This is a Class 1 exam

Lebanon Evangelical School
for Boys and Girls
Loueizeh - Baabda

SE01 Exam

10NOV2025

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

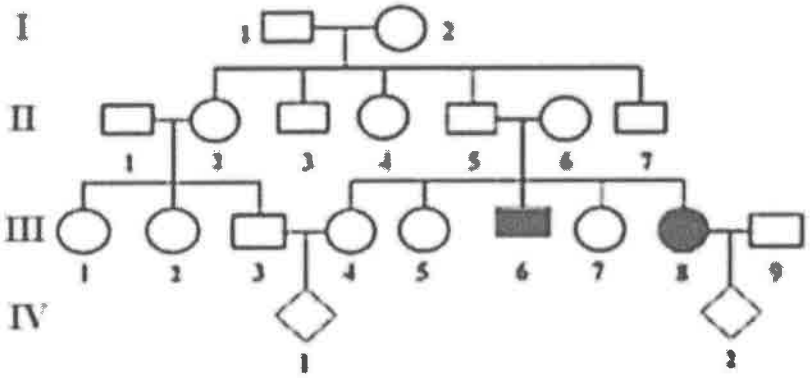
Subject :	Biology	
Class :	12E	
Teacher :	Christine Khalil	
Duration :	1 hour	
You are to answer the questions to this exam on : Lined paper		
Your name :		*Time taken to complete exam*
Your Class :		
Exam Room :		
How many sheets, including cover sheet :		

- 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.

Fructosemia or hereditary fructose intolerance was first clinically described in 1956. It's a rare disease occurring in 1 out of 10000 births worldwide and the estimated probability to be heterozygous in the human population is 1/50.

Fructosemia is a congenital/hereditary disease due to a deficiency in an enzyme called aldolase B located in the liver, small intestine and the kidneys. This disease can lead to the buildup of toxic substances in the liver and kidneys which leads to their damage with time.

Document 1 shows the pedigree of a family whose certain members are affected by fructosemia.



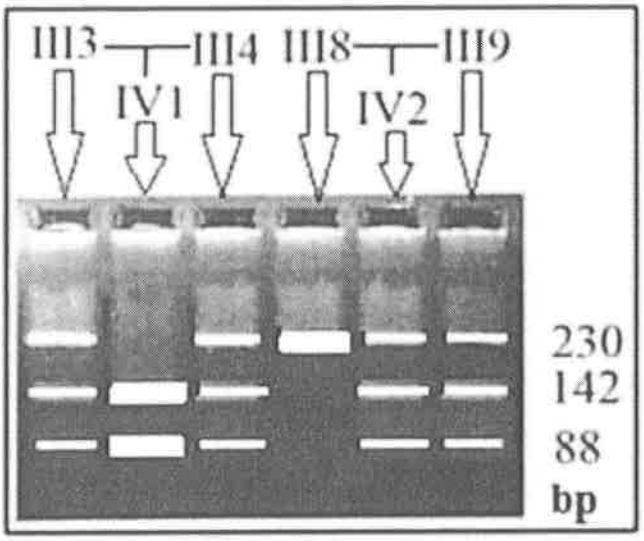
Document 1

1. Determine if the allele responsible for fructosemia is dominant or recessive. (1)
2. Determine the chromosomal localization of the gene responsible for fructosemia. (1.5)
3. Determine the risk for the fetus IV-2 to be affected by this disease. (2)
4. Show that the risk for the fetus IV-1 to be affected by this disease is higher than 1/10000. (2)
5. Why is the risk of fetus IV-1 higher than the worldwide risk? (0.75)

The mutations that can be detected by RFLP after addition of ethidium bromide either lead to the appearance of a new restriction site for a certain enzyme and are indicated by the name of the enzyme preceded by a + sign OR lead to the disappearance of a restriction site of that enzyme and are indicated by the name of the enzyme preceded by a – sign.

A DNA fragment of the aldolase gene is tested by RFLP using a restriction enzyme called BanI. The results of the electrophoresis for some family members are shown in document 2.

6. Identify the band (s) corresponding to the normal allele and the one (s) corresponding to the mutant allele. (1.5)
7. Indicate the genotypes and the phenotypes of both fetuses IV-1 and IV-2. Justify your answer. (2)
8. Is the mutation leading to this disease a +BanI or a -BanI? Justify your answer. (1.25)


III3
III4
III8
III9
IV1
IV2
230
142
88
bp

Answer Key SE01 12E

1. The couple II-5 and II-6 are phenotypically normal but have affected children III-6 and III-8 who should have received the affected allele from at least one of the parents. One of the parents at least have the mutant allele masked. Therefore, the allele responsible for the disease is recessive. N: normal dominant m: mutant recessive (1)
2. If the gene is located on the non-homologous segment of Y, boy III-6 who is affected and of genotype XYm should have received Ym from his father II-5 who should have been of genotype XYm and should have been affected but that's not the case. (1.5)
   If the gene is located on the non-homologous segment of X, girl III-8 who is affected and of genotype XmXm should have received Xm from her father II-5 who should have been of genotype XmY and should have been affected but that's not the case.
   If the gene is located on the homologous segment of X and Y, boy III-6 who is affected and of genotype XmYm should have received Ym from his father II-5. His sister III-8 who is affected and of genotype XmXm should have received Xm from her father II-5. Their father should have been of genotype XmYm and should have been affected but that's not the case. Therefore, this gene is autosomal.
3. Father III-9 has no family history, his probability to be heterozygous is 1/50 and his risk of giving the mutant allele is ½. Mother III-8 is affected of genotype mm. She will necessarily give the mutant allele to the fetus so her entire part is 1.
   P(fetus IV-2 to be affected)= 1/50 x1/2 x1= 1/100 (2)
4. Mother III-4 has a 2/3 probability to be heterozygous since she has affected siblings. Her probability to give the affected allele is ½. Father III-3 has no family history, his probability to be heterozygous is 1/50 and his risk of giving the mutant allele is 1/2.
   P(fetus IV-1 to be affected)= P(F to be heterozygous)x P(F to give the mutant allele) x P(M to be heterozygous) x P(M to give the mutant allele)= 1/50 x ½ x 2/3 x ½ = 1/300 which is higher than 1/10000. (2)
5. The parents of fetus IV-1 are cousins. Marriage within the same family increases the risk of having an affected child since both the mother and the father will have a possibility of carrying the same recessive mutant allele. (0.75)
6. Female III-8 is affected and since the mutant allele is recessive, she will definitely be mm. Document 2 shows a thick band at 230 bp which corresponds to 2 mutant alleles. Therefore, the 230 bp band corresponds the mutant allele. All the other individuals are normal having in addition to the 230 bp bands at the level of 142 and 88 bp which both correspond to the normal allele. (1.5)
7. Fetus IV-1 has a thick band on 142 bp and another on 88 bp which both correspond to the normal allele therefore he will be NN and normal. Fetus IV-2 has a band on 230 bp which corresponds to the mutant allele and a band on each of 142 bp and 88 bp which correspond to the normal allele therefore he will be Nm and normal since the normal allele is dominant. (2)
8. Bands 142 bp and 88 bp correspond to the normal allele and their sum is 230 bp which is equal to the length of the band corresponding to the mutant allele. The normal allele is cut within these 230 bp to yield these 2 fragments while the mutant allele won't be cut due to the absence of the restriction site of enzyme BanI within the 230 bp. Therefore, this mutation is –BanI. (1.25)