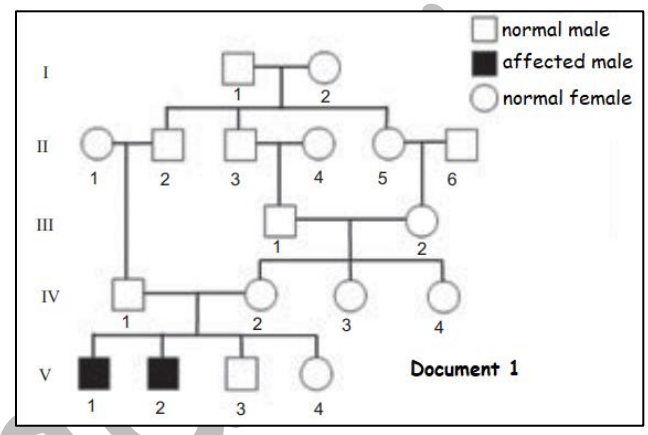
Leb.Bio.Academy
Grade 12 LS
Teacher: Mrs. Soha Al Adawi

Biology Sample exam
Academic year: 2021- 2021
Duration: 150 minutes

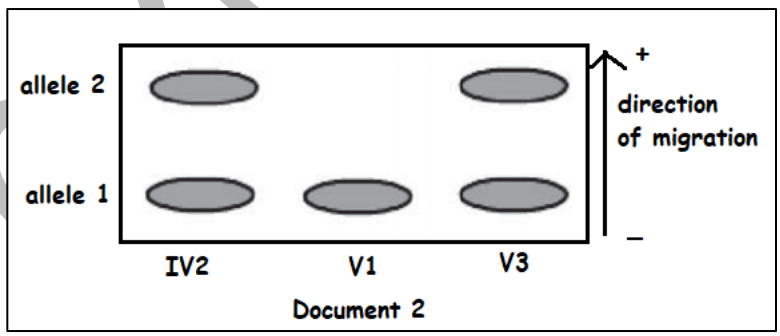
Exercise 1 **Inheritance of congenital myopathies**

Congenital myopathies can be inherited in a variety of ways; X-linked, autosomal dominant and autosomal recessive. Document 1 represents the pedigree of a family in Tunisia where some members are affected by one type of myopathy. In order to determine the localization of the gene responsible for this trait, we formulate two hypotheses.



Hypothesis 1: This type of myopathy is localized on the non-homologous segment of X.
Hypothesis 2: This type of myopathy is autosomal.

1- Specify the dominant and recessive alleles.
2- Show that, by referring to document 1 only, hypothesis 1 could be validated.



Document 2 shows the results of electrophoresis of the DNA corresponding to the gene responsible for this this form of myopathy of some family members represented in document 1. The electrophoresis shows both alleles, the normal allele and the allele responsible for the disease.

3- Based on document 2, identify the allele responsible for the disease.
4- Knowing that all boys of this family have a normal karyotype with 46 chromosomes, and based on document 2, determine which hypothesis is validated.
5- By referring to document 1 only, indicate the genotype (s) of IV1, V1 and V3. Justify.
6- Show that, electrophoresis is more specific in determining genotypes than pedigree.

To prepare the electrophoregram shown in document 2, we use a restriction enzyme X. The restriction enzymes cut in specific sites in DNA after recognizing a sequence of DNA called recognition site.

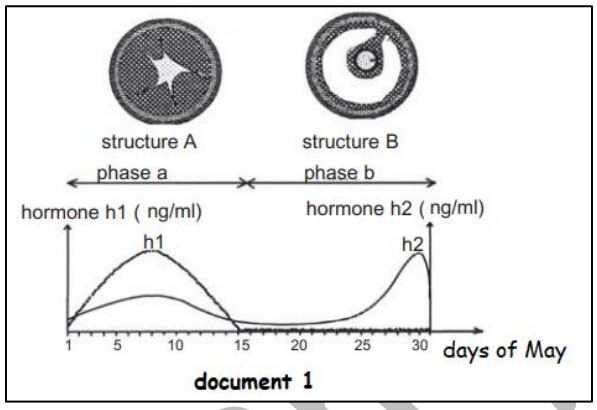
7- By referring to document 2, verify whether the allele responsible for the disease results from the disappearance or appearance of a recognition site of the restriction enzyme X in the normal allele.

The couple IV1 and IV2 expects a new child.

8- Determine the risk for this fetus to be affected.

Exercise 2: **Regulation of the sexual cycle.**

Document 1 shows the schema of 2 ovarian structures that are observed in the ovaries of an adult woman during a period of time between May 1st and May 31st. During the same period of time, we measure the amount of ovarian hormones in blood. The results are represented in the same document.



document 1

1- Name structures A and B.

Phases a and b represent phases during the female ovarian cycle.

2- Specify phase a and phase b.
3- Identify the hormones h1 and h2.

In order to explain the relation between the different organs responsible for the sexual cycle, we perform the following experiments.

Experiment 1: in a female monkey, the ablation of the pituitary gland interrupts the ovarian and uterine cycles.

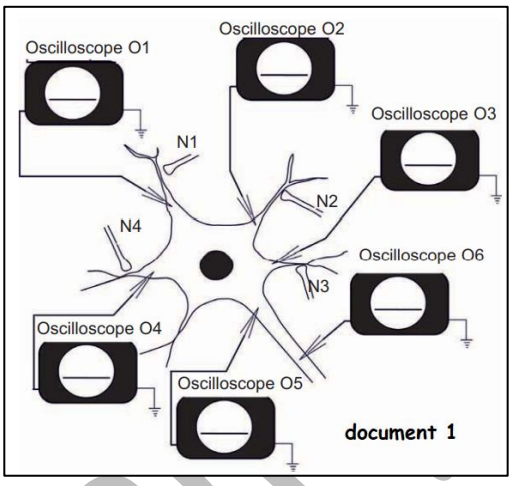
Experiment 2: the regular injection of FSH (a pituitary hormone) to this female (previously subjected to the ablation of pituitary gland) provokes the development of the follicles. But ovulation doesn't take place.

Experiment 3: in another female monkey, the bilateral ovariectomy (ablation of both ovaries) causes a regression of the uterus and an increase in the amount of LH in blood.

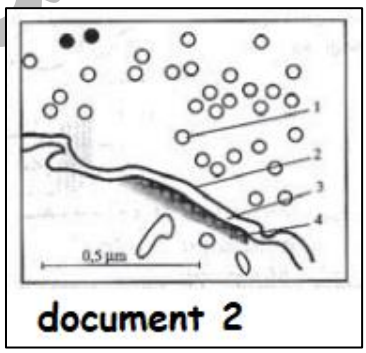
Experiment 4: lesions at the level of specific zones of the hypothalamus (secreting GnRH) of a female monkey shows an important fall in the amount of FSH, absence of ovulation and provoke the interruption of ovarian and uterine cycles.

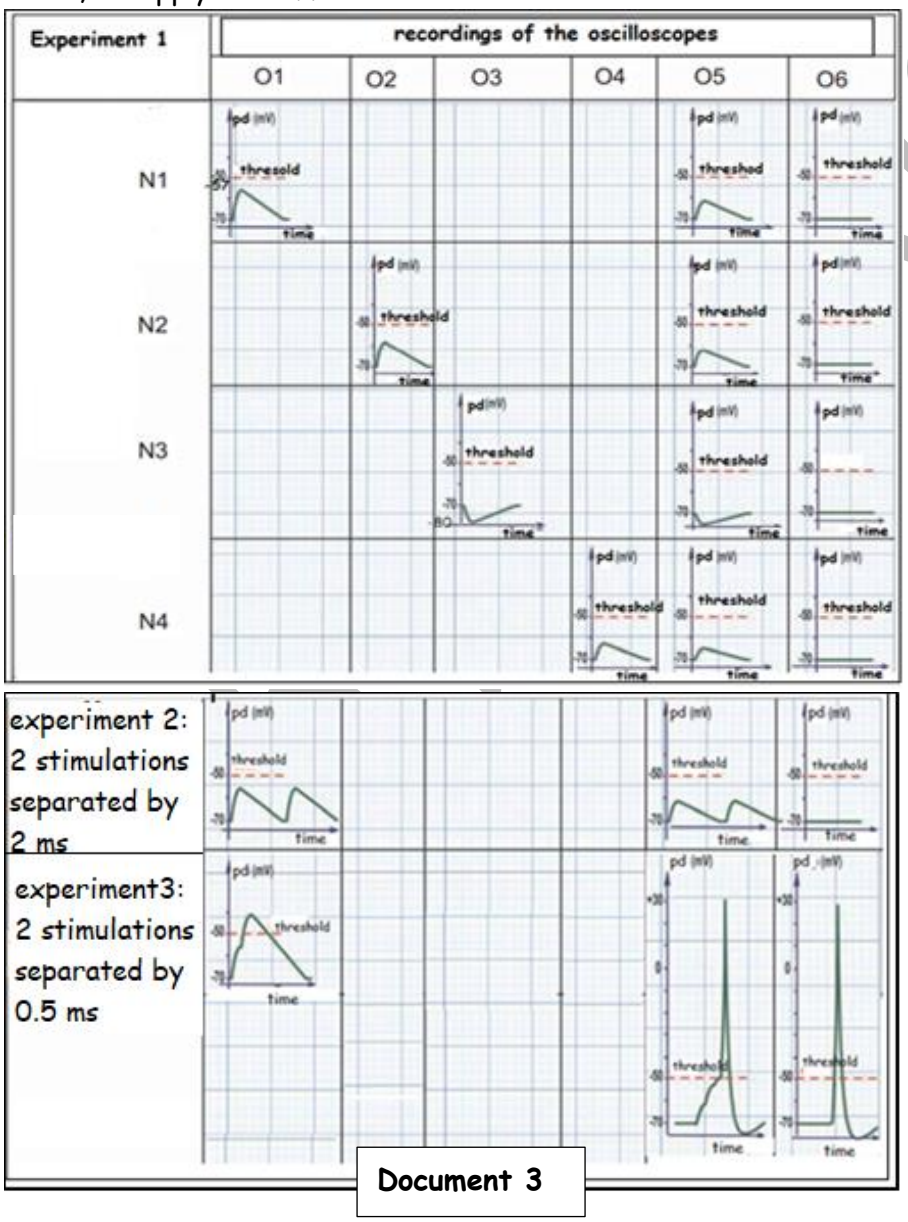
4- What can you draw out from experiment 1?
5- Indicate the roles of FSH and LH, as well as one role of estrogen and progesterone.
6- Interpret the results of experiment 2 and experiment 3.
7- Explain the results of experiment 4.
8- Establish, by referring to all what precedes, a functional diagram showing the relations existing between the different organs involved in the regulation of the sexual cycle.

Exercise 3: **A characteristic of the motor neuron**



At the level of the nervous centers, a motor neuron establishes several thousands of synapses with others neurons. It can receive simultaneously or successively a large number of afferent messages. In order to determine a characteristic of the motor neuron, we perform many experiments. Document 1 shows the experimental setup. Document 2 shows the details of the synapse between N1 and the motor neuron and document 3 shows the experimental conditions as well as the results obtained. In experiment 1 of doc.3, we apply one effective stimulation successively on the neurons N1, N2, N3 and N4. In experiments 2 and 3, we apply two effective stimulations on neuron 1.





Document 3

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1- Label document 2 (1 to 4).
2- Specify the type of synapse between N1 and the postsynaptic motor neuron and that between N3 and the motor neuron.
3- Interpret the results recorded in the oscilloscope 1 of experiments 2 and 3 in document 3.
4- Explain the presence of AP in experiment 3.

Dopamine is a neurotransmitter found in one of the above synapses. Dopamine plays a part in controlling the movements a person makes, as well as their emotional responses. A drug X, agonistic to dopamine, is used to increase the effect of dopamine. In order to study the mode of action of this drug X, we perform the following experiments then measure (estimate) the concentration of dopamine in the synaptic cleft.

- lot 1 of rats in injected by the drug X
- Lot 2 of rats in kept as a control (no injection of drug X).

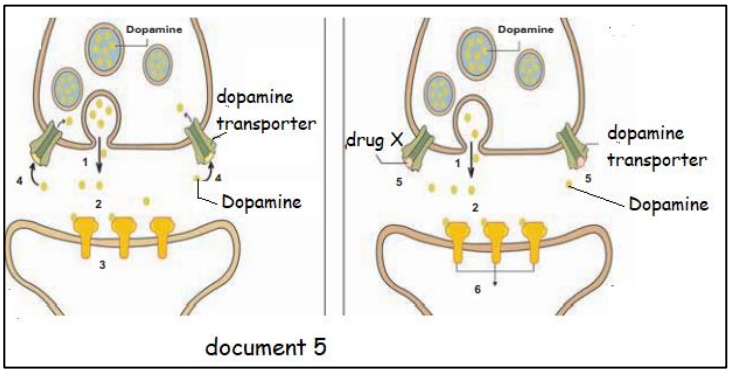
The results are shown in document 4.

Time in minutes		0	20	40	60	80	100	120	140
Concentration of dopamine in %	Lot 1	100	175	225	200	150	150	100	90
	Lot 2	100	105	110	110	110	105	100	100

Document 4

5- Draw in the same graph, the concentration of dopamine as a function of time in both lots 1 and 2.
6- Formulate two hypotheses explaining the mode of action of drug X at the level of the dopaminergic synapse (synapse with dopamine).
7- Show that the results of document 4 cannot validate any of the suggested hypotheses.

Document 5 shows the mode of action of drug X. The dopamine transporter is a transmembrane protein that is responsible for the reuptake of dopamine from the synaptic cleft and for the termination of the dopaminergic transmission.

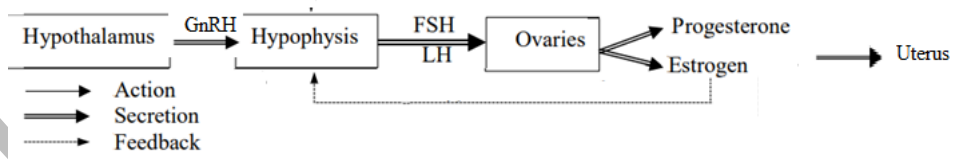


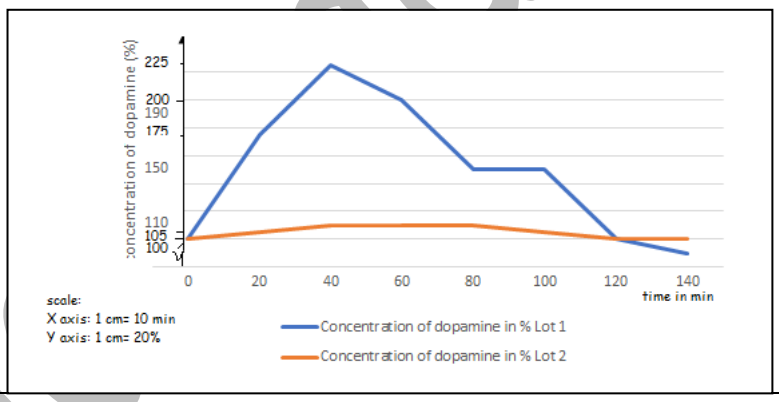
document 5

8- List the steps of the synaptic transmission.
9- Based on document 5, verify if one of your hypotheses is validated.
10- Name drug X.

Answer key

Part	Exercise 1 (6.5 pts) Inheritance of congenital myopathies	mark
1	The normal allele is the dominant allele and the allele responsible for the disease is recessive. The parents IV1 and IV2 are phenotypically normal. They have affected children V1 and V2 who inherited the allele responsible for the disease from at least one parent. Then, the allele responsible for the disease is masked in the parents and therefore recessive. N: normal m: myopathy	½
2	Only males are affected then the disease is gonosomal. If the gene is localized on the non-homologous segment of Y, V1 is affected and should have the genotype XYm, he should have had inherited Yn from his father IV1 who should have had the genotype XYn and should have been affected. But he is normal so this is not the case. Therefore, the disease is X-linked.	½
3	V1 is affected (doc. 1) and shows only a band at the level of the allele 1 (doc.2). therefore, allele 1 is the allele responsible for the disease.	½
4	V3, who is a male, is normal (doc.1). If the gene is localized on the non-homologous segment of X, this boy should show one band (one allele) only as males have only one X chromosome. Since V3 possesses two alleles, the normal allele and the allele responsible for the disease, therefore the gene is autosomal. Hence, hypothesis 2, This type of myopathy is autosomal, is validated.	1
5	IV1: Nm since he is normal and have affected children V1 and V2 who inherited one allele from each parent. V1: mm since m is recessive and can be expressed only in homozygous state. V3: NN or Nm. N is dominant, it is expressed in homozygous and heterozygous states.	1 ½
6	Doc. 3 shows that V3 have two alleles, allele 1 (the allele responsible for the disease) and allele 2 (normal allele). Then the genotype of V3 is certainly Nm. This real genotype couldn't be identified using the pedigree. Thus, electrophoresis is more specific in determining genotype than the pedigree.	½
7	The allele 1, which is responsible for myopathy, is larger in size than allele 2 responsible for the normal allele since it is nearer to the negative pole and didn't migrate far to the positive pole such the allele 2. Therefore, allele 1 results from the disappearance of recognition site.	1
8	IV1 and IV2 are heterozygous since they have affected children V1 and V2. Each parent gives two types of gametes: ½ N and ½ m. Then the risk to be affected is: ½ m × ½ m = ¼	1

Part	Exercise 2 (6.5 pts) Regulation of the sexual cycle	mark
1	Structure A: corpus luteum/ yellow body Structure B: Cavitary or graafian follicle	½
2	Phase a is the luteal phase since the structure found in this phase is the yellow body. Phase b is the follicular phase since the structure found in this phase in the follicles.	½
3	Since h1 is absent during the follicular phase and shows only one peak during the luteal phase then h1 is the progesterone. Since h2 is present in both phases, the follicular and luteal phases, and shows two peaks, where the peak in the follicular phase is greater than that in the luteal phase. Therefore, h2 is estrogen	1
4	The pituitary gland controls the ovarian and the uterine cycles.	½
5	FSH: stimulates the growth and maturation of the follicles. LH: Stimulates the development of the luteal body and its peak triggers ovulation. Estrogen: stimulates the proliferation of the uterine and vaginal mucosa Progesterone: stimulation of gland secretions in the uterine mucosa and the cervix.	1
6	In experiment 2, Ovulation does not occur; however, the follicles develop after injection of FSH to the female monkey subjected to ovariectomy. This indicates that, FSH stimulates the development of the follicles but has no effect on ovulation. (or does not trigger ovulation). While, in experiment 3, the uterus regresses and the amount of LH in blood increases after the bilateral ovariectomy. This means that ovaries control the development of the uterus and inhibit the secretion of LH.	1
7	The lesion at the level of the hypothalamus will interrupt the secretion of GnRH. This latter will not stimulate anymore the pituitary gland to secrete FSH and LH and this is the reason their amount decreases. In the absence of FSH, the follicles will not develop in the ovaries and since developed follicles are responsible for the secretion of high amounts of estrogen, then this amount will remain low. This leads to: - First, the endometrium will not develop and the uterine cycle is interrupted. - Second, the absence of ovulation due to the absence of positive feedback of estrogen on LH, causing the absence of LH peak. Moreover, in the absence of LH, the yellow body will not develop, no progesterone will be secreted and this explains also the interruption of the uterine cycle.	1
8	 a functional diagram showing the relations existing between the different organs involved in the regulation of the sexual cycle.	1

Part	Exercise 3 (7 pts) a characteristic of the motor neuron	mark
1	1: vesicles 2: presynaptic membrane 3: synaptic cleft 4: postsynaptic membrane	1
2	The synapse between N1 and the motor neuron is excitatory since a hypopolarization of amplitude 13 mv is recorded in the motor neuron after an effective stimulation of N1. The synapse between N3 and the motor neuron is inhibitory since a hyperpolarization of amplitude 10 mv is recorded in the motor neuron after an effective stimulation of N3.	1
3	No AP is recorded (only 2 EPSP with amplitude 10 mv) in the motor neuron when two effective stimulations separated by 2 ms are applied on N1. However, an AP with amplitude 100 mv is recorded in motor neuron after applying 2 effective stimulations on N1 separated by 0.1 ms. This shows that the summation of the two hypopolarizations is made only when they are close in time.	½
4	The two stimulations separated by 0.5ms leads to two successive hypopolarizations which due to temporal summation made by the motor neuron exceeds the threshold of depolarization -50 mv leading to the appearance of AP in the motor neuron.	½
5	the variation of the concentration of dopamine in both lots 1 and 2 as a function of time. 	1
6	Hypothesis 1: The drug X increases the exocytosis of dopamine Hypothesis 2: the drug X blocks the recapture of dopamine	1
7	In the absence of drug X, the concentration of dopamine is almost constant around 100% during 140 minutes. However, 40 minutes after the injection of drug X, this concentration increases abruptly to 225 % (2 times more). Then decreases to reach 90% at 140 minutes. This shows that this drug increases the concentration of dopamine in the synaptic cleft and this can result either from blocking the recapture of the dopamine or the increase in the exocytosis of dopamine.	½
8	• the AP arrives to dopaminergic neuron, • Calcium channels open, calcium ions enter the presynaptic neuron. • There is exocytosis of dopamine in the synaptic cleft. • dopamine binds to the postsynaptic receptors on postsynaptic neuron generating a PSP • then the dopamine is recaptured by the presynaptic neuron.	1

9	Drug X blocks the dopamine transporters that are responsible for the reuptake of dopamine from the synaptic cleft. Therefore, hypothesis 2, the drug X blocks the recapture of dopamine, is validated.	
10	Cocaine	$\frac{1}{2}$