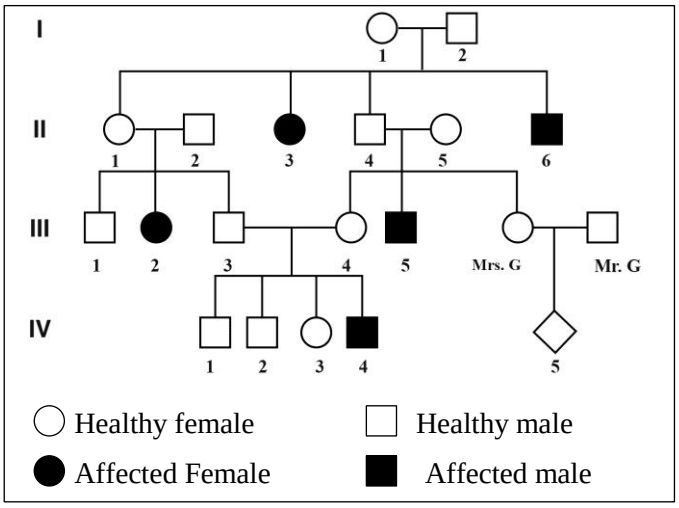
مسابقة في مادة علوم الحياة
المدة: ثلاث ساعات

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.



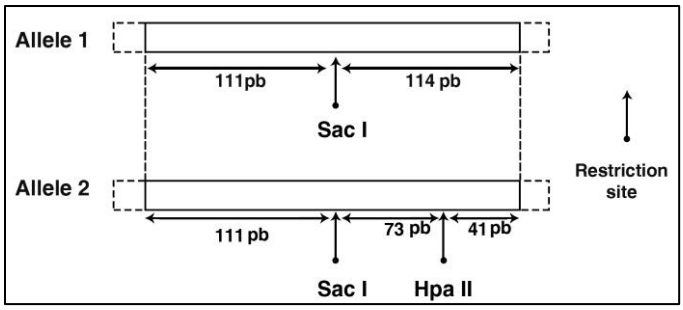
Document 1

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.

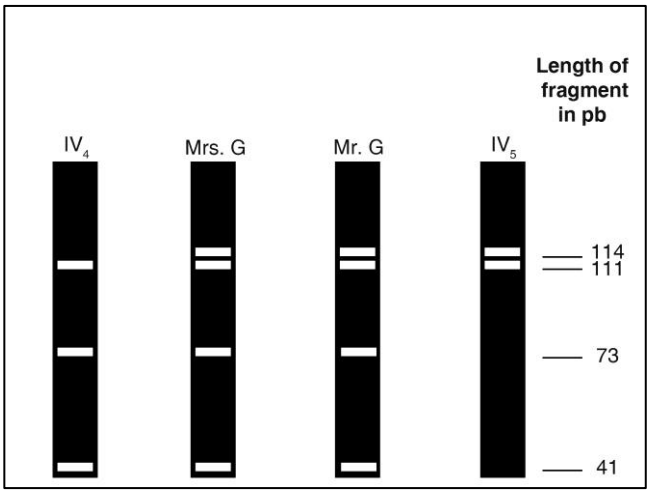
4. Determine the risk for the expected child, IV5, to be diseased.

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.



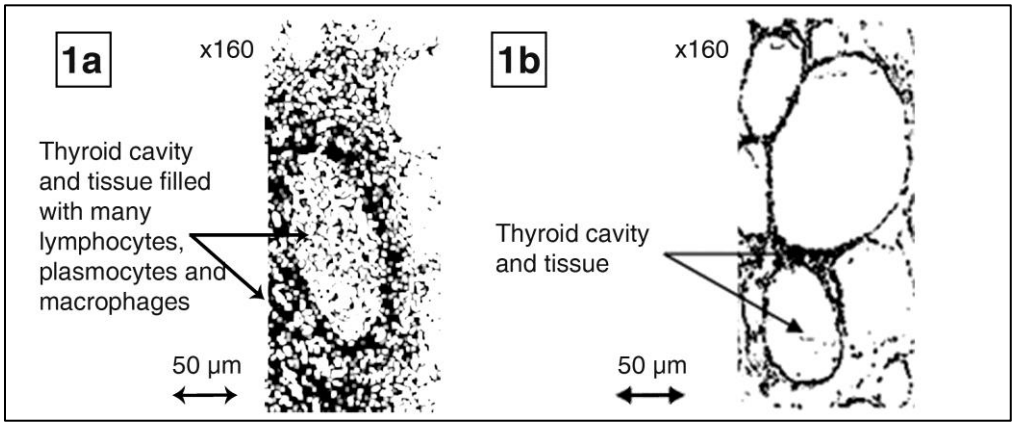
Document 3

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.

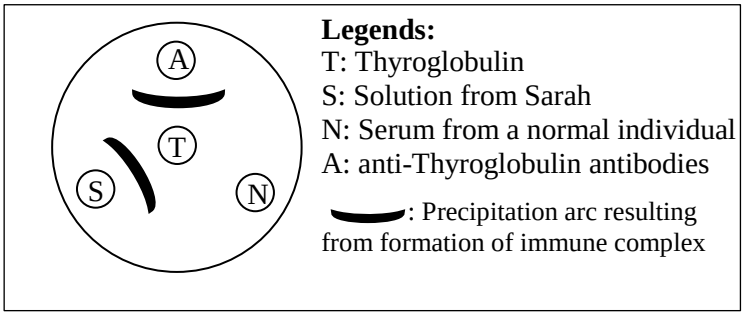
The immune and thyroid cells extracted from the thyroid gland of Sarah, are cultured in 3 different media. The conditions as well as the results are shown in document 2.

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.

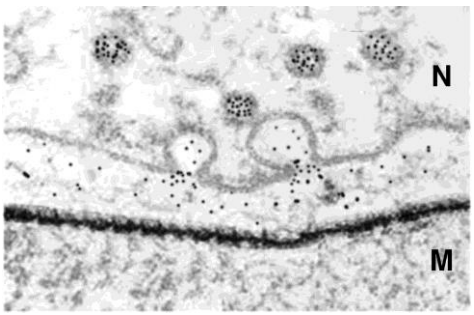
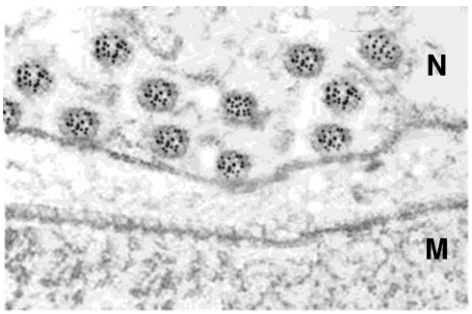
These researchers performed experiments 3, 4, and 5 to verify these hypotheses**.**

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		

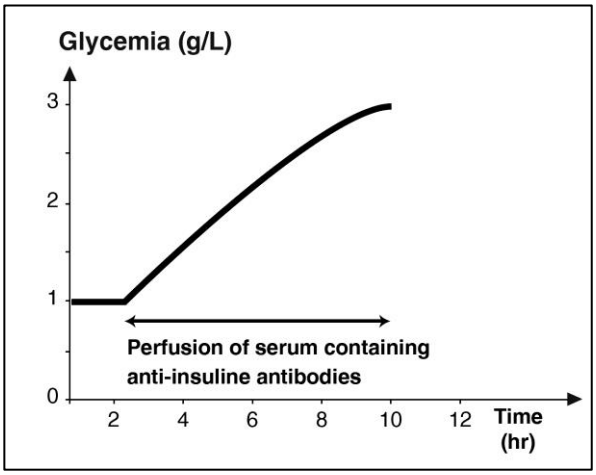
Document 2

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.

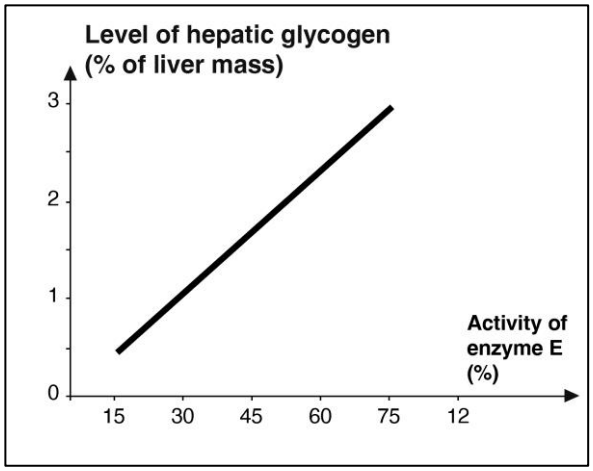
Concentration of insulin (µg/L)	5	10	15	20	40
Rate of absorption of glucose by the hepatic cells (a.u.)	10	20	40	60	90
Activity of hepatic enzyme E (%)	15	45	60	75	85

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.

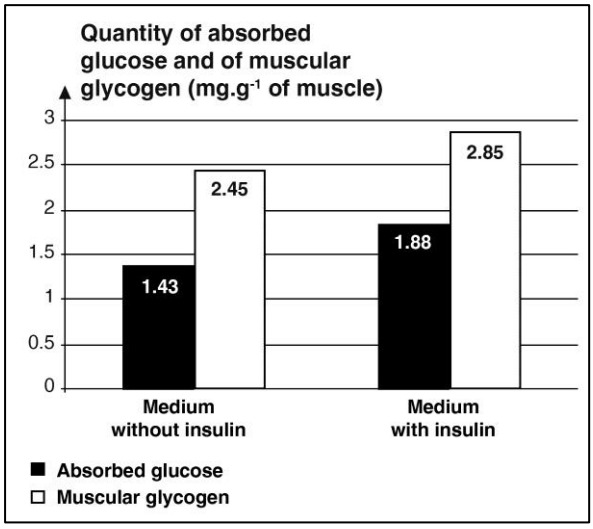


Document 3

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

Q1	Exercise 1 : Diagnosis of Galactosemia Correction	Marks
1	The allele of the disease is recessive. Couple 1 and 2 are normal but have affected children 3 and 6.This shows that the allele of the disease is carried at least by one of the parents who do no not show phenotypically, so the allele is masked, that's why it is recessive (g) with respect to the normal allele N.	**0.5**
2	If the gene is carried by the proper part of Y , First argument: there are no girls affected because	
3		

Exercice 2 (5 points) **Case of Thyroditis**

Q 2	Correction	Marks
1	Hypothesis : Sarah may have an infection in the thyroid gland. Sarah may have an auto-immune disease. Sarah may have a cancer at the level of the thyroid gland	

2	A large amount of antibodies is secreted in culture 3 in the presence of four types of cells : thyroid gland, B lymphocytes , macrophages and T4 lymphocytes. On the contrary, no antibodies are secreted in the absence of T4 lymphocytes (culture 2) and absence of macrophages (culture 1). This means that secretion of antibodies by B lymphocytes nécessitâtes the presence of T4 lymphocytes and macrophages in the presence of an antigen, in this case the infected thyroid cells of sara.	0.5
3	Document 2 reveals the secretion of antibodies, therefore, the nature of specific immune response is humoral.	
4	When a macrophage phagocytoses and digests a cell or protein, resulting peptides are attached to HLA class II molecules and presented on the cell surface. The macrophage migrtaes to te closetst lymph node, where it becomes an antigen presenting cell or APC. The T helper cells that are specific for the peptides presented by this APC remain attached to it. Then they are activated and they proliferate	
5	The anti-thyroglobulin antibodies in well A moves along the gel where it recognizes the thyroglobulin protein, fix to it and form an immune complex which appears as a precipitaion arc. A similar precipitation arc (immune complex) is formed between well S and T which means that serum of sara contains antibodies specific to the protein thyroglobulin where they move along the gel and forms an immune complex . No such arc is revealed between well A and well N which lacks the anti-thyroglobulin antibodies since well N contains serum of a normal individual. Thus. Sara cells serete antithyroglobulin antibodies which attack the thyroglobulin protein in her thryoid gland leading to problems in metabolism and swollen neck. This shows that Sara has auto immune disease.	

Q4.	**Correction**	**Marks**
1.1	At T=0 , the level of glycemia of the rat is 1g/l> This glycemia rests constant at this level till t=2 hr, time of injecting the rat with a continuous amount of anti-insulin antibodies. This perfusion induces an augmentation of glycemia from 1g/l to 3 g/l at t=10 hr.	**0.5**
1.2	Insulin plays a hypoglycemic role.	**0.25**

<table>
<tr><td>2</td><td>With an insulin concentration of 5 Mg/l…., the absorption rate of glucose by the hepatic cells is 10 a.u. and the activity of hepatic enzyme E is 15% .
As insulin concentartion increases, the absorption rate of glucose increases to reach 90 a.u (about 90 % increase) and also the activity of enzyme E increases to reach 85% (about 5 times increase) when insulin concentartion reaches 40 Mg/l .
This shows that insulin acts on the hepatic cells and favors the absorption of glucose by the hepatic cells and activate enzyme E.</td><td>1</td></tr>
<tr><td>3</td><td>The hepatic enzyme E favors the augmentation of hepatic reserves.</td><td>0.5</td></tr>
<tr><td>4</td><td>Table showing the variation of the quantity of glucose absobred by hepatic cells and quantity of muscular glycogen in the absence and absence of insulin.
<table><tr><td></td><td>Glucose stored (mg/g)</td><td>Muscular glycogen (mg/g)</td></tr><tr><td>Medium without insulin</td><td>1.43</td><td>2.45</td></tr><tr><td>Medium with insulin</td><td>1.88</td><td>2.85</td></tr></table></td><td></td></tr>
<tr><td>5</td><td>In a medium without insulin, the quantity of stored glucose is 1.43 mg/g and that of muscular glycogen is 2. 45 mg/g.
However, in the presence of insulin, the quantity of stored glucose increases to 1.88 mg/g and that of muscular glycogen also increases to reach 2.45 mg/g.
We deduce that insulin allows the muscle to absorb glucose and store it in the form of glycogen</td><td>1</td></tr>
</table>

Q	Exercise 3	Marks
1	Experiment 1 shows that both muscle contracts when they receive direct effective stimulations. So Both muscles are functional. Experiment 2 shows a nervous message at the level of motor neuron N when it is stimulated .So motor neuron N is functional. On the contrary, muscle M of the paralyzed animal doesn't show any contraction as a consequence of a nervous message that it receives contrary to the muscle of the normal animal. This shows that the paralysis in the animal is neither due to a disfunctioning of motor neurone N nor due to disfunctioning of muscle M. So it is due to dysfunctioning at the level of neuromuscular synapse.	1.5
2	As vesicles are filled with acetylcholine as in the normal animal, this permit us to reject H3 which proposes a deficiency in acetylcholine. The muscular contraction of the paralysed animal upon injection of acetylcholine in the synaptic shows that these neurotransmitters are fixed on their postsynaptic receptors at the level of the muscle .This evident rejects H2.	2
3	The electromyographies show black traces at the level of synaptic membrane. A- Synaptic vesicles are marked in black. This indicates the presence of acetylcholine. B- The vesicles during exocytosis and the black spots of acetylcholine are observed uniquely in the normal individual. A radioactive marker is observed uniquely on the post Synaptic membrane of the muscle of the control animal. So the cause of the paralysis of the animal is due to the absence of exocytosis, and consequently absence of liberation of acetylcholine. The message is not transmitted and the muscle remains relaxed.	1