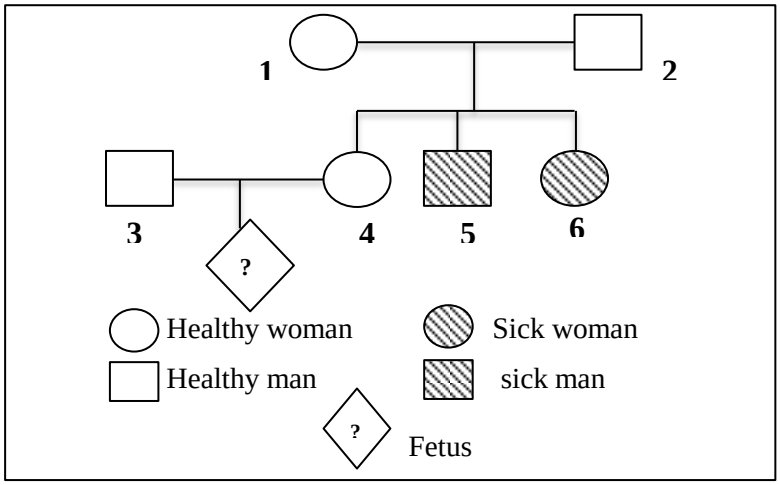
مسابقة في مادة علوم الحياة
المدة: ساعتان ونصف

الاسم:
الرقم:

Answer the following exercises.

Exercise 1 (4 points) Transmission of a Hereditary Character

Fructosemia is a disease caused by deficiency of aldolase B enzyme. The following pedigree in document 1 shows the transmission of this disease in a family.



Document 1

1. Indicate whether the allele responsible for the disease is dominant or recessive. Justify the answer.
2. Determine the chromosomal localization of the gene responsible for this disease.
3. Write the possible genotypes of each of the individuals 3 and 4.

Document 2 represents the results of the electrophoresis performed on the alleles of the studied gene in individuals 3, 4 and the fetus.

Individual / Allele	3	4	Fetus
Normal	▬▬	▬▬	▬▬
Mutant	▬▬		

Document 2

4. Specify, by referring to document 2:
   4.1. the real genotype of each of individuals 3 and 4.
   4.2. the phenotype of the fetus.
5. What advantage does this technique provide to the determination of the genotype of an individual?

Exercise 2 (4 points) **Fructosemia**

Congenital fructosemia is an intolerance to fructose, preventing the absorption of fructose and all sugars containing fructose**.** It is due to a deficiency in aldolase B, an enzyme located in the liver, small intestine and kidneys. Aldolase B enzyme is responsible for the cleavage of fructose -1- phosphate into two molecules: DHAP and glyceraldehyde. Children affected by this disease show a dysfunction of the liver and kidney weakness with abnormal high levels of sugar, amino acids and salts in the urine.

Document 1

1. Pick out from document 1:
- **1.1.** the cause of fructosemia.
- **1.2.** the consequences of this disease.

Document 2 represents the partial sequence of the nucleotide of DNA in the normal and the mutant alleles of the gene determining the synthesis of the enzyme aldolase B.

Allele	Nucleotide sequence of the **transcribed** strand of DNA
Normal	1↓ ↓24 TTA CCT GAC CAT GGA TAA CAA CTT
Mutant	1↓ ↓18 TTA CCT GGA TAA CAA CTT

Document 2

2. Compare these two sequences (document 2).

3. Indicate the type of the revealed mutation.

4. Write, referring to documents 2 and 3:
- **4.1.** the mRNA that corresponds to each allele.
- **4.2.** the sequence of amino acids that corresponds to each allele.

5. Explain how the modification of the nucleotide sequence of the allele leads to the appearance of fructosemia.

Nucleotides position 1	Nucleotides position 2 U	Nucleotides position 2 C	Nucleotides position 2 A	Nucleotides position 2 G	Nucleotides position 3
U	UUU, UUC: phenyl-alanine; UUA, UUG: leucine	UCU, UCC, UCA, UCG: serine	UAU, UAC: tyrosine; UAA, UAG: non-sens	UGU, UGC: cysteine; UGA: non-sens; UGG: tryptophane	U C A G
C	CUU, CUC, CUA, CUG: leucine	CCU, CCC, CCA, CCG: proline	CAU, CAC: histidine; CAA, CAG: glutamine	CGU, CGC, CGA, CGG: arginine	U C A G
A	AUU, AUC, AUA: isoleucine; AUG: methionine	ACU, ACC, ACA, ACG: threonine	AAU, AAC: asparagine; AAA, AAG: lysine	AGU, AGC: serine; AGA, AGG: arginine	U C A G
G	GUU, GUC, GUA, GUG: valine	GCU, GCC, GCA, GCG: alanine	GAU, GAC: aspartic acid; GAA, GAG: glutamic acid	GGU, GGC, GGA, GGG: glycine	U C A G

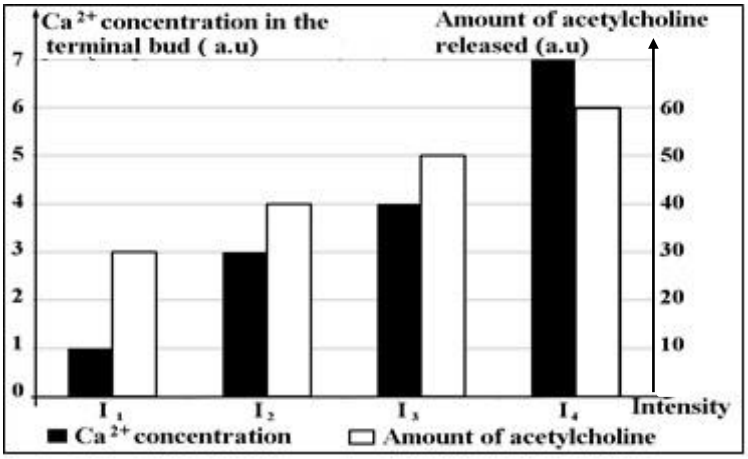
A : Adenine U : Uracile G : Guanine C : Cytosine.

Document 3: Genetic Code

Exercise 3 (6 points) **Synaptic Transmission**

Nerve messages are transmitted along the nerve fibers and across synapses. In the framework of studying the synaptic transmission of the nervous message, the following experiments are performed.

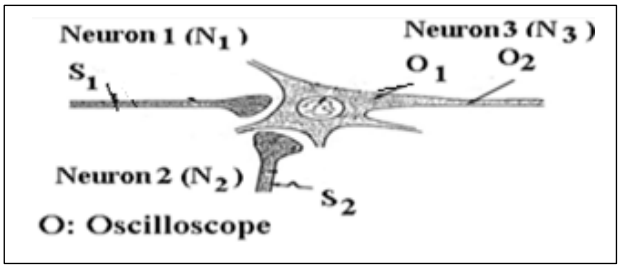
Experiment 1: In a physiological medium and using an experimental set up, four effective stimuli of increasing intensities ($I_1 < I_2 < I_3 < I_4$) are applied on a motor neuron innervating a skeletal muscle.



Document 1

The concentration of Ca^{2+} in the presynaptic terminal bud as well as the amount of acetylcholine released into the synaptic cleft are measured. The obtained results are represented in document 1.

1. List the steps of the transmission of the nervous message at the level of the synapse.
2. Analyze the obtained results.
3. What can you conclude concerning the coding of the nervous message revealed by document 1?



Document 2

Experiment 2: Effective stimulations S_1 and S_2 are applied separately on the presynaptic neurons N_1 and N_2, document 2. The responses are recorded at the level of the postsynaptic neuron N_3.

The results are represented in document 3.

4. Indicate the nature of each of the synapses N_1-N_3 and N_2-N_3. Justify the answer.

Conditions	Recordings	
	O_1	O_2
Stimulation S_1	+30 mV AP -70 mV	+30 mV AP -70 mV
Stimulation S_2	-70 mV -72 mV	-70mV

Document 3

Effective stimulations S_1 and S_2 are simultaneously applied on the presynaptic neurons N_1 and N_2 respectively. The obtained recordings are represented in document 4.

5. Explain the results obtained at the levels of **O_1 and O_2** in document 4.

Conditions	Recordings	
	O_1	O_2
Stimulations $S_1 + S_2$	-68 mV -70 mV	-70mV

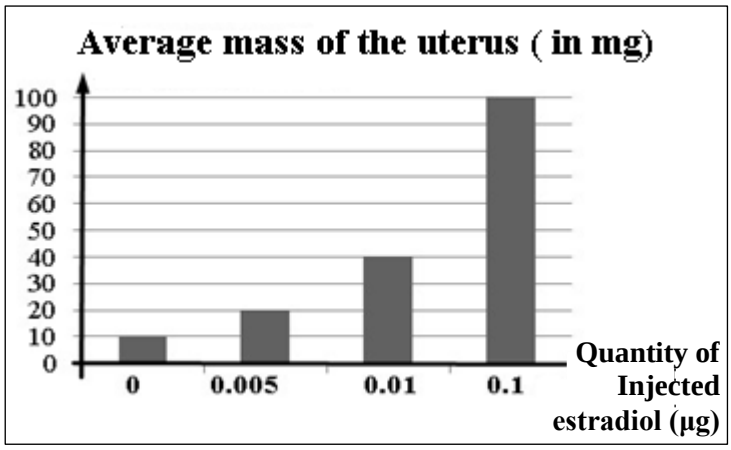
Document 4

Exercise 4 (6 points) **Uterus and Ovarian Hormones**

The uterus, an organ of the female reproductive system, is made up of 3 layers: an external layer (serous external), a muscle layer (myometrium) and an internal layer (endometrium). This endometrium undergoes cyclic development controlled by the ovarian hormones.

In the framework of studying the effect of the ovarian hormones on the uterus, the following experiments are performed.

Experiment 1: Estradiol of increasing quantities is injected to lots of ovariectomized mice at puberty. Document 1 shows the obtained results.



Document 1

1. Name the ovarian structures that secrete estradiol during female sexual cycle.
2. Draw a table that represents the variation of the average mass of the uterus (mg) as a function of the quantity of the injected estradiol (µg).
3.1. Analyze the obtained results.
3.2. What can you conclude?

Experiment 2: Estradiol and progesterone are injected to lots of ovariectomized mice at puberty. The conditions and the results are represented in document 2.

Lots / Injections	Lot 1	Lot 2	Lot 3	Lot 4
Estradiol	-	+	-	+
Progesterone	-	-	+	+

(+): presence (-): absence

Results	No thickening of the endometrium. No uterine lace	Endometrial thickening. No uterine lace	No thickening of the endometrium. No uterine lace	Endometrial thickening with uterine lace

Document 2

4. Specify, referring to document 2:
4.1. the role of estradiol.
4.2. the role of progesterone.
5. Show that the action of progesterone on the endometrium of the uterus necessitates the presence of estradiol.

وزارة التربية والتعليم العالي
المديريّــة العامة للتربية
دائرة الامتحانات الرسمية

امتحانات الشهادة الثانوية العامة
فرع علوم الحياة

دورة العام 2021 العاديّة

مسابقة في مادة علوم الحياة
اسس التصحيح

الاسم:
الرقم:

Q.	Exercice 1: Transmission of a Hereditary Character Answer key	Note
1	The fructosemia allele is recessive (1/4). Because, parents 1 and 2 of healthy phenotype have affected children 5 and 6 (1/4). These children inherited the disease allele from at least one of the parents. This parent has the morbid allele in the masked state. Let N be the symbol of the Normal, dominant allele. Let m be the symbol of the allele responsible for recessive fructosemia.	**1/2**
2	If the gene is localized on the non-homologous segment of Y chromosome, then none of the female should be affected. This is not the case, since female 6 is an affected one. Or father and son would be of the same phenotype because the boy inherits his Y from his father. Or, sons should have the same phenotypes as their fathers (they have inherited the Y chromosome from their father). (1/4). The affected male 5 would have as genotype $X//Y^m$, he has inherited Y^m from his father who would have as genotype $X//Y^m$. Possessing such genotype, the father should be affected, which is not. (1/4). If the gene is localized on the non-homologous segment of X chromosome, then the affected female 6 would have as genotype $X^m//X^m$, she has inherited X^m from his father 2 who would have as genotype $X^m//Y$. Possessing such genotype, the father should be affected, which is not. (1/4). If the gene is localized on the homologous segments of X and Y chromosomes, then the affected female 6 would have as genotype $X^m//X^m$, and similarly the affected male 5 would have as genotype $X^m//Y^m$, female 6 has inherited X^m from her father while male 5 has inherited Y^m from the same father. The father as such should have as genotype $X^m//Y^m$. Possessing such genotype, the father should be affected, which is not. Thus the gene is not gonosomal, therefore autosomal. (1/4).	**1**
3	3 : N//m or N//m 4 : N//m or N//m	**1/2**
4.1	The electrophopregram of individual 3 shows a band that corresponds to the normal allele and another that corresponds to the mutant allele (1/4). Consequently, the genotype of individual 3 is N//m (1/4).. The genotype of individual 4 is N//N (1/4) because he has one band that corresponds to the normal allele (1/4).	**1**
4.2	The fetus has only the normal allele, so he has two copies of the allele N (1/4). He has a normal phenotype (1/4).	**1/2**
5	The electrophoresis permits determining the real genotype of the individual.	**1/2**

Q.	Exercise 2 Fructosemia Answer Key	Note
1.1	Deficiency of aldose B.	**1/2**
1.2	Children affected by this disease show a dysfunction of the liver and kidney weakness with abnormal high levels of sugar, amino acids and salts in the urine.	**1/2**
2	The number of nucleotides in the mutated allele is smaller than in the normal allele 18 <24 (1/4) Nucleotides 7,8,9,10,11 and 12 (or 8, 9, 10, 11, 12 and 13) are absent in the mutated allele (1/4). However, the remaining nucleotides are identical (1/4). Or the first six nucleotides (or the first seven nucleotides) are identical in both sequences (1/4). However, the remaining nucleotides are different (1/4).	**3/4**
3	Mutation by deletion.	**1/4**
4.1	mRNA that corresponds to the normal allele: AAU GGA CUG GUA CCU AUU GUU GAA mRNA that corresponds to the mutant allele: AAU GGA CCU AUU GUU GAA	**1/2**
4.2	The amino acid sequence Asp - Gly - Leu - Val- Pro - Ile- Val –Glu. Diseased mRNA Amino acid sequences: Asp - Gly - Pro - Ile - Val - Glu	**1/2**
5	The mutation by deletion in DNA was transcribed at the level of the mRNA by the absence of codons which results in an absence of the two amino acids Leu and VaL. This new amino acid sequence affects the three-dimensional form of the protein (Enzyme aldolase B) which becomes non-functional. As this enzyme is responsible for the cleavage of fructose 1 phosphate, the change in its function is manifested by fructose intolerance.	**1**

+

Q.	Exercise 3: Synaptic transmission Answer key	Note
1	- Arrival of an action potential to the presynaptic nerve ending. - Opening of calcium channels and entrance of Ca^{2+} ions into the presynaptic membrane. - Liberation of neurotransmitters by exocytosis into the synaptic cleft. - Fixation of neurotransmitters to postsynaptic receptors. - Opening of chemo-dependent channels. - Genesis of PSP in the postsynaptic element. Recapture of neurotransmitters by the presynaptic neuron or its degradation.	1 1/2
2	As the intensity of the stimulation increases from I_1 to I_4, the concentration of Ca^{2+} in the presynaptic element increases from 1 to 7, similarly, the amount of released acetylcholine increases from 30 to 60 a.u.	1
3	The nerve message at the level of a synapse is coded by modulation in Ca^{2+} concentration and the neurotransmitter acetylcholine concentration as a function of the intensity of the stimulation.	1/2
4	The synapse N-N3 is excitatory, because following the application of S_1, an AP is obtained at the level of O_1. The synapse N_2-N_3 is inhibitory, because following the application of S_2, a hyperpolarization is obtained at the level of O_1.	2
5	When S_1 and S_2 are applied simultaneously, an EPSP of amplitude +2mV less than threshold is generated at the level of O_1, but a RP at the level of O_2. In fact, the motor neuron plays an integrative role. It sums up algebraically the IPSP produced at the level of the synapse N2N3 and the EPSP produced at the level of the synapse N1N3. These PSPs are added (spatial summation), producing, thus, an EPSP of amplitude +2mV. Since this amplitude is less than threshold of depolarization, it remains incapable of generating a propagating AP at the level of N3 neuron.	1

Q.	Exercice 4: Uterus and Ovarian Hormones Answer key	Note
1	Estradiol is secreted by: the follicle during the follicular phase and by the corpus luteum during the luteal phase.	1
2	<table><tr><td>The quantity of the injected estradiol (µg).</td><td>0</td><td>0.005</td><td>0.01</td><td>0.1</td></tr><tr><td>The average mass of the uterus (mg)</td><td>10</td><td>20</td><td>40</td><td>100</td></tr></table>The variation of the average mass of the uterus (mg) as a function of the quantity of the injected estradiol (µg).	1 1/2
3.1	The average mass of the uterus increases from 10 to 100 mg when the quantity of the injected estradiol increases from 0 to 0.1 µg.	1
3.2	Thus, estradiol favors the development of the uterus.	1/2
4.1	Estradiol is responsible for the thickening of the endometrium because following its injection (lot 2), thickening of the endometrium is observed.	1/2
4.2	Progesterone is responsible for the formation of the uterine lace because following its injection with estradiol (lot 4), formation of the uterine lace and also thickening of the endometrium are observed.	1/2
5	Since the injection of progesterone alone (lot 3) does not provoke the formation of uterine lace whereas with estradiol (lot 4) provokes the formation of uterine lace, then the action of progesterone on the endometrium necessitates the presence of estradiol.	1