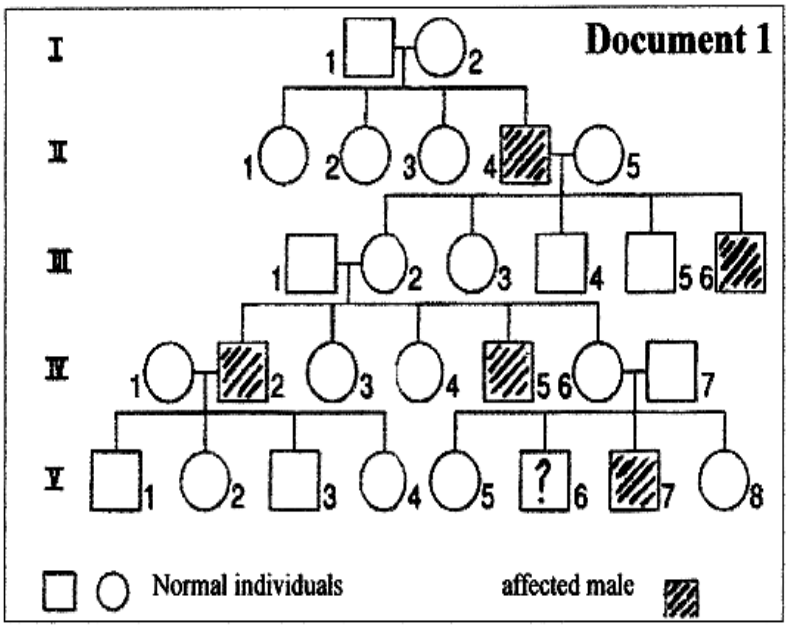
ثانوية الصويري الرسمية/1497	امتحان : السعي الثاني	التاريخ:23/1/2024
رقم الهاتف 08/565075	المادة: علوم الحياة	العلامة:/10
محافظة البقاع/قضاء البقاع الغربي	الصف:الثالث الثانوي علوم حياة	المدة: 45 min
	أستاذ المادة: نجمة أبوياسين	

Exercise 1(10 pts) **The Transmission of Hemophilia in Humans**

Hemophilia A is a hereditary disease characterized by an anomaly in the mechanism of blood coagulation and is due to absence or anomalies of a protein implied in the coagulation, the factor VIII. A portion of 142 bp of the gene of this illness is polymorphic at the level of a restriction site for the enzyme Bcl I, if the enzyme recognizes the site, it cuts the sequence of DNA in two pieces of 99 and 43 bp.

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



1- Determine the mode of the transmission of the gene of hemophilia.
2- Determine the genotypes of the individuals: III_3, III_4, IV_5 and V_5.

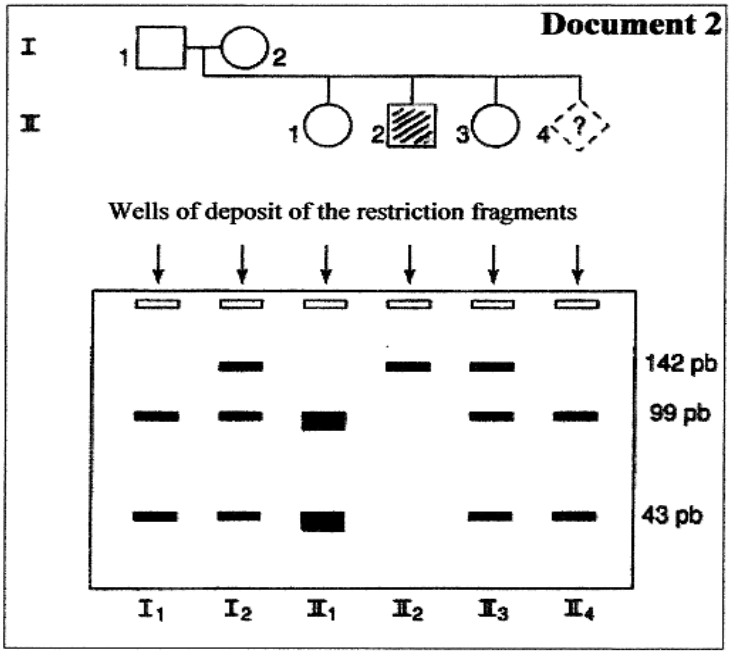
Knowing that the phenotype of the individual V_6 is unknown.

3- Indicate the risk for him to be hemophilic.

Giving that the frequency of heterozygous females in the population is 1/40:

4- Determine the risk for the couple IV (1-2) to have an affected child.

The individual V_1 married the individual V_5, their pedigree appears in the document 2 with the results of the electrophoresis of the portion of the gene mentioned above. Either the normal allele or the allele of the disease has the restriction site mentioned above.



Basing on the above text and on the results of the electrophoresis of the document 2:

5- Specify the fragments corresponding to the normal allele and to the mutant allele.
6- Specify if the mutation had created or suppressed a restriction site for the enzyme Bcl I.

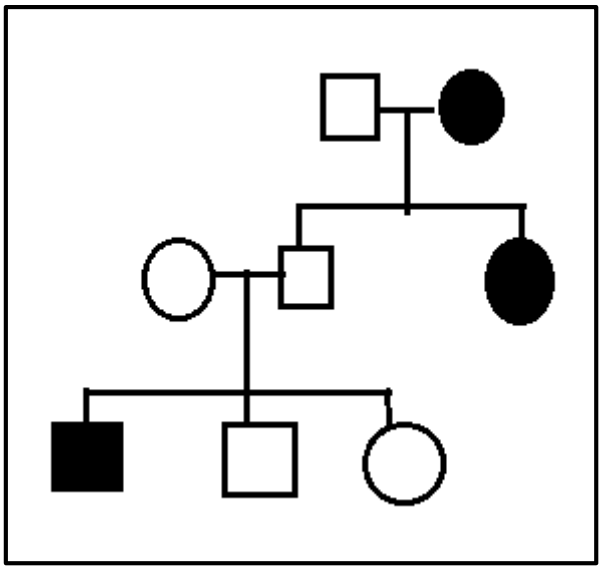
(**Bonus**)A hemophilic girl is born in this family. This result originates from an abnormal meiosis in the father or the mother.

1- Identify the meiosis of the parent at the origin of the birth of this girl.

(Bonus)

A pedigree of a family affected by a hereditary disease is shown in the adjacent document.

1- Indicate if the allele of the disease is dominant or recessive.
2- Show that this disease is autosomal.



Answer key

Exercise 1(10 pts) The Transmission of Hemophilia in Humans

1- Since the parents I-1 and I-2 are normal but they have an affected child II-4, therefore at least one of the parents carry the allele of the disease but it is masked by the normal allele. So the allele of disease is recessive.
Let N be the symbol of the normal allele and d be the symbol of the mutant allele.

Since the pedigree shows only affected males and no affected females, then the diseases is neither autosomal nor X and Y linked (homologous segment of X and Y). Therefore, the disease is sex linked. Moreover, if the disease is Y- linked (present on non-homologous segment of Y), then the affected boy II-4 should inherit Y^d from his father I-1 that should be $X\ Y^d$ affected, but he is normal so it is not the case.

Therefore, the diseases is X-linked (present on the non- homologous segment of X).

2- III_3: X^NX^d , normal heterozygous, since she is a normal female so she has X^N , but she has an affected father who must have transmitted X^d to her.

III_4: **X^NY**, normal, since he is a normal male thus he should carry the allele N on X chromosome.

IV_5 : **X^dY**, since he is an affected male thus he should carry the allele m on X chromosome.

V_5: **X^NX^N or X^NX^d** , since she is normal so she necessarily has X^N and the normal allele is dominant so it can be expressed both in homozygous and heterozygous states.

3- **risk to have affected boy=** probability for the mother to be heterozygous× probability for the mother to transmit the allele of the disease × probability of the father to transmit Y
His father is X^NY normal, he necessarily transmits Y to V-6 since he is a male.
His mother is heterozygous **X^NX^d** since she is normal and she has affected boy V-7 who must have inherited X^d from her. So her probability to be heterozygous is 1 and her probability to transmit the allele of the disease is ½.
Risk= 1×1/2×1=1/2.

4- **Risk= risk to have affected girl + risk to have affected boy** = (probability for the mother to be heterozygous× probability for the mother to transmit the allele of the disease × probability of the father to transmit the allele of the disease) + (probability for the mother to be heterozygous× probability for the mother to transmit the allele of the disease × probability of the father to transmit Y)
The mother is normal she could be either **X^NX^N or X^NX^d .** if she was homozygous, then she will give the normal dominant allele to all of her children who will all be normal, so the risk will be zero.
Her probability to be heterozygous is 1/40 since she has no family history. Her probability to transmit the allele of the diseases is ½.
The father is affected X^dY so his probability transmit the allele of the disease is ½ for the baby to be a girl.
His probability to transmit Y so the baby could be a boy is ½.
Risk= (1/40×1/2×1/2)+(1/40×1/2×1/2)= 1/80.

5- Fragment 99 and 43bp correspond to the normal allele since the male I-1 is normal X^NY so he has only the normal allele and he has two bands on the gel of sizes 99 and 43bp.
Fragment 142bp corresponds to the allele of the disease since the male II-2 is affected X^dY so he has only the allele of the disease and he has one band on the gel of size 142bp.

6- The mutation had suppressed a restriction site for the enzyme Bcl I since the considered portion of the gene is 142bp and if it recognized by the enzyme it will be cut into two fragment 99 and 43bp which are the same fragments corresponding to the normal allele, then the normal allele contains a restriction site. While the mutant allele remains 142bp so it lacks a restriction site.

Bonus 1:

Since the father in this family is normal so all of his daughters must be normal but this girl is born hemophilic, therefore, she did not receive X^N from her father and received only an X^d from her mother. So there was nondisjunction of chromosomes in her father during meiosis 1 or 2.

Bonus 2:

1- Recessive.
2- If the gene of the disease is localized on the non homologous segment of X, then the female which is affected should be X^cX^c receiving X^c from her father that should be X^cY ,affected, but it is not the case.

If the gene of the disease is localized on the non homologous segment of Y, then the male which is affected should be XY^c receiving Y^c from his father that should be XY^c ,affected, but it is not the case.

If the gene of the disease is localized on the homologous segment of X and Y, then the female which is affected should be X^cX^c receiving X^c from her father that should be XcY^N since he is normal. He should transmit Y^N to his normal son who also receives X^c from his affected mother. In turn, this male must transmit Y^N to his sons who must all be normal but it's not the case.

Therefore, the gene is autosomal.