LEBANESE UNIVERSAL ACADEMY

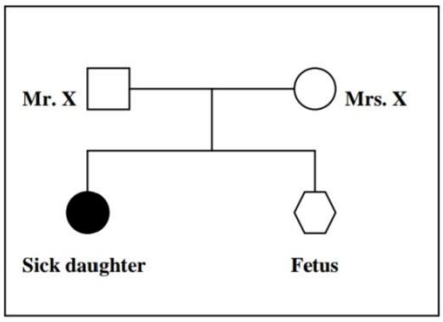
Class: LS
Subject: Biology
Duration: 90 min.

Answer the following questions.

Exercise1: (5 points)

Mr. and Mrs. X have a daughter suffering from sickle cell anemia, document 1. This hereditary sickness, whose mode of transmission is autosomal, is characterized by an abnormality in the β-globin molecule which leads to the deformation of the red blood cells. Mrs. X is pregnant and the couple demand prenatal diagnosis to know if their second child will be affected by sickle cell anemia.

1- Specify if the allele responsible for the sickle cell anemia is dominant or recessive.
2- Indicate the genotype of Mr. and Mrs. X and that of their sick daughter. Justify the answer.
3- Determine the genetic risk for the fetus to be affected by sickle cell anemia.



Document 1

Document 2 reveals the sequences of parts of the non-transcribed strands of the β-globin alleles: HbA is the normal allele while HbS is the mutant allele of the β-globin gene responsible for sickle cell anemia.
A direct diagnostic method by radioactive probe is done for this family. Many copies of the parts of the
β-globin gene can be obtained from the DNA of each person by this technique. These copies are separated in two lots, and each lot is placed in the presence of a different radioactive probe, document 3; each probe is capable to bind with either allele HbA or HbS. The results of autoradiography are shown in document 4.

Position of the nucleotide	1 20
HbA	**CTCCTGAGGAGAAGTCTGCC**
HbS	**CTCCTGTGGAGAAGTCTGCC**

Document 2

Probe n°1	**GAGGACACCTCTTCAGACGG**
Probe n°2	**GAGGACACCTCTTCAGACGG**

Document 3

	Mr. X	Mrs. X	Daughter	Fetus
Probe n°1	▬▬	▬▬	▬▬	
Probe n°2	▬▬	▬▬		▬▬

Document 4

4- Specify, based on document 2, the location of the mutation and its type. Justify the answer.
5- Determine, in reference to documents 2 and 3, which allele corresponds to each probe used.
6- Do the results of document 4 confirm the genotypes you have indicated in question a? Justify the answer. Draw out the genotype and the phenotype of the fetus.
7- Justify why prenatal diagnosis is more accurate than a pedigree in detecting a genetic disease.

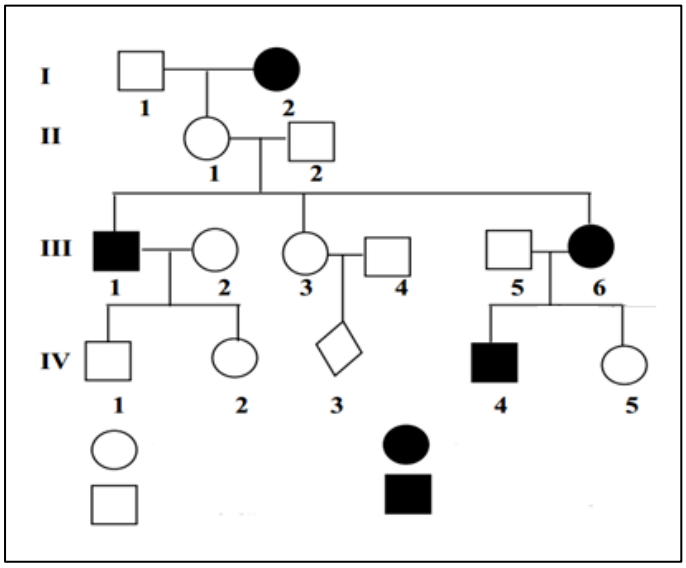
Exercise 2: (5points)

Albinism is a hereditary disease caused by the absence of melanin, a pigment responsible for hair color. Document 1 represents the pedigree of a family where some members are affected by the disease.

1- Specify if the allele responsible for albinism is dominant or recessive.
2- Determine the localization for the gene of albinism.
3- Indicate the genotypes of individuals I-1, II-1, and III-6.

In the population where this family lives, among each 100 normal individuals 15 are heterozygous.

4- Determine the genetic risk for the fetus IV-3 to be albino**.**



Document 1

Researchers have identified gene E1 coding for enzyme E1 and gene E2 coding for enzyme E2. These enzymes are essential for the synthesis of melanin according to the following reactions:

Precursor____**Enzyme 1**____Tyrosine____**Enzyme 2**_____ Melanin

To find the exact origin of albinism in this family, the researchers determined the nucleotide sequences of specific parts of the non-transcribed DNA strand of gene E1 (document 2a) and gene E2 (document 2b) for a normal individual and an albino individual of this family.

	Non-transcribed DNA strand of gene E1 1 2 3 4 5 6 7 8 9
Normal individual	….ACG AGG CCT ACG GGC TTA TGG GGC GAA…
Albino individual	….ACG AGG CCT ACG GGC TTA TGG GGC GAA…

Document 2a

	Non-transcribed DNA strand of gene E2 1 2 3 4 5 6 7 8 9
Normal individual	…ATC ATG CGA ACC GGC TGC TCA AAC CCA...
Albino individual	…ATC ATG CGA ACC GGC TGC TGA AAC CCA...

Document 2b

3. Show that the gene responsible for albinism in this family is gene E2.
4. Determine, using the genetic code (document 3), the amino acid sequence of enzyme E2 that corresponds to each of the two individuals, the normal and the albino.
5. Explain how the modification in the nucleotide sequence of the allele coding for enzyme E2 leads to albinism in this family.

GOOD LUCK.