



Al-Mahdi High Schools

In his Name

Chapter 3:

**Genetic variation and polymorphism
worksheet 2**

Documents 2: mutation and multiple alleles

AY: 2020/2

Exercise 1: complete the following tables:

Normal non-transcribed strand of DNA with corresponding peptide	Mutant non-transcribed strand of DNA with corresponding peptide
DNA: CCA-GAG-ACG-TAC-ACT-GGC-CAC	DNA: CCA-GAG-AGG-TAC-ACT-GGC-CAC
Peptide: Pro-Glu- Thr- Tyr- Thr-Gly-His....	Peptide: ...Pro-Glu-Arg- Tyr -Thr-Gly-His....

Type of mutation:

Effect of the mutation:

Normal non-transcribed strand of DNA with corresponding peptide	Mutant non-transcribed strand of DNA with corresponding peptide
DNA :CCA-GAG-ACG-TAC-ACT-GGC-CAC	DNA: CCA-GAG-ACC-TAC-ACT-GGC-CAC
Peptide...Pro-Glu-Thr-Tyr-Thr-Gly-His....	Peptide: ...Pro-Glu-Thr -Tyr-Thr-Gly-His....

Type of mutation:

Effect of the mutation:

Normal non-transcribed strand of DNA with corresponding peptide	Mutant non-transcribed strand of DNA with corresponding peptide
DNA: CCA-GAG-ACG-TAC-ACT-GGC-CAC	DNA: CCA-GAG-ACG-TAG-ACT-GGC-CAC
Peptide: Pro- Glu-Thr- Tyr-Thr -Gly-His--	Peptide...Pro-Glu-Thr

Type of mutation:

Effect of the mutation:

Normal non-transcribed strand of DNA with corresponding peptide	Mutant non-transcribed strand of DNA with corresponding peptide
DNA: CCA-GAG-ACG-TAC-ACT-GGC-CAC....	DNA: CCA-GGA-CGT-ACA-CTG-GCC-AC..
Peptide: ...Pro-Glu-Thr-Tyr-Thr-Gly-His....	Peptide: ...Pro-Gly-Arg-Thr-Leu-Ala-....

Type of mutation:

Effect of the mutation:

Normal non-transcribed strand of DNA with corresponding peptide	Mutant non-transcribed strand of DNA with corresponding peptide
<u>DNA: CCA-GAG-ACG-TAC-ACT-GGC-CAC</u>	<u>DNA: CCA-GAG-ATC-GTA-CAC-TGG-CCA-C</u>
<u>Peptide: Pro-Glu-Thr- Tyr-Thr- .</u>	<u>Peptide: Pro-Glu -Ile-Val- His- Trp- Pro-...</u>
Type of mutation: Effect of the mutation:	

Normal non-transcribed strand of DNA with corresponding peptide	Mutant non-transcribed strand of DNA with corresponding peptide
<u>DNA :CCA-GAG-ACG-TAC-ACT-GGC-CAC</u>	<u>DNA: CCA-TGA-GAC-GTA-CAC-TGG-CCA-C</u>
<u>Peptide: Pro-Glu-Thr-Tyr -Thr-Gly-His....</u>	<u>Peptide: ...Pro</u>
Type of mutation: Effect of the mutation:	

Normal non-transcribed strand of DNA with corresponding peptide	Mutant non-transcribed strand of DNA with corresponding peptide
<u>DNA: CCA-GAG-ACG-TGG-ACT-GGC-CAC</u>	<u>DNA: CCA-GAG-ACG-TGA-CTG-GCC-AC</u>
<u>Peptide: ...Pro-Glu-Thr-Trp-Thr-Gly-His....</u>	<u>Peptide: ...Pro-Glu-Thr</u>
Type of mutation: Effect of the mutation:	

Exercise 2:

β globin polypeptides constitute the iron binding protein (Hemoglobin). Hemoglobin is a hetero protein found in the red blood cells which is responsible for transportation of respiratory gases. Some allelic mutations may result in the gene coding for β globin to different types of Thalassemia. This mutation leads to the appearance of dysfunctional hemoglobin molecules that cannot bind iron properly.

Case	Codon	Mutation	Site	Clinical Signs
1	5	Substitution	3 rd Nitrogenous base by A	Normal hemoglobin
2	9	Substitution	2 nd Nitrogenous base by A	Sickle cell anemia
3	9	Substitution	1 st Nitrogenous base by T	Mild Thalassemia
4	19 20	Deletion Substitution	3 rd Nitrogenous base 1 st Nitrogenous base by A	Severe Thalassemia

The table below represents the types of Thalassemia presents in Lebanon.

The following DNA segment represents the transcribed strand that codes for normal B globin:

CGG TTT TAC CAC GTG GAC TGA GGA CTC CTC TTC AGA CGG CAA TGA
1 2 3 4 5 6 7 8 9 10 11 12 13 14 15
CGG GAC ACC CCG TTC CAC.....
16 17 18 19 20 21

a-Determine the amino acid sequence coded by this normal gene.

b- By referring to the table, determine the type of Thalassemia in a person showing in his gene the following DNA sequence.

CGG TTT TAC CAC GTG GAC TGA GGA CAC CTC TTC AGA CGG CAA TGA
1 2 3 4 5 6 7 8 9 10 11 12 13 14 15
CGG GAC ACC CCG TTC CAC.....
16 17 18 19 20 21

c-Determine the amino acid sequence for a person having severe Thalassemia, knowing that the mutation occurs within his gene doesn't lead to a change in the reading frame.

d-How can you explain the clinical sign of the first case.

Exercise 3: Genetics and Cancer

Billions of cells of the organism, which have a limited life span, are constantly renewed by cell divisions, these divisions are controlled by a regulatory system. The dysfunction of this regulatory system can produce a clone of cells, forming a tumor.

Cancer is the proliferation of abnormal and "sick" cells in one part of the body. When they develop and contaminate surrounding tissues and organs, we talk about metastasis.

1- Pick out from the text:

1-1- The cause of tumors.

1-2- The definition of cancer

1-3- The definition of metastasis.

To understand the origin of a type of cancer, several studies have been carried out concerning the p53 gene, which codes for the p53 protein. This protein is involved in the regulation of cell divisions.

Document 1 shows the nucleotide sequence of the non-transcribed strand of the two alleles (the normal allele p53+ and the mutant one p53-) involved in this study.

Gene p53	Nucleotid sequence of the nontranscribed strand of DNA	
N° of the codon	↓244	↓250
Allele p53+	GGC GGC ATG AAC CGG AGG CCC	
allele p53-	GGC GGC ATG AAC CGG AGT CCC	

Document1

2- Specify the type of mutation at the origin of this cancer.

3-Determine, using the table of the genetic code, the peptide sequence coded by each of the two alleles.

4- Explain how the modification of the nucleotide sequence leads to the appearance of tumors.