مسابقة في علوم الحياة
المدة: ٣ ساعات

الاسم:
الرقم:

Answer the following questions.

Question I (6½ pts)

A-

Document 1 represents the pedigree of a family whose certain members, colored in black, are affected by hemophilia B. This disease is observed only in male individuals. The presence of this gene in two copies in a genotype provokes the death of the embryo.

a- Is the allele responsible for this disease dominant or recessive ? Justify the answer.

b- Is this gene sex-linked ? Justify the answer.

Document 1

c- Indicate the genotype of each of the individuals 8,13 and 14. Justify, for each genotype, the answer.

d- Make the necessary analyses to determine the possible proportions of the descendants of female 5 in each of the following cases:
1- If her husband is not affected by hemophilia.
2- If her husband is affected.

We performed a special technique for the analysis of DNA of the couple 8-9 and their children 12, 13, and 14. We obtained different DNA fragments of variable lengths measured in kilo bases (document 2).

e- By referring to document 2, identify the allele of the hemophilia B.

Document 2

B-

Document 3 presents the karyotype of an individual with an abnormality while document 4 presents the karyotype of a normal individual.

Document 3

Document 4

f- Compare these two karyotypes. What is the abnormality revealed?

Document 5 shows the Karyotype of spermatocyte II that is obtained from a man of a normal karyotype. This spermatocyte II is identical to that, which has allowed the birth of the individual affected by the abnormality (document 3).

g- What information does document 5 provide?

h- Make a labeled diagram of the anaphase stage of the division, which produces this type of spermatocyte II. (Present, only, in the diagram the sex chromosomes X and Y and the pair of chromosomes concerned with the disease).

Document 5

Question II (5pts)

Cancer cells are abnormal cells whose rapid and uncontrolled division frequently leads to the death of the individual. Being a part of the organism, these cells present abnormal markers on their membranes.

In the framework of studying the reaction of the organism against these cells, we perform, on mice, the experiment shown in the adjacent document.

a- In a few lines, describe the performed experiment.

b- Interpret this experiment.

c- By referring to the acquired knowledge, explain the immune mechanism involved in the elimination of the cancer cells by the organism.

d- Formulate a hypothesis, which permits to explain the obtained results after the injection of preparation 2.

Question III (4pts)

Experiment 1: After 25 days from the ovariectomy (castration) of a female monkey, we introduce under the skin an implant of estradiol, which liberates continuously and in a small amount this hormone in the blood. In this way, the plasma concentration of estradiol is maintained during several days, at an amount near to that which normally exists at the start of the follicular phase of the menstrual cycle. We inject, 15 days after placing the implant, a large amount of estradiol (600pg/mL). We measure the plasma concentration of LH in this female during the experiment; the results are shown in document 1.

Experiment 2 : Another female monkey is subjected to the same treatment: castration, introduction of estradiol implant, and the injection of a large amount of estradiol. In addition, we maintain a large amount of progesterone in the blood starting from the beginning of castration. We measure the plasma concentration of LH during this second experiment; the results are shown in document 2.

Document 1 **Document 2**

a- In the same table, represent the variation of the concentration of LH as a function of time, in these two females.
b- Interpret each of these two documents. What can you deduce concerning the role of these two hormones on the variation of the plasma concentration of LH?

Question IV (4½ pts)

Myasthenia is a muscular disease characterized by great fatigue when doing an effort. The motor deficit appears, particularly during a sustained or repeated skeletal muscular activity. When fatigue is at **maximum**, the muscle remains excitable directly by an electric stimulation but it cannot be excited by means of its nerve.
Document 1-A represents the simultaneous recordings of the electric response (a) and the mechanical response (b) of the adductor muscle of the thumb by the stimulation of the cubital nerve at a frequency of 3 per second in an individual having myasthenia.
Document 1-B shows the recordings in the same individual and by the same stimulation done 15 minutes after the injection of 4mg of prostigmine. Prostigmine is an inhibitor of acetylcholinesterase, an enzyme that destroys acetylcholine.

Document 1

a- Compare the recordings before and after the injection of prostigmine.
b- By referring to the given and to the acquired knowledge, how can you explain the results obtained in B?
c- Are the muscle fibers affected in this individual? Justify the answer.
d- The nerves, in individuals having myasthenia, are healthy. Formulate two hypotheses concerning the cause of this disease.

Research has shown that 95% of the individuals having myasthenia have antibodies, which block or destroy the membrane receptors of the acetylcholine.

e- To which type of disease does myasthenia belong?
f- Which of the two hypotheses is valid? Justify the answer.
g- A pregnant woman having myasthenia, gives birth to an infant who presents, at birth, muscular paralysis, that disappears after a few weeks or a few months. How can you explain this particularity?

Answer Key

Question I (6½pts)

a- The allele responsible for hemophilia is recessive because individual 14 is hemophiliac and his parents, 8 and 9 are healthy. The defective allele is present in the parents but is not expressed. (½ pt) Symbols : h for hemophilia and N for healthy or non-hemophilia .

b- Yes, because the presence of this gene in two copies provokes the death of the embryo. The hemophiliac boys thus, possess one allele h which is possible only if the inheritance is sex linked and not autosomal. Therefore, this gene is found on a segment of sex chromosome, which has no homologous segment on the other chromosome. (¾ pt)

c- Individual 8 is X^N/X^h since she is healthy. Being healthy, she must have the allele N, but since her father is sick, he gives her an X^h.
Individual 13 is X^N/Y since he is healthy, his chromosome X carries allele N.
Individual 14 is X^h/Y since he is sick, his chromosome X carries allele h.(1 ¼ pt)

d- The genotype of female 5 is the same as that of female 8: X^NX^h since her father is sick.

1rst case : X^N/X^h x X^N/Y
½ X^N ½ X^h ½ X^N ½ Y

Table

♀ \ ♂	½ X^N	½ Y
½ X^N	¼X^NX^N	¼ X^NY
½ X^h	¼ X^NX^h	¼ X^hY

Analysis of table
¼ X^NX^h healthy carrier girl
¼ X^NX^N healthy girl
¼ X^NY healthy boy
¼ X^hY hemophiliac boy (¾ pt)

2nd case : X^N/X^h x X^h/Y
½ X^N ½ X^h ½ X^h ½ Y

Table

♀ \ ♂	½ X^h	½ Y
½ X^N	¼X^NX^h	¼ X^NY
½ X^h	¼ X^hX^h	¼ X^hY

Analysis of table
X^hX^h hemophiliac girl: born dead
⅓ X^NX^h healthy carrier girl
⅓ X^NY healthy boy
⅓ X^hY hemophiliac boy (¾ pt)

e- The comparison between electrophoreses 9 and 13 of healthy men on one hand and the electrophoresis of individual 14, hemophiliac man, on the other hand, permits to say that the radioactive band of 1.3 kb, which characterizes the allele h, is responsible of hemophilia B. (½ pt)

B-

f- The karyotype of the normal individual shows the sex chromosomes X and Y. All its autosomes are present in pairs.
The karyotype of the affected individual shows the sex chromosomes X and X. All its autosomes are present in pairs except chromosome 21 that exists in three copies. (¼ pt)
Trisomy 21. (¼ pt)

g- Document 5 presents the karyotype of spermatocyte II. Each chromosome exists in one copy except chromosome 21, which exists in two copies. (½ pt)

h- (1pt)

Question II (5pts)

a-

- We obtain cancer cells and serum from mouse $A_1$and we put them in an appropriate culture medium. Five days later, we inject this mixture into mouse A_2 which dies after three months.
- We obtain cancer cells and a few lymphocytes from mouse $A_1$and we put them in an appropriate culture medium. Five days later, we inject this mixture into mouse A_3 which dies after three months.
- We obtain cancer cells and many lymphocytes from mouse $A_1$and we put them in an appropriate culture medium. Five days later, we inject this mixture into mouse A_4 which survives.
- Mouse A_1 dies after three months. (2pts)

b- The presence of cancer cells in mouse A_1 provokes its death after three months. This indicates that cancer is fatal. Similarly, mice A_2 and A_3 die since A_2 received cancer cells and serum from A_1 and A_3 received cancer cells and a few lymphocytes from A_1. This indicates that the serum as well as the limited number of lymphocytes do not protect the mice from cancer. On the contrary, mice A_4, who received cancer cells cultured with many lymphocytes remains alive. Thus, lymphocytes in large quantities destroy cancer cells.(1pt)

c- Tumor cells express on their surface peptides, which are recognized by the TL as foreign bodies. This recognition activates the TL specific to these antigens: TL_4, which secrete interleukins and TL_8, which, after proliferation, differentiate into TLc. These cells will destroy (lyse) the cancer cells. (1½ pt)

d- Hypothesis: The lymphocytes which are specific to the tumoral antigen are not present in this culture medium.

Or

Specific TL are not enough to destroy, in 5 days, all the cancer cells. (½ pt)

Question III (4pts)

a- (1pt)

Plasma concentration of LH (in ng/mL) / Time (in days)	Graph of document 1	Graph of document 2
5	20	5
15	20	5
25	20	5
40	5	5
41	35	5
43	15	5
50	5	5

Variation of the plasma concentration of LH as a function of time

b- We observe that, 25 days after the castration, the amount of LH is constant and equal to 20ng/mL. The introduction of estradiol implant at day 25, provokes a decrease in the amount of LH that reaches 5 ng/mL at day 40. The injection of a large quantity (600pg/mL) provokes a peak of LH (35 ng/mL) within one day then the amount decreases to 5 ng/mL at day 50.
This implies that a small amount of estradiol inhibits the secretion of LH (negative feedback); and in the case of large amount of estradiol there is an LH peak (positive feedback). (1 ½ pt)
On the other hand, we observe in graph B that as we inject estradiol (in small or in large amount) together with a large amount of progesterone, the amount of LH is weak and remains constant at 5 ng/mL.
This implies that progesterone prevents the increase in the amount of LH (negative feedback) whatever the amount of estradiol is. (1 pt)
Therefore, The plasma concentration of LH depends on the amount of estradiol and the presence of progesterone. (½ pt)

Question IV (4½ pts)

a- For a frequency of 3 per second before the injection of prostigmine, the electromyogram reveals that the amplitude of the first response is about 10 mV then this amplitude decreases gradually to become very weak at the 5^{th} stimulation. The same for the mechanical response the amplitude is high (1kg) at the beginning then, it decreases progressively. After the injection of prostigmine and for the same frequency of stimulations, we admit that the amplitude of the electrical and that of the mechanical responses decrease very slightly.(1pt)

b- The acetylcholinesterase is an enzyme that degrades acetylcholine in the synaptic cleft and thus prevents its prolonged fixation on the postsynaptic membrane. After the inhibition of the acetylcholinesterase by the prostigmine, a prolonged fixation of acetylcholine on the receptors is observed which explains the obtained recording. (½ pt)

c- The muscle fibers are not affected because at the maximum of fatigue, the muscle remains excitable directly by an electric stimulation while it cannot be excited by its nerve. (½ pt)

d- 1rst Hypothesis: The quantity of Ach, which fixes on the postsynaptic receptors, is not sufficient. (½ pt)
2nd Hypothesis: The number of Ach receptors is not sufficient. (½ pt)

e- It is an autoimmune disease. (½ pt)

f- The second hypothesis is valid because the researches have revealed the presence of antibodies, which block or destroy the Ach receptors. (½ pt)

g - At birth, some maternal antibodies are present in the infant's blood which disappears at the age of some months. These maternal antibodies are at the origin of myasthenia in the infant and the disease disappears with the disappearance of these antibodies. (½ pt)

Chromosome X
migrates towards one pole

Chromosome Y
migrates towards the
other pole
of the spindle

The two chromosomes 21
migrate towards the
same pole

Spermatocyte II

Spermatocyte II