مسابقة في مادة علوم الحياة

المدة: ثلاث ساعات

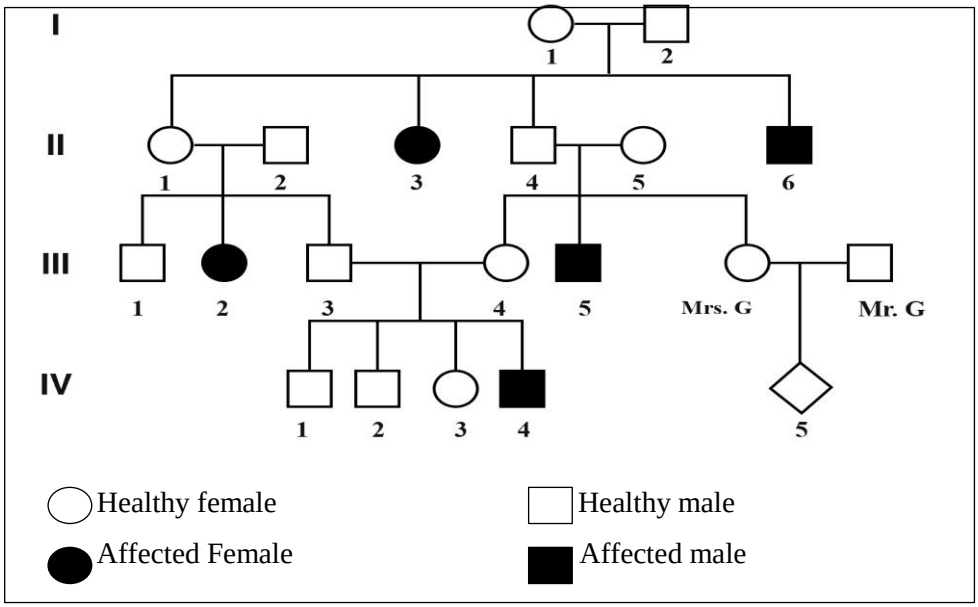
(باللغة الإنكليزية)

الاسم:.....

الرقم:.....

Exercise 1 (5.5 points) **Diagnosis of Galactosemia**

Galactosemia is a genetic disease which results from a deficiency in the enzyme transforming galactose to glucose. On the long term, infants would show retarded growth and later they may have mental retardation.Mr. and Mrs. G are expecting a child. Mrs. G is worried because several members in her family are affected by this disease as shown in the pedigree presented in document 1.



Document 1

-I represents the first generation: the mother is represented by I-1 and the father is represented by I-2.

-II represents the second generation: II-1, II-3, II-4 and II-6 are children of the first generation. II-2 is the partner of II-1 and II-5 is the partner of the II-4.

-III represents the third generation: III-1, III-2, III-3 are the children of II-1 and II-2.

II-4, II-5 and Mrs. G are the children of II-4 and II-5. Mr.G is the partner of Mrs.G.

-IV represents the fourth generation: IV-1, IV-2, IV-3 and IV-4 are the children of III-3 and II-4. IV-5 is the fetus of Mr. and Mrs.G.

1. The allele for the disease is recessive. **Justify**.

2. Choose the best answers:

a- The allele of the disease is not localized on the proper part of Y, because the affected boy II-6 has a healthy father I-2.

b- The allele of the disease is localized on the proper part of X, because the diseased girl II-3 has a healthy father I-2.

c- The allele of the disease is localized on the common part of X and Y, because II-3 and II-6 are diseased but have a healthy father I-2.

d- The allele of the disease is autosomal.

3.1- Match between the individuals below and their corresponding genotype(s)

individual IV-4	•	• N//N
Mrs G	•	• N//m
		• m//m

3.2- Justify your choice.

In the world wide population, the probability of an individual to be heterozygote is 1/100.

4.1 Indicate the risk that the mother is heterozygous.

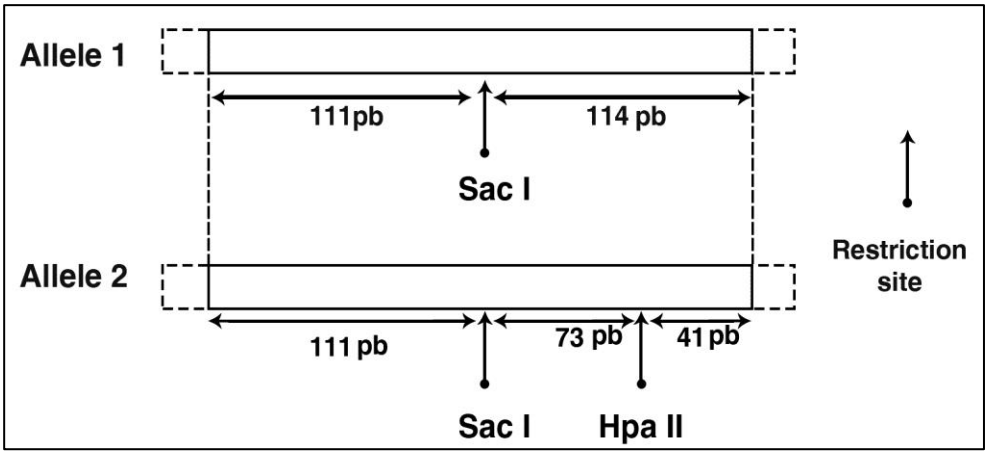
4.2 Indicate the risk that the father is heterozygous**.**

4.3 Indicate the risk that a heterozygous couple has an affected child**.**

4.4 Calculate the risk that the fetus IV-5 will have the disease.

The GALT gene is responsible for galactosemia.

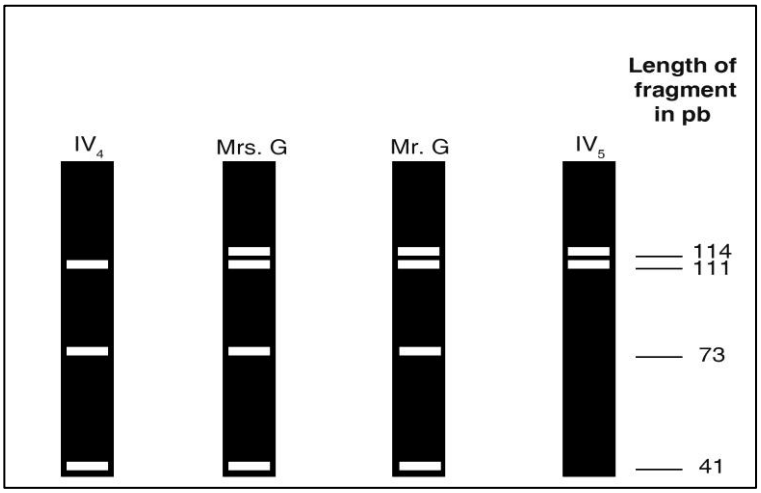
Document 2 shows the cleavage sites of two restriction enzymes, Sac 1 and Hpa II, at the level of a part of two alleles of this gene: Allele 1 and allele 2.



Document 2

1.5. **Indicate**, by referring to document 2, the number and size of restriction fragments obtained by the enzymatic digestion of allele 1 and allele 2.

Document 3 represents the results of electrophoresis obtained after the combined action of enzymes, Sac 1 and Hpa II on allele 1 and allele 2 of GALT gene of certain family members.



Document 3

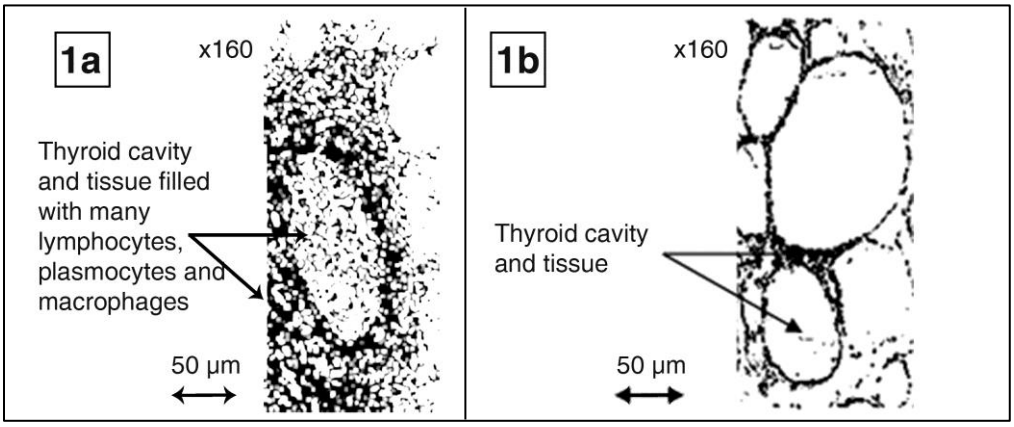
6. Determine the allele which corresponds to the mutant one.

7. The fetus IV5 is diseased? **Justify** the answer of the document 3.

Exercise 2 (5 points) **A Case of Thyroiditis**

Sarah has a swelling of the neck at the level of thyroid gland and suffers from many troubles of metabolic origin. Blood analysis of Sarah shows that the concentration level of the thyroid hormones is noticeably lower than the normal values. The synthesis of these thyroid hormones necessitates the presence of a protein named thyroglobulin.

A biopsy is performed on the thyroid gland of Sarah. Document 1 represents the results of the microscopic observations of the sections of thyroid gland of Sarah (1a) and those of the normal thyroid gland (1b).



Document 1

1. The thyroid gland of Sarah is presenting a probable infection. **Justify** your answer.

The immune and thyroid cells extracted from thyroid gland of Sarah, are cultured in 3 different media. The conditions as well as the results are shown in document 2.

Culture	Cultivated Cells	Results
1	Thyroid cells + B Lymphocytes	Absence of antibodies
2	Thyroid cells + B Lymphocytes + Macrophages	Absence of antibodies
3	Thyroid cells + B Lymphocytes + Macrophages + T_4 Lymphocytes	Presence of a large amount of antibodies

Document 2

2.1- What can you draw out from result of culture 1?

2.2- What can you draw out from result of culture 2?

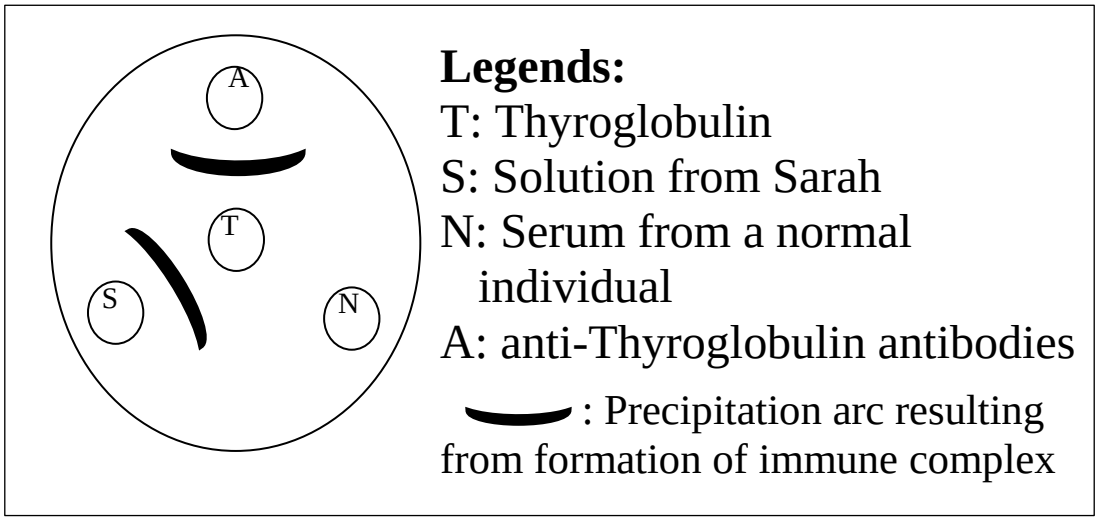
2.3- What can you draw out from result of culture 3?

3. Identify the nature of the specific immune response evidenced in document 2.

4. For explaining the following statement "Macrophages induce specific immune response", **complete** the following text:

The …….. phagocytose the antigen, will be transformed into …….. and will present the antigen to ……. inside ……. that is very close to the site of infection.

Afterwards, immunodiffusion gel test is applied. A solution containing the protein thyroglobulin (T) is deposited in the central well, and three other different solutions are separately deposited in three peripheral wells: A solution of antibodies from Sarah (S), anti-thyroglobulin antibodies (A), and serum from a normal individual (N). The results are shown in document 3.



Document 3

5.1. Explain the formation of the precipitation arc in the presence of solution from Sarah and the thyroglobulin.

5.2 Draw out the name of the disease that Sarah suffers from.

Exercise 3 (4.5 points) **Cause of Muscle Paralysis**

In the framework of studying certain cases of muscle paralysis, researchers work on animals which exhibit complete paralysis of their muscles. In order to determine the origin of this paralysis, the following experiments are performed on a normal animal another paralyzed one. These experiments are performed on the motor neuron N connected to muscle M by synapse F.

Experiment 1:

Effective stimulations are directly applied on muscle M in each of the two animals. Muscular contraction is observed in both cases.

1. What can you draw out from experiment 1?

Experiment 2:

Effective stimulations are applied on motor neuron N innervating muscle M in each animal. The results and the experimental conditions are shown in document 1.

	Results of effective stimulation of motor neuron N	
Normal animal	Nerve Message recorded at level N	Contraction of muscle M
Paralyzed Animal	Nerve Message recorded at level N	No contraction of muscle M

Document 1

2.1 Show that in the normal animal, the transmission of the nerve message across the muscular synapse is normal**.**

2.2 Show that in the paralysed animal, the transmission of the nerve message across the muscular synapse is not normal**.**

A group of researchers formulate the following hypotheses concerning the cause of the synaptic dysfunctioning in the animal affected by muscle paralysis.

H1: Muscle paralysis is due to the blockage of exocytosis of acetylcholine in the synaptic cleft.

H2: Muscle paralysis is due to nonfunctional postsynaptic receptors of acetylcholine.

H3: Muscle paralysis is due to a deficiency in the production of acetylcholine by the presynaptic neuron.

These researchers performed experiments 3, 4, and 5 to verify these hypotheses**.**

Experiment 3:

The analysis of the content of the synaptic vesicles of the neuromuscular synapse in the paralyzed animal reveals the presence of acetylcholine, similar to that in the normal animal.

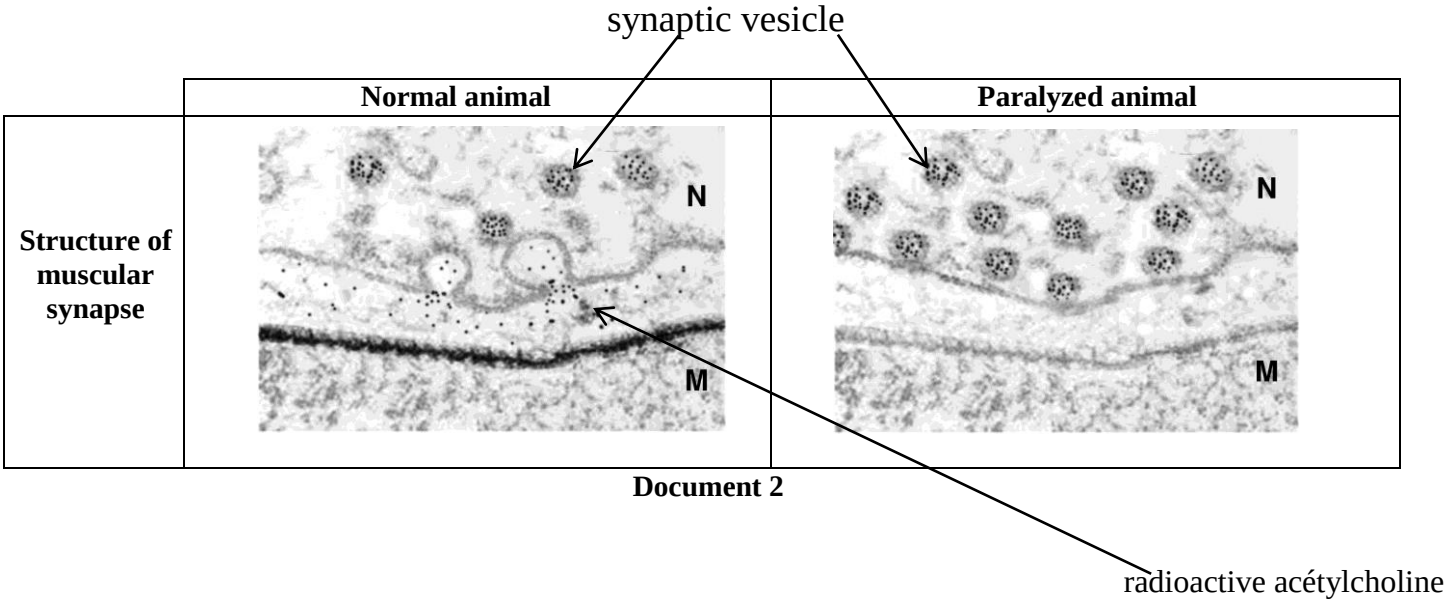
Experiment 4:

Acetylcholine in the neuromuscular synapse of the paralyzed animal is extracted and injected into the synaptic cleft between N and M, in both the paralyzed animal and the normal animal. Muscular contraction of muscle M is obtained in both animals.

3.1 Verify that hypothesis 3 is rejected by the experiment 3.

3.2 Verify that hypothesis 2 is rejected by the experiment 4.

Experiment 5: Radioactive choline, a substance transformed by the neuron into acetylcholine, is injected into neuron N of the normal and paralyzed animals. Then, neuron N in both animals is stimulated. Document 2 shows the electromyographies of the synapse after nervous stimulation. The radioactivity appears in the form of black spots.



Document 2

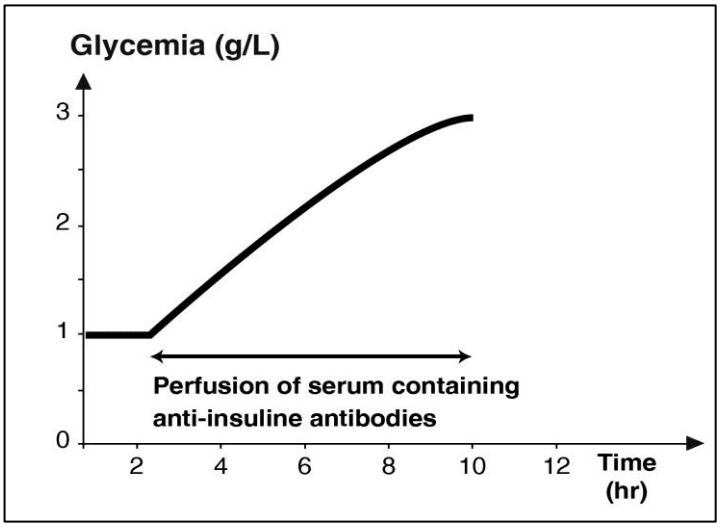
4. Indicate the cause of the paralysis in the animal. Justify your answer.

Exercise 4 (5 points) **Role and Mode of Action of Insulin**

In order to understand the role and mode of action of insulin in an organism, the following experiments are performed.

Experiment 1 :

A rat is perfused (continuously injected) by a serum containing anti-insulin antibodies. These antibodies neutralize insulin, thus preventing it from binding to its receptors on target cells. Afterward, the variation of glycemia is studied. The results are presented in document 1



Document 1

In reference to document 1, answer the following questions:

1.1. How does the glycemia change before the perfusion of serum containing antibodies anti-insulin?

1.2. How does the glycemia change during the perfusion of the serum anti-insulin?

1.3. Conclude the role of insulin evident in the document.

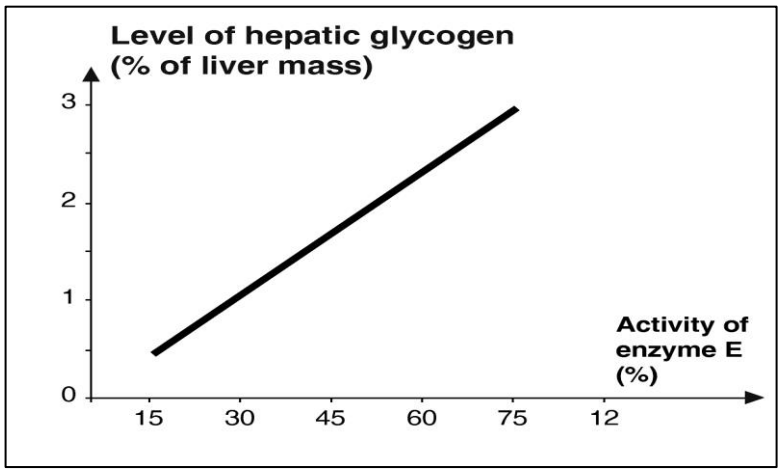
Experiment 2:

1.4. The rate of absorption of glucose by hepatic cells and the hepatic enzyme activity involved in glycogenesis are measured as a function of the concentration of insulin. The results of the experiment are presented in document 2.

Concentration of insulin (µg/L)	5	15	40
Rate of absorption of glucose by the hepatic cells (a.u.)	10	40	90
Activity of hepatic enzyme E (%)	15	60	85

Document 2

2. "Insulin favors the absorption of the glucose in the hepatic cells and increases the activity of the enzyme E". Justify this statement.



Document 3

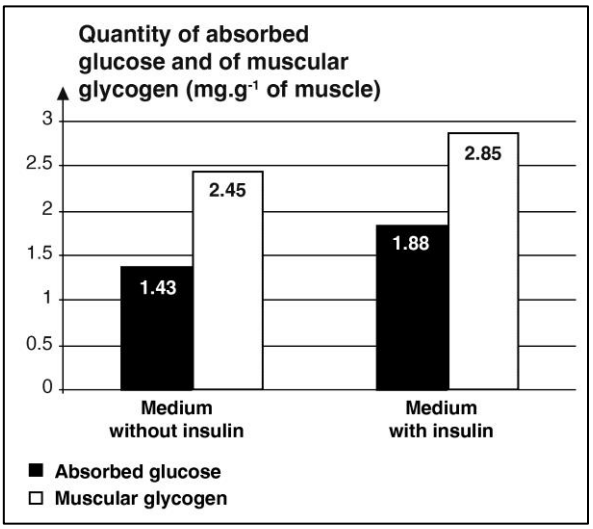
Experiment 3:

The evolution of hepatic reserve in glycogen is studied. The results are shown in document 3.

3. Draw out, from document 3, the role of enzyme E.

Experiment 4:

A muscle is placed in a medium containing glucose with or without insulin for 10 minutes. Then, the quantity of glucose absorbed by the muscle and the quantity of stored glycogen are measured in both media. The results are shown in document 4.



Document 4

4. Fill the following table showing the variation of the quantity of absorbed glucose and that of muscular glycogen with and without insulin.

	Quantity of absorbed glucose by the muscle (in mg/g)	
Medium without insulin		

5-1. compare the quantity of glucose absorbed by the muscle in a medium without insulin to that placed in the presence of insulin.

5-2. Compare the quantity of glycogen in the muscle placed in a medium without insulin to that placed in the presence of insulin.

5-3. Draw out the effects of insulin on the muscle.