This is a Class 1 exam

Lebanon Evangelical School for Boys and Girls

Lebanon Evangelical School

for Boys and Girls Loueizeh - Baabda

Stage Exam 01

06NOV2023

Do NOT write in this box. Your MARK will be placed here.

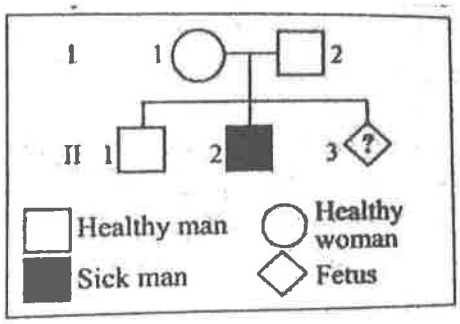
Subject :	Biology	
Class :	12E	
Teacher :	Christine Khalil	
Duration :	1 hour	
Your name :		*Time taken to complete exam*
Your Class :		
Exam Room :		
How many sheets, including cover sheet :		

- Do not open this paper until the teacher in charge instructs you to do so.
- The most important thing to remember in taking an exam is NEVER PANIC.
- If you are caught cheating in this exam, you will be awarded zero for ALL exams that are taken on this day.

Sickle Cell Anemia: A Genetic Disease

Sickle cell anemia is a severe hereditary autosomal disease caused by the synthesis of a non-functional hemoglobin HBS.

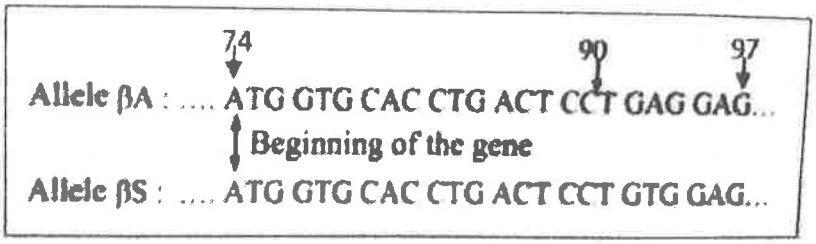
Document 1 presents the pedigree of a family having a child affected by sickle cell anemia.



Document 1

1. Is the allele responsible for the disease dominant or recessive? Justify your answer. (1.5)
2. Indicate the possible genotypes of I 1, II 1 and II 2. (1.5)

Document 2 presents the non-transcribed nucleotide sequences of two parts of the two alleles of the beta globin gene: allele βA coding for normal hemoglobin HBA and allele βS coding for abnormal hemoglobin HBS.

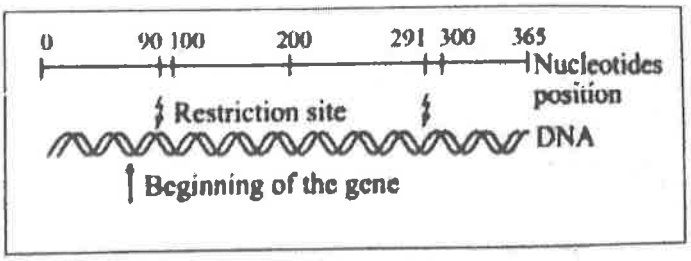


Document 2

3. After comparing the two nucleotide sequences presented in document 2 and their corresponding amino acid sequences, explain the origin of sickle cell anemia. (Table is provided at the end) (4)

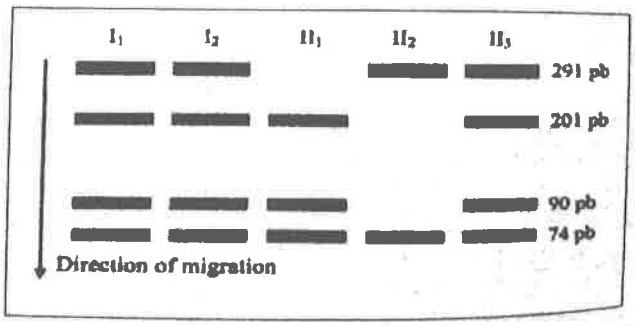
A restriction enzyme, Dde I, cuts the DNA between C and T at the level of the recognition site CTGAG. Two DNA fragments of 365 nucleotides each, one carrying a part of the allele βA and the other carrying a part of the allele βS, are cut with this enzyme. The restriction sites corresponding to the allele βA are presented in document 3.

4. Determine, by referring to documents 2 and 3, the number and the length of the restriction fragments obtained from the alleles βA and βS. (2)



Document 3

Couple I 1 and I 2 asked for prenatal diagnosis to know if their fetus would be affected. The DNA of each member of this family was isolated and cut by the enzyme Dde I. The obtained electrophoregrams are presented in document 4.



Document 4

5. Do the electrophoregrams confirm the genotypes of II 1 and II 2? Justify your answer. (2)
6. Draw out the genotype and the phenotype of the fetus. (2)
7. Once II 1 gets married, is there any risk of him having a child suffering from this disease? (2)

Second letter

First letter	U	C	A	G	Third letter
U	UUU Phe, UUC Phe, UUA Leu, UUG Leu	UCU Ser, UCC Ser, UCA Ser, UCG Ser	UAU Tyr, UAC Tyr, UAA Stop, UAG Stop	UGU Cys, UGC Cys, UGA Stop, UGG Trp	U C A G
C	CUU Leu, CUC Leu, CUA Leu, CUG Leu	CCU Pro, CCC Pro, CCA Pro, CCG Pro	CAU His, CAC His, CAA Gln, CAG Gln	CGU Arg, CGC Arg, CGA Arg, CGG Arg	U C A G
A	AUU Ile, AUC Ile, AUA Ile, AUG Met	ACU Thr, ACC Thr, ACA Thr, ACG Thr	AAU Asn, AAC Asn, AAA Lys, AAG Lys	AGU Ser, AGC Ser, AGA Arg, AGG Arg	U C A G
G	GUU Val, GUC Val, GUA Val, GUG Val	GCU Ala, GCC Ala, GCA Ala, GCG Ala	GAU Asp, GAC Asp, GAA Glu, GAG Glu	GGU Gly, GGC Gly, GGA Gly, GGG Gly	U C A G

Bareme for 12E SE01

1. The couple I 1 and I 2 is phenotypically normal but they have an affected boy II 2 who should have received the affected allele from at least 1 of his parents. One of his parents at least possesses the affected allele but is not expressed in his/their phenotypes indicating that the allele responsible for this disease is masked in the parents and therefore is recessive.
   N: normal allele, dominant.
   a: affected allele, recessive. (1.5)
2. I 1: Na II 1: NN or Na II 2: aa (1.5)
3. The allele sequences of beta A and beta S are identical in all positions except at position 93 where nucleotide A in beta A was replaced by T in beta S.
   mRNA of beta A: AUG GUG CAC CUG ACU CCU GAG GAG
   amino acid Met Val His Leu Thr Pro Glu Glu
   sequence of beta A

   mRNA of beta S: AUG GUG CAC CUG ACU CCU GUG GAG
   amino acid Met Val His Leu Thr Pro Val Glu
   sequence of beta S

   The amino acid sequence of beta A is identical to that of beta S in all positions except in amino acid n7 where Glu in beta A was replaced by Val in beta S. This change leads to the formation of a protein that is not functional. Therefore, people having the beta S allele will have an unfunctional hemoglobin leading to the appearance of sickle cell anemia. (4)
4. In beta A: two restrictions sites are found: the first at 90 pb and the second at 291 pb. The enzyme will cut at these levels and generates 3 fragments:
   The first from 0 to 90= 90 pb. The second: 291-90=201 pb. And the third: 365-291=74 pb
   In beta S: a mutation changed the restriction site found at 90 pb. Therefore, the allele beta S only has one restriction site at 291 pb. The enzyme will cut at this site and generate 2 fragments:
   The first from 0 to 291=291 pb. The second 365-291= 74 pb. (2)
5. The 3 fragments 90, 201 and 74 pb correspond to HBA which is the normal allele while the 2 fragments 291 and 74 pb correspond to HBS which is the affected allele.
   II 1 has two of each of the following bands at the levels 74, 90 and 201 pb which are fragments corresponding to the normal allele indicating that II 1 is homozygous normal NN which is one of genotypes suggested in question 2.
   II 2 has two of each of the following bands at the levels 74 and 291 pb which are fragments corresponding to the affected allele indicating that II 2 is homozygous affected aa which is compatible with the genotypes suggested in question 2. (2)
6. The fetus has the following bands: 74, 90 and 201 pb corresponding the normal allele and another 74 pb with a 291 pb corresponding to the affected allele. Therefore, the genotype of the fetus is Na and he will be normal. (2)
7. Since II 1 is homozygous normal, whatever the genotype of his wife, his children will receive N from him. Whatever they receive from their mother, since N is dominant, they will be normal. (2)