translocation
    d. Chromosomal Duplication

19. The Homologous pair of chromosomes are arranged in what characteristics?
    a. Size
    b. Numbers
    c. Activity
    d. Frequency

20. How do three chromosomal aberrations differ from one another?
    a. They are different by the frequency of smaller genes
    b. They are different by the number of genes within the chromosome.
    c. They are different by the descending sizes of the genes within the chromosome.
    d. They have no distinct difference.

21. This abnormality is caused by an extra chromosome in chromosome 18.
    a. Down Syndrome
    b. Edwards Syndrome
    c. Klinefelter's Syndrome
    d. Turner's syndrome

22. This abnormality causes the female sexual characteristic to be underdeveloped.
    a. Down Syndrome
    b. Edwards Syndrome
    c. Klinefelter's Syndrome
    d. Turner's syndrome

23. Cri du chat which is an abnormality that causes babies to make high-pitched cries is a result of?
    a. Deletion of the short part of chromosome 5
    b. Duplication of the short part of chromosome 6
    c. Deletion of the short part of chromosome 6
    d. Duplication of the short part chromosome 5

24. Why is a specific base pairing essential for transcription and translation?
    a. Because specific base pairing dictates the particular type of amino acids to be created
    b. Because specific base pairing powers the translation and transcription processes
    c. Because specific base pairing is our defense for specific kinds of mutations
    d. The specific base pairing has no influence on the two-process stated.

25. Protein X's eleventh amino acid should be serine, but due to a mutation, it now has alanine at that position. What type of mutation may have resulted in this change?
    a. Missense
    b. Nonsense
    c. Silent
    d. Insertion

Thank You

COMPETENCY 2: MUTATION