HaretHreik Official School For Girls

Governate: Mount of Lebanon
District: Baabda

Academic Year: 2016-2017
Number: 1611

Name:
Class: G12(LS)
Subject: Biology

Midterm Exam

Duration: 180 min
Points: / 20
Date: 17 / 1 / 2017

Exercise 1: 5pts **Hemophilia A**

Hemophilia A is a hereditary disease determined by a recessive allele; it is due to an abnormality of blood clotting factor VIII. Document 1 shows the transmission of hemophilia in a certain family. Document 2 shows the number of alleles of the gene coding for this disease in the members of this family:

1. Determine, from documents 1 and 2, the chromosomal location of the gene responsible for hemophilia.
2. Identify the sex of the fetus.
3. Determine the risk of the fetus to be affected

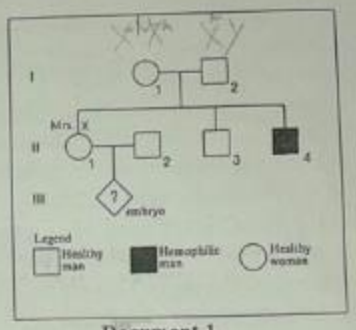
Probe DX 13 is hybridized with a complementary sequence near the gene coding for factor VIII. Scientists consider that it characterizes mutation of this gene. Document 3 shows the restriction sites of the enzyme Bgl II and the hybridization site of probe DX 13.

4. Indicate, by referring to document 3, the number and the size of the fragments obtained for each allele, after the treatment of DNA, by the enzyme Bgl II.

To determine the prenatal diagnosis of the fetus, the doctor proposed DNA analysis for Mrs. X, the fetus and individuals II-3 and II-4 using the Southern blotting technique.

The considered DNA fragment is cut with the restriction enzyme Bgl II . the resulting restriction fragments are separated by electrophoresis and denatured, then, they are hybridized with radioactive probe DX 13. An autoradiography is obtained and the results are presented in document 4:

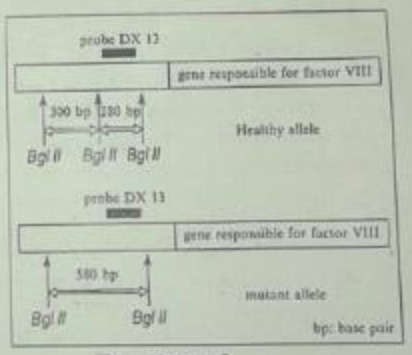
5. Explain why only two fragments are observed for Mrs.X
6. Determine, by referring to document 4, the prenatal diagnosis of the fetus
7. Explain why the diagnosis is not 100% reliable.



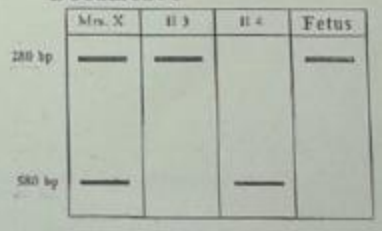
Document 1

Individual	1-1	1-2	II-1	II-2	II-3	II-4	II
Number of alleles	2	1	2	1	1	1	1

Document 2



Document 3

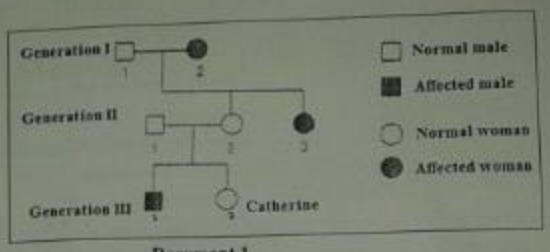


Document 4

Exercise2: 5pts **Retinoblastoma**

Retinoblastom is a kind of cancer characterized by the appearance of tumor in the retina of the eye. The transmission of retinoblastoma in a certain family is shown in document 1:

1. Indicate if the gene responsible for retinoblastoma is dominant or recessive. Justify the answer.
2. Show that retinoblastoma is not a sex linked disease,



Document 1

Normal individuals have a gene on chromosome 13; this gene regulates cell division and suppresses tumor development. In babies and infants affected by retinoblastoma, two types of mutation are detected in this gene. Cancer develops when the individual has two mutated alleles.

A part of the non-transcribed DNA strand of the cancer suppressor gene in Catherine's parents and her brother is shown in document 2. DNA analysis revealed that the father of Catherine has two normal alleles for the cancer suppressor gene.

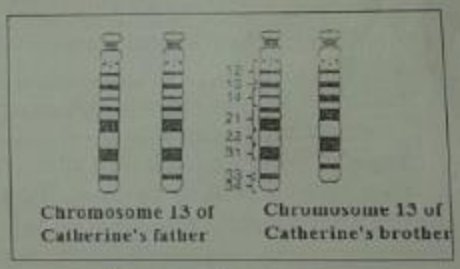
Non-transcribed DNA strand:
Catherine's father: A C T A A C A A A
Catherine's mother and brother: A C T A A T A A A

Document 2

3. Identify the type of mutation detected in the cancer suppressor gene.
4. Indicate the problem posed from the development of the disease in Catherine's brother

Document 3 shows chromosome 13 and its different gene loci in Catherine's brother and father:

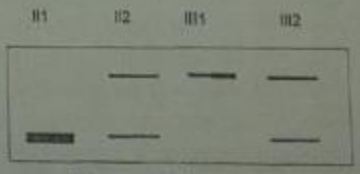
5. Compare the chromosome pair 13 of Catherine's brother to that of his father.



Document 3

Document 4 shows DNA analysis for the cancer suppressor gene on chromosome 13 in different members of the pedigree shown in document 1:

6. Explain, based on what all preceded, the development of the disease in Catherine's brother.



Document 4

Exercise 3:5pts **Determining The Cause Of A Fertility Problem**

After determining the conditions of stimulation of the secretion of LH by the pituitary gland, we perform different incubations in vitro of 4 lots of pituitary cells having the same number of cells.

Document 1 shows the experimental procedure and the results obtained:

Pituitary gland cells

	Lot 1	Lot 2	Lot 3	Lot 4
First phase: incubation during 36 hours in medium A	① ooooo	② ooooo	③ ooooo	④ ooooo
Estradiol in medium A (with concentration > 30 pg.mL^{-1})	No	No	Yes	Yes
Second phase: incubation during 5 hours in medium B	ooooo	ooooo	ooooo	ooooo
GnRH in medium B (0.2 ng.mL^{-1})	No	Yes	No	Yes
Quantity of LH found in medium B at the end of incubation (µg)	< 0.2	0.7	< 0.2	3.3

Document 1

1. Describe the above experiment indicating its results.
2. Interpret the results obtained.
3. Explain, based on what precedes and on your acquired knowledge, the mechanism that leads to LH peak.

Katty is a 30 years old woman consults a doctor for the cause of her sterility. The doctor prescribed measurement of LH level. The results obtained are shown in document 2.

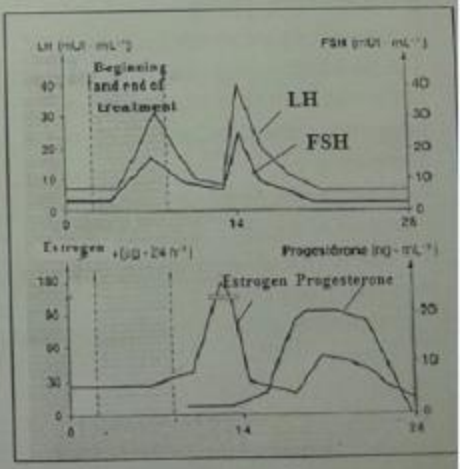
4. Determine the cause of sterility of this woman

Days after the beginning of menstruation	0	4	10	12	14	18	24	28
Amount of LH (mIU.mL)	5.3	6.8	6.3	7	6.2	6	7.3	6.5

Document 2

The measurement of estrogen shows that the level of this hormone remains fluctuating between 29 amd 30.5pg/ml in Katty. So, the doctor proposed clomiphene treatment for Katty. This molecule has a structure analogous to estrogen. It binds to the receptors of estrogen on the hypothalamo pituitary axis and prevents the action of this hormone. The results are shown in document 3.

4. Deduce, from the result obtained during treatment, the origin of Katty's sterility
5. Explain, based on the results obtained in document 2 and document 3, why the treatment was successful for Katty.
6. Propose another treatment that can give the same result.

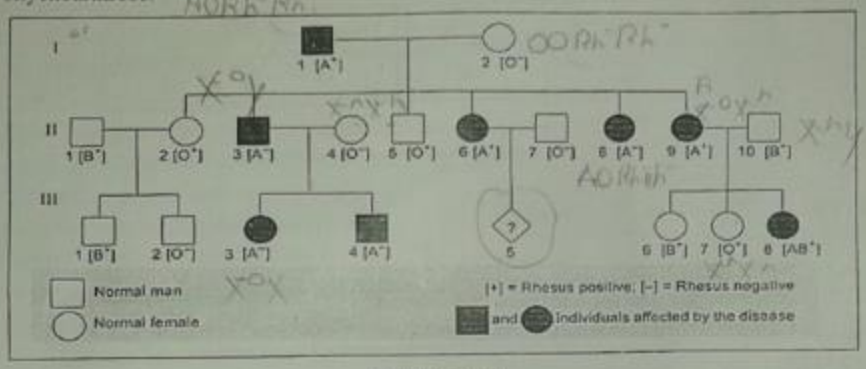


Document 3

Exercise 4: 5pts **Oncho-Arthrose And Blood Groups**

The blood types in ABO system are determined by a gene situated on chromosome 9. This gene has three alleles: A, B and O. alleles A and B are co-dominant but both are dominant over allele O which is recessive. However, the blood types in rhesus system are determined by a gene situated on chromosome1. This gene has two alleles: dominant allele Rh+ and recessive allele Rh-

Onychoarhtrose is a very rare genetic disease characterized by the absence of kneecaps and nail development. Document 1shows the genealogical tree of a family that has some members affected by onychoarthrose.



Document 1

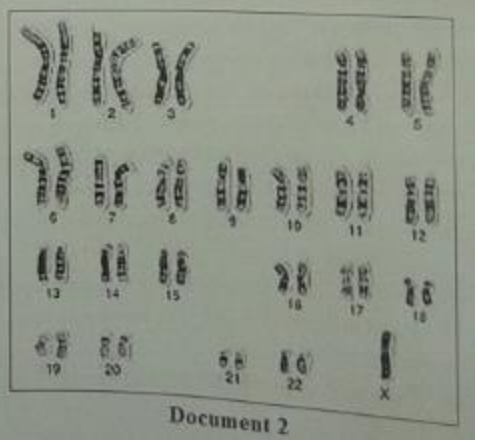
1. Indicate if the alleles responsible for onychoarthrose is dominant or recessive. Justify the answer.
2. Determine the location of the gene responsible for onychoarthrose.
3. Establish the relation between the gene of the disease and that of the ABO system,

Fetus III5 is affected by the disease. His blood type is [O $^{+}$] .

4. Determine the genotype of the fetus and his mother.
5. Explain why the fetus is affected although his blood group is O
6. Illustrate by explanatory drawing the phenomena responsible for the phenotype of the fetus.

Girl III-6 suffers from sexual and physical troubles. She is also color blind like her mother while her father has normal vision. Her karyotype is shown in document 2.

7. Determine the parent responsible for the abnormal phenotype of this girl.
8. 1. Indicate the phase of meiosis at which this abnormality took place,
8. 2. Illustrate this phase by explanatory drawing.



Document 2