This is a Class 1 exam

Lebanon Evangelical School
for Boys and Girls
Loueizeh - Baabda

SE02 Exam

15DEC2025

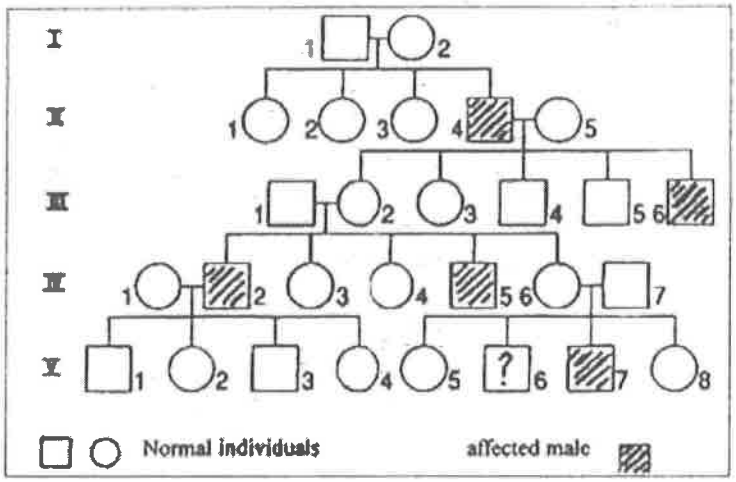
Do NOT write in this box. Your MARK will be placed here.

Subject :	Biology	
Class :	12E	
Teacher :	Christine Khalil	
Duration :	1 hour	
You are to answer the questions to this exam on : Lined paper		
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.

Hemophilia A is a lethal hereditary disease characterized by an abnormality in the blood clotting mechanism and results in the absence/ abnormalities of a protein involved in blood clotting, the factor VIII.

The pedigree of document 1 shows the transmission of this disease in a family.



Document 1

1. Indicate if the allele responsible for hemophilia is dominant or recessive. (1)
2. Determine the chromosomal localization of the hemophilia gene. (1.5)
3. Specify the genotypes of individuals III-3, III-4 and V-5. (1.5)
4. Knowing that the phenotype of individual V-6 is unknown, determine his risk to have hemophilia. (0.75)

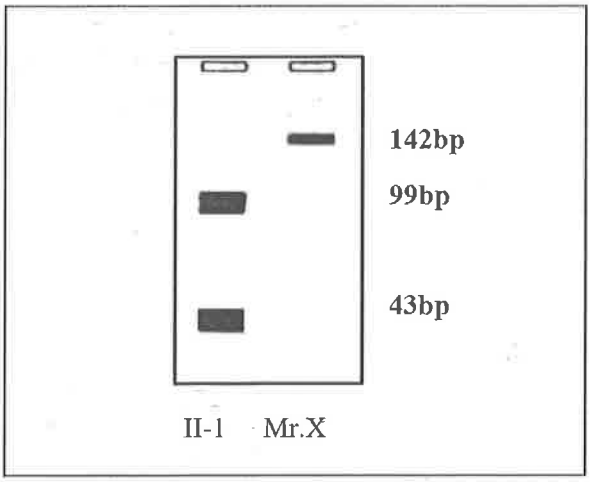
A 142 bp portion of the gene leading to this disease is polymorphic at a restriction site for the Bcl I enzyme. This enzyme is unable to cut the 142 bp DNA portion of the mutant allele while it cuts that of the normal allele into 2 pieces of 99 and 43 bp. Document 2 shows the restriction site of the enzyme Bcl I.



Document 2

5. Justify that the mutation leading to hemophilia A is a substitution mutation of one of the 4 nucleotides A, T, C and G. (1.5)

II-1 married a hemophilic man, Mr. X, and are expecting a child. Their restriction maps are presented in document 3.



Document 3

6. Determine the risk of this couple to have an affected child. (1)

II-1 gave birth to a girl having sexual abnormalities: atrophy of the ovaries, poorly developed breasts, in addition to being affected by hemophilia.

7. Formulate a hypothesis concerning the birth of this baby girl which is hemophilic with sexual abnormalities. (0.75)
8. Determine in this girl:
   a. The parental origin of hemophilia. (1)
   b. The parental origin of the sexual abnormalities. (1)

Answer Key for SE02 12E

1. The couple I-1 and I-2 are phenotypically normal but have an affected child II-4 who should have received the mutant allele from at least one of his parents. At least one of his parents has the mutant allele masked. Therefore, the allele responsible for the disease is recessive.
   Let N be the symbol of the normal dominant allele. Let m be the symbol of the mutant recessive allele. (1)
2. In this pedigree, only boys are affected. Therefore, the gene is definitely not autosomal and not located on the homologous segment of X and Y. If the gene is located on the non-homologous segment of Y, boy II-4 who is affected and of genotype XYm should have received Ym from his father I-1 who should have been of genotype XYm and be affected but this is not the case. Therefore, this disease is X-linked. (1.5)
3. III-3: XN Xm, she is normal so she should have the normal allele and she should have received the X chromosome carrying the mutant allele from her affected father.
   III-4: XN Y, he is a normal male, he expresses the only allele he has on the X chromosome.
   V-5: XN XN or XN Xm, the normal allele is dominant, it can be expressed in the homozygous or in the heterozygous state. She received XN from her normal father and could have received XN or Xm from her heterozygous mother. (Each is 0.5)
4. Mother IV-6 is heterozygous with genotyope XN Xm. Boy V-6 necessarily took Y from his father and has a risk of ½ to take Xm from his mother thus making him affected. (0.75)
5. The sizes of the 2 fragments of the normal allele are 99 and 43 bp. Their total leads to 142 bp which is equal to the size of the fragment of the mutant allele. Since the size didn't change after the mutation, then this is not a deletion nor an insertion; it is a substitution mutation. The restriction site of the enzyme where the mutation is located contains all kinds of nucleotides A, T, C and G so at least one of these nucleotides has been replaced by mutation. (1.5)
6. II-1 is a girl and she only has the 2 fragments 99 and 43 bp corresponding to the normal allele. So, she has two normal alleles and she is of genotype XN XN. Whether the child is a boy or a girl, he/she will receive XN from the mother. Therefore, whatever he/she receives from the father, he/she will still be normal since the normal allele is dominant. The risk of having an affected child is 0. (1)
7. Hypothesis: The girl suffers from a gonosomal abnormality and may have 1 X chromosome instead of 2. (0.75)
8. Since this disease is lethal, the girl can't be of genotype Xm Xm or she would have died before birth. She survived with several sexual abnormalities which usually characterize girls with Turner syndrome. Since she is affected, she must be of genotype Xm. Her mother is of genotype XN XN so she can't give her the mutant allele. She must have received Xm from her affected father of genotype Xm Y. Therefore, the parental origin of hemophilia is paternal. (1)
9. The father's meiosis occurred normally and he gave his daughter Xm. On the contrary, the mother should have had an abnormal meiosis leading to a gamete lacking a gonosome leading to the girl only having Xm that she received from her father. She suffers then from Turner syndrome which is characterized by sexual abnormalities. Therefore, the parental origin of these abnormalities is maternal. (1)