

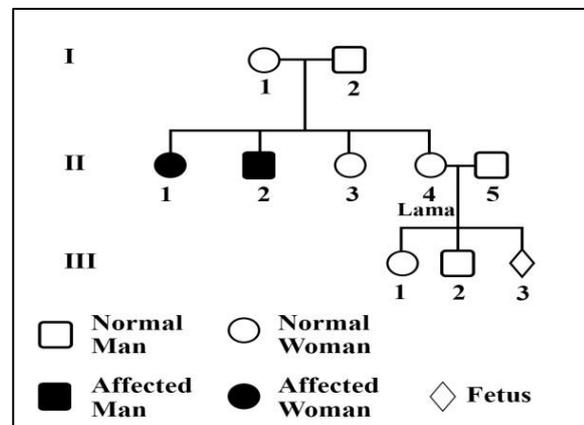
## Answer the following four exercises.

### Exercise 1 (5 points)

### Cystic Fibrosis

Cystic fibrosis is a serious genetic disease. It is characterized by respiratory and digestive disorders. The gene responsible for the disease is carried on chromosome n°7. Document 1 represents the pedigree of a family whose certain members are affected by this disease.

- 1-1. Specify if the allele responsible for this disease is dominant or recessive.
- 1-2. Designate by symbols the corresponding alleles.
2. Indicate the possible genotype(s) of Lama (II<sub>4</sub>). Justify the answer.



Document 1

Lama, whose certain members of her family are affected by this disease, is pregnant. The doctor demanded a specific test, for her and her fetus, that reveals the types and the number of alleles of the studied gene. The obtained results are represented in document 2.

3. Determine the real genotype of Lama, document 2.
4. Is Lama's fetus affected by cystic fibrosis? Justify the answer.

Individuals	Lama II <sub>4</sub>	Fetus
Normal allele	1	0
Affected allele	1	2

Document 2

### Exercise 2 (5 points)

### The Gout

Urea, eliminated by the kidneys in the urine, is a toxic substance. The elevated level of urea in blood might provoke a disease that affects the kidneys and the joints: The Gout.

1. Pick out, from the text, the effect of the high quantity of urea in blood.

In order to determine the origin of urea in the blood, a study is performed on healthy individuals to show the relation between the quantity of consumed proteins and the concentration of urea in plasma. The obtained results are represented in the document below.

<b>Quantity of consumed protein (g/Kg of Body mass)</b>	0.5	1.5	2
<b>Concentration of urea in plasma (g/L)</b>	0.20	0.40	0.45

2. Draw a curve showing the variation in the concentration of urea in the plasma as a function of the quantity of consumed protein.
- 3-1. Analyze the obtained results.
- 3-2. Draw out the origin of urea in plasma.
4. Explain the cause of Gout disease.

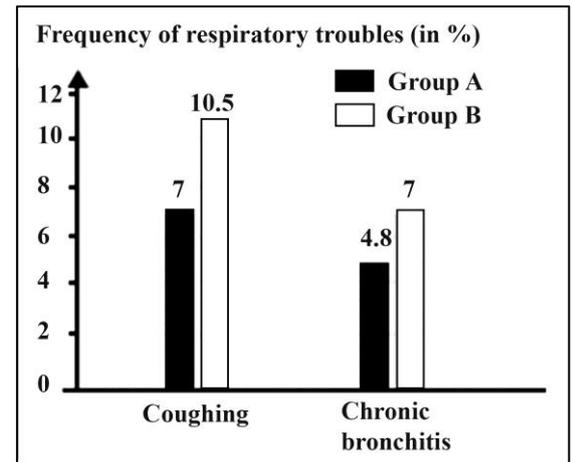
### Exercise 3 (5 points) Passive Smoking

Passive smoking is when someone involuntarily inhales cigarette smoke produced by neighboring smokers. Each year, 3000 to 6000 non-smokers die due to passive smoking.

1. Pick out, from the text, the definition of passive smoking.

The adjacent document represents the frequency of the respiratory troubles in two groups of non-smokers:

- Group A: not exposed to cigarette smoke for long periods of time in their environment.
- Group B: exposed to cigarette smoke in their environment.

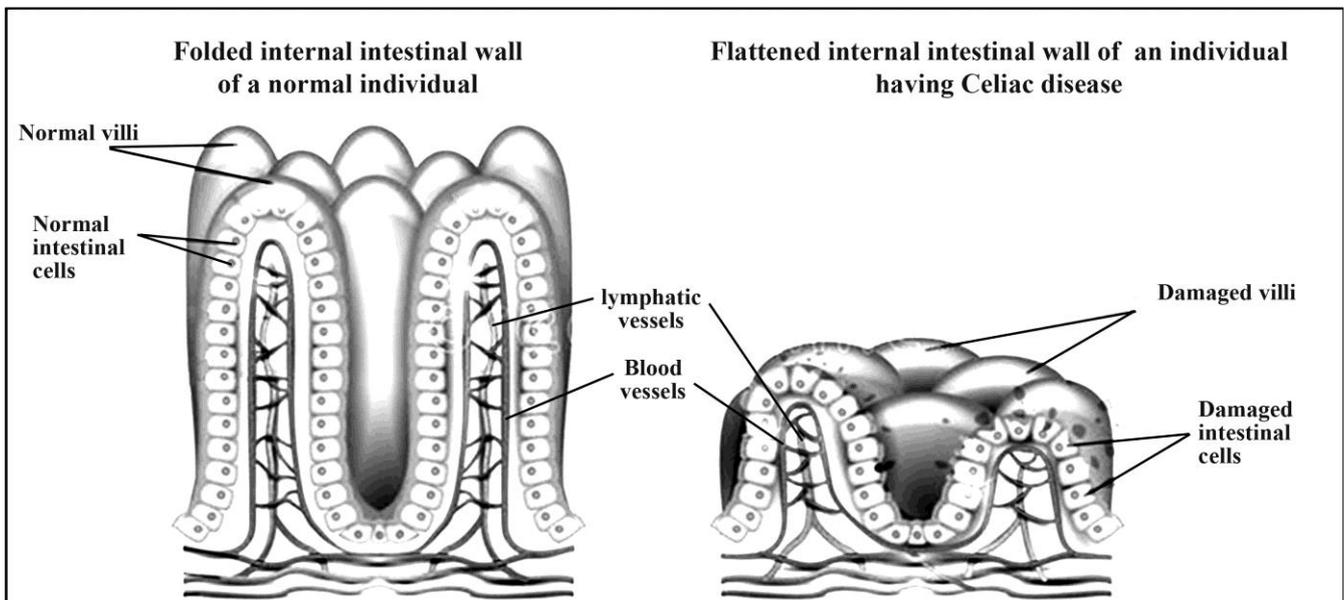


2. Draw a table showing the variation in the frequency of different respiratory troubles in both groups A and B.

3. Justify the following statement: “Inhibition of smoking in closed public areas is a major action that protects non-smokers”.

### Exercise 4 (5 points) Celiac Disease

The intestinal villi are small finger-like projections that line up the inner intestinal walls of the small intestine and increase considerably its surface area. Certain individuals who are affected by celiac disease show symptoms such as fatigue, anemia and diarrhea upon eating food diet rich in gluten. The document below, shows the difference in the inner walls of the small intestine of a normal individual and of an individual affected by celiac disease.



1-1. Indicate the role of the intestinal villi.

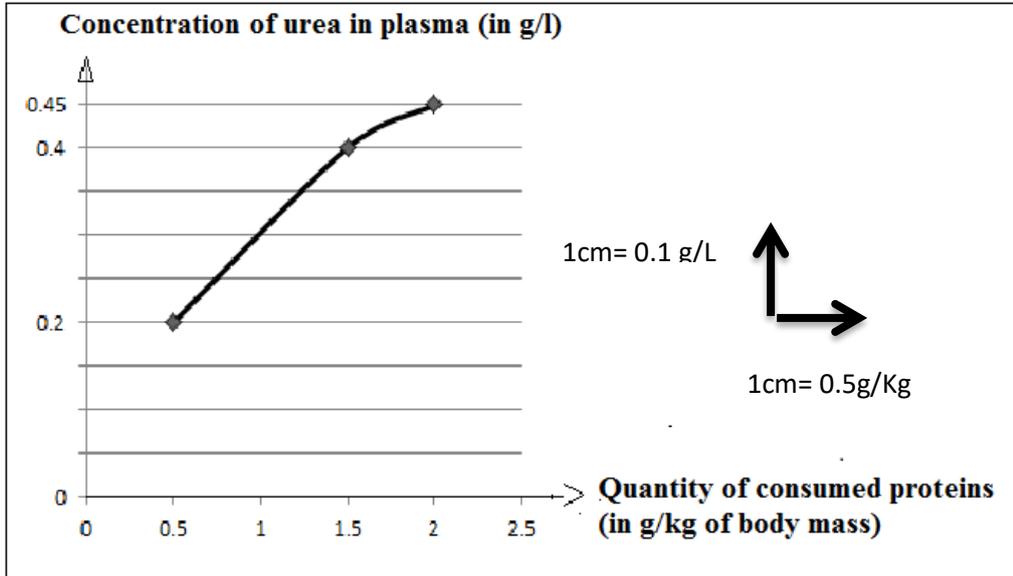
1-2. List the three characteristics of the intestinal wall which favor this role.

2-1. Compare the aspect of the inner wall of the small intestine in these two individuals.

2-2. Draw out the characteristic(s) of the inner wall in the individual affected by this disease.

3. Justify why this disease can slow down the growth of the infant although he consumes a balanced food diet.

Q	Exercise 1 Cystic Fibrosis	Mark
1-1.	The allele responsible for this disease is recessive because the healthy couple (I <sub>1</sub> and I <sub>2</sub> ) gave birth to two affected children II <sub>1</sub> and II <sub>2</sub> who received the allele responsible for the disease from their parents. This indicates that the allele responsible for the disease is present in the parents but is masked by the normal allele, and not expressed phenotypically.	1.5
1-2.	Let "N" be the symbol of the dominant allele that is responsible for the normal phenotype. Let "d" be the symbol of the recessive allele that is responsible for the disease cystic fibrosis.	0.5
2.	The possible genotypes of Lama are N//N or N//d. Since Lama is normal, she possesses the normal allele (N) responsible for the normal phenotype. The parents I <sub>1</sub> and I <sub>2</sub> are heterozygous (they are normal and have affected children), She can receive either one normal allele (N) from each parent or a normal allele (N) from one parent and an allele (d) responsible for the disease from the other parent.	0.5 1
3	Since the results in document 2 show that Lama has two types of alleles: one normal allele and one allele responsible for the disease, therefore her genotype is N//d.	0.75
4	Yes, the fetus is affected by cystic fibrosis, because he possesses the two alleles responsible for the disease.	0.75

Q	Exercise 2 The Gout	Mark
1	The elevated level of urea in blood might provoke a disease that affects the kidneys and the joints: The Gout.	0.5
2	 <p>Graph showing the variation of the concentration of urea in the plasma as a function of the quantity of consumed proteins.</p>	1.5
3-1	The concentration of urea in plasma increases from 0.2 g / L to 0.45 g / L when the quantity of consumed protein increases from 0.5 g / Kg to 2 g / Kg of body mass.	1
3-2	The origin of urea in plasma is the consumed proteins.	0.5
4	Urea is produced by the degradation of proteins. As the quantity of consumed proteins increases (from 0.5 g / Kg to 2 g / Kg ), the concentration of urea in the plasma increases from(0.2 g / L to 0.45 g / L ) . This high concentration of urea provokes the gout disease.	1.5

Q	Exercise 3 Passive Smoking	Mark												
1	Passive smoking is when someone involuntarily inhales cigarette smoke produced by neighboring smokers	1												
2	<table border="1" style="margin-left: auto; margin-right: auto;"> <thead> <tr> <th colspan="3">Frequency of respiratory troubles (%)</th> </tr> <tr> <th>Group</th> <th>Coughing</th> <th>Chronic bronchitis</th> </tr> </thead> <tbody> <tr> <td>A</td> <td>7</td> <td>4.8</td> </tr> <tr> <td>B</td> <td>10.5</td> <td>7</td> </tr> </tbody> </table> <p>A table showing the variation in the frequency of respiratory troubles in two groups A and B</p>	Frequency of respiratory troubles (%)			Group	Coughing	Chronic bronchitis	A	7	4.8	B	10.5	7	1.5
Frequency of respiratory troubles (%)														
Group	Coughing	Chronic bronchitis												
A	7	4.8												
B	10.5	7												
3	By referring to the results in the document, the frequency of coughing is 10.5% in the individuals of group B exposed to the cigarette smoke which is greater than that of the individual of group A that are not exposed to the cigarette smoke which is 7%. Moreover the frequency of chronic bronchitis is 7% in the individuals of group B that are exposed to cigarette smoke, which is greater than that of individuals of group A that are not exposed to the cigarette smoke which is 4.8%. This shows that passive smoking favors the development of respiratory troubles (coughing and chronic bronchitis). This is why the inhibition of smoking in closed public areas is a major action that protects non-smokers from passive smoking.	2.5												

Q	Exercise 4 Celiac Disease	Mark
1-1	The role of the intestinal villi is the intestinal absorption.	0.5
1-2	The characteristics of the intestinal wall are: -Large surface area -very thin wall -richness in blood vessels	1.5
2-1	In the normal individual, the internal wall of the small intestine shows normal villi, while in the affected individual having celiac disease; the internal intestinal wall shows damaged villi. In the normal individual, the internal wall of the small intestine shows normal intestinal cells while in the affected individual having celiac disease shows damaged intestinal cells. In the normal individual, the internal wall of the small intestine is folded while in the affected individual having celiac disease is flattened. In the normal individual, the villi are more vascularized than those of the affected individual.	1
2-2	The characteristic of the internal wall of an affected individual: - Decrease in the surface area of intestinal absorption - Less vascularized	1
3	The decrease in the surface area of intestinal villi leads to a decrease in the quantity of absorbed nutrients. The child will be unable to produce energy (by oxidation) and to form a new molecule (by assimilation) which allows growth. Thus the growth will be slowed down although his food diet is balanced.	1