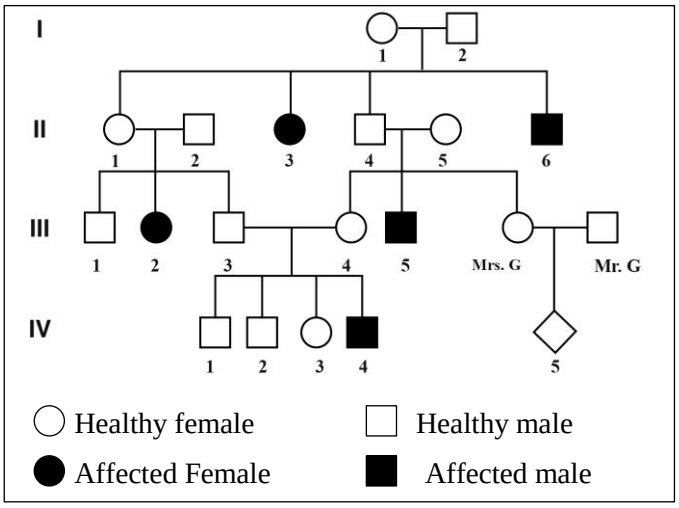
مسابقة في مادة علوم الحياة
المدة: ثلاث ساعات

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. Several days following the consumption of milk or milk products, the following clinical signs appear: vomiting, diarrhea, ….. . 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.



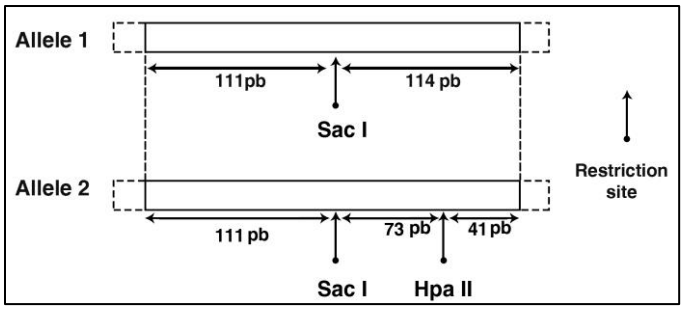
Document 1

1. Indicate if the allele responsible for the disease is dominant or recessive. Justify the answer.
2. Determine the chromosomic location of the gene responsible for this disease.
3. Specify the possible genotype(s) of Mrs. G and individual IV-4.

Worldwide, the probability of individuals to be heterozygous for the gene responsible for this disease is 1/100.

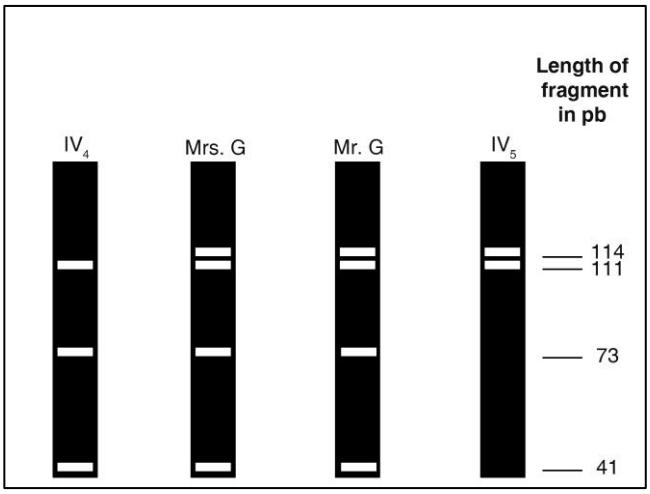
4. Determine the risk for the expected child, IV5, to be diseased.

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 **(**from nucleotide 1367 to nucleotide 1605) of two alleles of this gene: Allele 1 and allele 2.



Document 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.



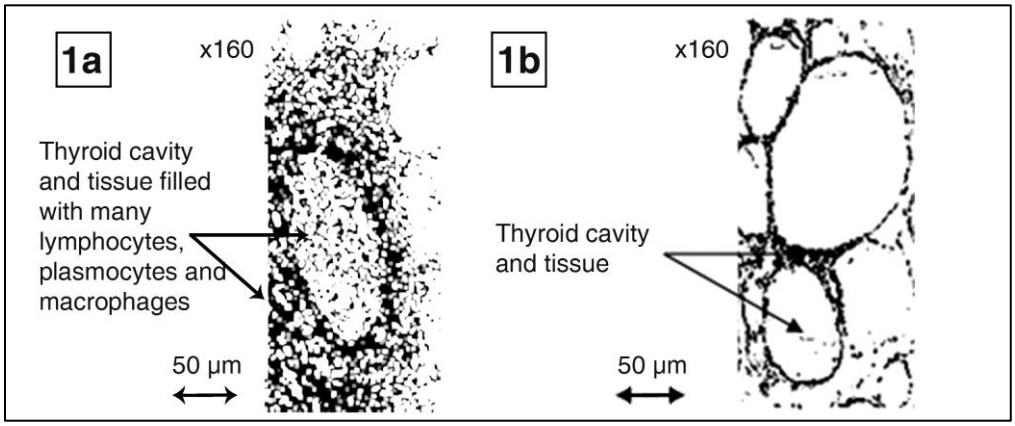
Document 3

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.
6. Determine the allele which corresponds to the mutant one.
7. Verify if the fetus IV5 will be affected by galactosemia.

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. Formulate a hypothesis that can explain the results of biopsy of the thyroid gland of Sarah.

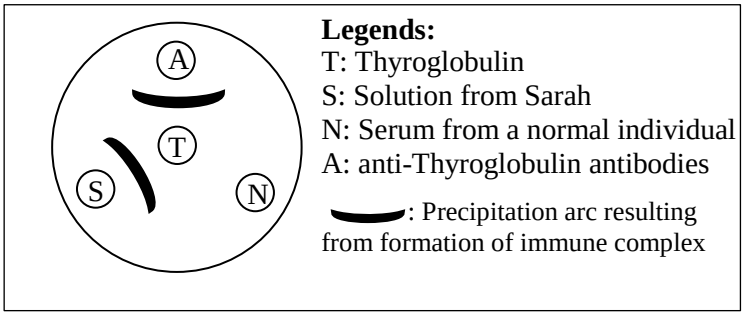
The immune and thyroid cells extracted from the thyroid gland of Sarah, are cultured in 3 different media. The conditions as well as the results are shown in document 2.

2. Interpret the results shown in document 2.
3. Identify the nature of the specific immune response revealed in document 2.
4. Explain the following statement: "Macrophages induce specific immune response".

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

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. Show that Sarah suffers from an auto-immune disease directed against the self.

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

In the framework of studying certain cases of muscle paralysis, researchers carried on experiments 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.

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.

1. Show that the paralysis of this animal is due to dysfunctioning of the synapse.

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

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

Document 1

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.

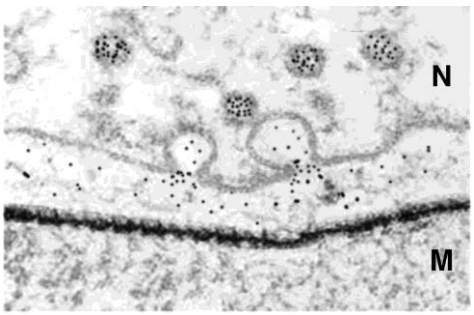
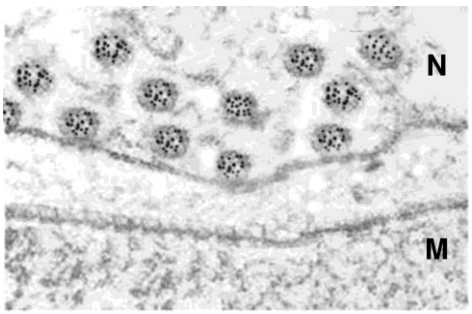
These researchers performed experiments 3, 4, and 5 to verify these hypotheses**.**

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. Contraction of muscle M is observed in both animals.

2. Determine, after studying the results of each of the experiments 3 and 4, the two rejected hypotheses.

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.

	Normal animal	**Paralyzed animal**
Structure of neuro-muscular synapse		

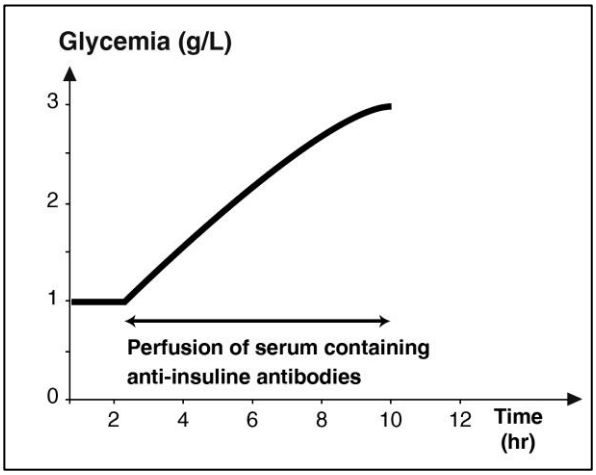
Document 2

3. Specify the cause of muscle paralysis in the animal.

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 neutralise insulin, thus preventing it from binding to its receptors on target cells. Afterwards, the variation of glycemia is studied. The results are presented in document 1



Document 1

1.1. Analyse document 1.
1.2. Conclude the role of insulin evident in the document.

Experiment 2:
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.

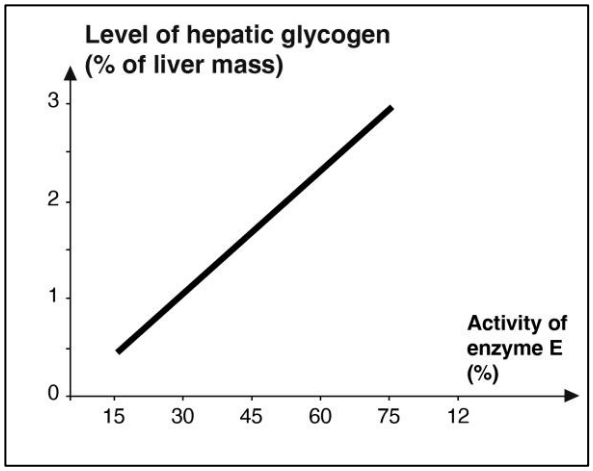
Concentration of insulin (µg/L)	5	10	15	20	40
Rate of absorption of glucose by the hepatic cells (a.u.)	10	20	40	60	90
Activity of hepatic enzyme E (%)	15	45	60	75	85

Document 2

2. Interpret the results represented in document 2.

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

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

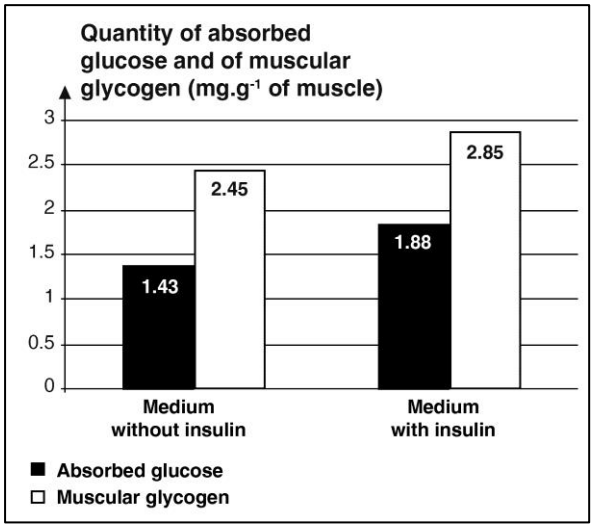


Document 3

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.

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

5. What can be deduced concerning the effect of insulin on the muscle?



Document 4