Exercise 1 (6 points)

Huntington Disease

Huntington disease is a rare neurodegenerative disease of the central nervous system. It is characterized by uncoordinated and involuntary movements of great amplitude and by psychological problems. It is due to a mutation at the level of the gene coding for a protein called huntingtin which is essential for the survival of the neurons.

Document 1

1- Pick out from the text:

1.1- the origin of Huntington disease.

1.2- the symptoms of this disease.

A study is performed on individuals carrying the mutated allele responsible for this disease. Document 2 represents the variation of the percentage of individuals showing the symptoms of the disease as a function of their age.

2- Interpret the obtained results.

Document 3 shows the genealogical tree of a family which certain members are affected by the disease.

3- Indicate whether the allele of this disease is recessive or dominant. Justify the answer.

4- Determine the localization of the gene responsible for this disease.

DNA analysis is performed on certain individuals of this family using the Southern blot method. The used probe permits to distinguish the mutated allele from the normal one of the studied gene. The obtained results are shown in document 4.

- **5-** Specify the band which corresponds to the mutated allele.
- **6-** Determine the genotype and the phenotype of the fetus.

Age (years)	10	30	40	60	70
Percentage of individuals showing the symptoms of the disease (%)	0	30	60	90	100
-					





Document 3

	Individuals						
Bands	5	6	Fetus				
Α							
В							

Document 4

Exercise 2 (6 points) Evolution of the Ovarian Hormones with Age

Female fertility evolves with age; beyond 50 years old, the cyclic menses as well as ovulation disappear. Studies are done to explain the mechanisms at the origin of these modifications.

Study 1: During a period of 28 days, the levels of ovarian hormones are measured in two women: woman (A) whose age is 25 years old and woman (B) whose age is 50 years old, at menopause. The obtained results are presented in document 1.



3- Indicate a role of estradiol and a role of progesterone.

Study 2: The ovarian follicles which are responsible for the secretion of estradiol and progesterone are evaluated throughout the female life span. The obtained results are presented in document 2.

4- Deduce the cause of the variation of the ovarian hormones observed at menopause.

In order to determine the cause of the disappearance of the ovarian follicles, two hypotheses are proposed:

Hypothesis 1: The disappearance is due to the aging of the ovary itself.



Document 3 represents the evolution in the average plasma level of FSH (pituitary hormone) which stimulates the growth and maturation of ovarian follicles as a function of the age of a woman.

5- Determine which of the two hypotheses is valid.

183										
1000	1									
-	4									
1000	0.000									
-										
	/	$\overline{\ }$							Y	ears
		- 10	20	40	50	60	70	80	Y	ears

Age (years)	20-29	34-39	48-54
FSH (in mg.L ⁻¹)	22	34	60

Document 3

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Exercise 3 (4.5 points)

Cocaine

In the framework of studying the mode of action of cocaine at the level of the dopamine synapses, the following experiments are performed.

Experiment 1: Dopamine percentage is measured at the level of the synaptic clefts in two lots of rats: the rats of **lot 1** which are considered as control group, and those of **lot 2** which are injected with cocaine at time t = 0 minute.

Document 1 represents the obtained results.

- **1-** Represent in the same table, the variation of the percentage of dopamine in the two lots as a function of time.
- **2.1** Analyze the obtained results.
- 2.2- What can you conclude?
- **3-** Propose two hypotheses explaining the mode of action of cocaine at the level of this synapse.



Document 1

Experiment 2: In order to determine the validity of the proposed hypotheses, cocaine is injected into the synaptic cleft of a dopamine synapse.

Document 2 shows the aspect of two synapses, synapse 1 not injected with cocaine (control) and synapse 2 injected with cocaine.





N.B.: Dopamine transporters are responsible for the recapture of dopamine by the presynaptic neuron.

- 4- Which of the two proposed hypotheses is valid? Justify the answer.
- **5-** Explain the following statement: "Cocaine consumption induces a state of dependence and a state of tolerance".

Exercise 4 (3.5 points)

Synaptic Transmission

In order to study certain aspects of the synaptic transmission, the following experiments are performed using the experimental setup shown in document 1.

Experiment 1: Fiber F₁ is stimulated by an effective intensity (E₁). The results are recorded by oscilloscopes at the level of the three post-synaptic structures: the cone of implantation of the axon F_2 (O₁), the axon of the interneuron (O_2) and the cone of implantation of axon F_3 (O_3) . The obtained results are shown in document 2.



Document 1

	-
O 2	Action Potential
O ₃	Hyperpolarization
	Document 2

O₁

Recordings

Hypopolarization

followed by an action potential

1- List the steps of the transmission of the nervous message at the level of a synapse.

2- Specify the nature of each of the synapses S₁, S₂ and S₃.

Experiment 2: Two types of neurotransmitters, acetylcholine and GABA, are injected into the synaptic clefts of S_1 and S_3 . The experimental conditions and the results are presented in document 3.

Synapses	Injected Neurotransmitters	Oscilloscopes		
S .	Acetylcholine	0	Hypopolarization	
51	GABA	01	Resting potential	
Q.,	Acetylcholine		Resting potential	
53	GABA	03	Hyperpolarization	

Document 3

3- Indicate the site of action (synapse(s) S_1 and/or S_3) for each of the utilized neurotransmitters. Justify the answer.

الاسم: الرقم: امتحانات الشهادة الثانوية العامة فرع علوم الحياة

مسابقة في مادة علوم الحياة اسس المتصحيح

Q.	Exercise 1 : Huntington Disease Answer key (6 points)	Note
1.1	The origin of Huntington disease is a mutation at the level of the gene coding for a protein called huntingtin.	0.5
1.2	The symptoms of this disease are uncoordinated and involuntary movements of great amplitude and psychological problems.	0.5
2	The percentage of individuals showing the symptoms of the disease increases from 0 % to 100% as the age of these individuals increases from 10 to 70 years. Therefore, the expression of Huntington disease symptoms is enhanced with age.	1.5
3	The mutated allele is dominant over the normal allele because the affected parents (3 and 4) have healthy children (8 and 9). This means that the allele for normal is present in the parents but it is masked by the allele responsible for the disease. (D = allele responsible for Huntington disease; n = normal allele) D> n	0.5
4	 If the allele responsible for the disease is carried on the non -homologous segment of chromosome Y then, it should be transmitted from father to son; however the affected father (3) has a healthy son (8). Therefore, the allele is not carried by the non-homologous segment of chromosome Y. If the allele of the disease is carried on the non-homologous segment of chromosome X, then the affected father (3) should transmit this dominant allele to all his daughters who should be all affected; however, his daughter (9) is healthy, thus the allele is not carried on the non-homologous segment of X chromosome. If the allele is carried on the homologous segment of chromosome X and Y, then boy (8) who is normal should have the genotype Xⁿ Yⁿ, inheriting Xⁿ from his mother(4) and Yⁿ from his father (3). His sister (9) who is normal should have the genotype XⁿYⁿ and should be healthy but it is not the case. Therefore, the allele is not carried by the homologous segment of chromosomes X and Y. Hence the allele responsible for the disease is carried on an autosome. 	1
5	The band at the level of B corresponds to the mutant allele. This is because the DNA analysis of individual 5 who is healthy shows only a thick band at the level of A indicating that band A corresponds to the normal allele. On the other hand, individual 6 who is diseased shows 2 thin bands at the level of A and another at the level of B. Hence, that band at the level of B corresponds to the mutant allele which is responsible for the disease.	1
6	Since the fetus has two thin bands at the levels of A and B corresponding to the normal allele and mutant allele respectively, then the genotype of the fetus is D//n. Since the mutant allele D is dominant over the normal allele n, the phenotype of the fetus is [D].	1

Q.	Exercise 2: Evolution of the C Answer ke	Ovarian Hormones with Age y (6points)	Note			
1.1	In woman A, during a period of 28 days, the exreaches approximately 250 pg/ml around day 1 pg/ml around day 21. On the other hand, Wom fluctuates around 50 pg/ml during the same interval.	stradiol level shows two peaks: the first peak 2, and a second lower peak that reaches 100 an B shows constant variation of estradiol that rerval of time (from day 0 till day 28).	1			
1.2	In woman A, during a period of 28 days, the preaches approximately 15 ng/ml around day 21 constant variation of progesterone that fluctuat time (from day 0 till day 28).	rogesterone level shows only one peak that I. On the other hand, Woman B shows tes around 3ng/ml during the same interval of	1			
2	At menopause, the cyclic variation of ovarian hormones disappear indicating the cease of the ovarian activity.					
3	Estrogen (any of the following roles) - Proliferation of the uterine and vaginal mucosa - Development tube-like glands of the endometrium. - Development of the cervical glands - Growth of blood vessels.	Progesterone (any of the following roles) - Stimulation of gland secretions in the uterine mucosa and the cervix. - Development of spiral arterioles. - Increase in the body temperature. - Inhibition of the uterine contractions.	1			
4	The number of the ovarian follicles which are responsible for the secretion of estradiol and progesterone decreases from 10 a.u. at around 7 years old to reach 0 a.u. beyond 50 years old. Therefore, the constant level of ovarian hormones and the absence of their cyclic variation observed at menopause are due to the depletion of ovarian follicles.					
5	Given that FSH is responsible for the growth a by referring to doc 3, the plasma level of FSH years old), then the cause of the disappearance of the stimulation of the ovary by the anterior first hypothesis (The disappearance of the folliv valid one.	nd maturation of ovarian follicles and since increases to 60 mg/l at menopause (48 -54 of ovarian follicles is not due to the stoppage pituitary hormones (FSH), and therefore the cles is due to the aging of the ovary) is the	1.5			

Q.			l Ar	Exercise 3: C swer key (4.	ocaine 5 points)			Note
	Time (min	utes)	0	40	80	120		
	Percentage	Lot 1	100	100	100	100		
1	dopamine (%)	Lot 2	100	200	150	100		1
	Variation of th rats.	e percent	tage of dopar	nine (a.u.) as	a function of	time (minute	es) in 2 lots of	
2.1	The percentage of dopamine is 100% at time= 0 min in both lots of rats, lot 1,the control group, and lot 2 which are injected with cocaine. The % of dopamine remains constant(100%) during 120 min while it duplicates after 40 min in lot 2 to reach maximum of 200% then it decreases back to the initial level (100%) at 120 minutes.							0.5
2.2	Cocaine ampli	fies the l	evel of dopar	nine in the sy	naptic cleft fo	or a certain p	eriod of time.	0.5
3	Hypothesis1: Cocaine increases the release of dopamine into the synaptic cleft. Hypothesis2: Cocaine prevents or decreases the recapture of dopamine by the presynaptic neuron						1	
4	Hypothesis 2 is valid, because according to document2, dopamine transporters which are responsible for the recapture of dopamine after being released in to the synaptic cleft are blocked in the presence of cocaine and eventually it leads to excess dopamine in the synaptic cleft for a longer duration of time.						0.5	
5	The consumption continue its use The repetitive of the product in order to obtain	ion of co e. This le use of co and loss ain the de	caine leads to eads to a state ocaine will le of its effect. esired effect.	o a sensation of of dependen ad to the adap Consequently This leads the	of pleasure fo ce. tation of the t, the consume e consumer to	llowed by a c body to the re er tends to in a state of to	lesire to epeated doses crease the dose lerance.	1

Q.	Exercise 4: Synaptic transmission Answer key (3.5 points)	Note
1	 The steps of the transmission of the nervous message at the level of a synapse are: Arrival of action potential at the presynaptic terminal buds leads to the opening of calcium voltage gates. The inflow of Ca²⁺ ions into the presynaptic terminal bud causes the fusion of vesicles that contain neurotransmitters with the presynaptic membrane. The release of neurotransmitters by exocytosis into the synaptic cleft. The Binding of neurotransmitters to postsynaptic receptors allow the opening of chemical-dependent channels that modifies the membrane potential thus creating PSP at the level of the postsynaptic membrane. Later, the neurotransmitters are rapidly destroyed by a specific enzyme or recaptured by the presynaptic neuron. 	1
2	S_1 and S_2 are excitatory synapses because O_1 and O_2 connected to the fibers of the postsynaptic neurons recorded A.P after applying an effective intensity of stimulation, E_1 , to the nerve fiber F_1 of the presynaptic neuron. S_3 is an inhibitory synapse because for the same effective stimulation E_1 , hyperpolarization is recorded by O_3 connected to the post synaptic neuron F_3 .	1.5
3	Acetylcholine acts at the level of the synapse S_1 only because it records a hypolarization at the level of O_1 but not O_3 where it records a resting potential. On the other hand, GABA acts at the level of the synapse S_3 only because it records a hyperpolarization at the level of O_3 but not O_1 where it records a resting potential.	1