This is a Class 1 exam

Lebanon Evangelical School for Boys and Girls

Lebanon Evangelical School
for Boys and Girls
Loueizeh - Baabda

Class 12 : Mock 2

10JUN2024 – 14JUN2024

Do NOT write in this box. Your MARK will be placed here.

Subject :	Biology	
Class :	12E	
Teacher :	Christine Khalil	
Duration :	2 hours 30 minutes	
Your name :		*Time taken to complete exam*
Your Class :		
Exam Room :		
How many sheets, including cover sheet :		

- Do not open this paper until the teacher in charge instructs you to do so.
- The most important thing to remember in taking an exam is NEVER PANIC.
- If you are caught cheating in this exam, you will be awarded zero for ALL exams that are taken on this day.

Exercise 1 (6 points)

Some chromosomal abnormalities in the fetus are at the origin of spontaneous abortions (fetal death). They can be detected by the preparation of a fetal karyotype. In order to detect the relationship between spontaneous abortions in women and the chromosomal abnormalities, some studies were done and the results are represented in document 1.

Chromosomal Abnormalities		Rate of spontaneous abortions
Trisomy	13	100%
	14	100%
	21	75%
Monosomy	13	100%
	14	100%
	21	100%

Document 1

1. Draw a histogram that represents the results of document 1. (1.5)

80% of the fetuses with chromosomal abnormalities can be identified through many signs that are revealed through the following 3 tests:

- The skinfold thickness of the neck of the fetus identified by ultrasound at 13 weeks of pregnancy.
- Blood levels of the fetal hormone HCG which is essential for the nutrition of the fetus during the first 3 months.
- Blood levels of fetal protein AFP.

Mrs. D suffered from several spontaneous abortions. Document 2 shows the results of these 3 tests during her last pregnancy that was interrupted at 13 weeks.

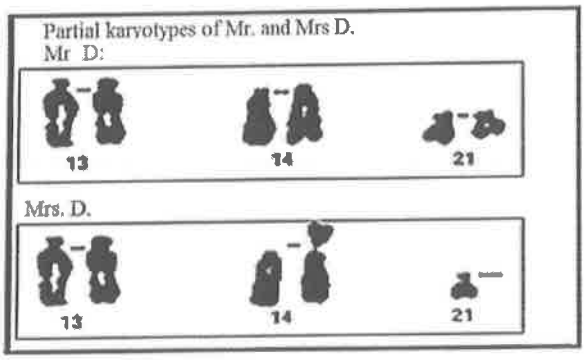
		Measured Values	Normal Values
Fetus echography at week 13	**Thickness of the skinfold at the neck**	5 mm	Less than 3 mm
Mrs. D blood tests at week 13	**AFP**	23.6 $U.mL^{-1}$	33.8±2.7 $U.mL^{-1}$
	HCG	80 $U.mL^{-1}$	35±25 $U.mL^{-1}$

Document 2

2. Deduce, referring to document 2, the cause of abortion of Mrs. D. (1)

Mrs. D is pregnant again and consulted a doctor who asked her to do a karyotype test with her husband. Document 3 shows the 3 pairs of chromosomes (13, 14 and 21) from the karyotypes of Mrs. D and her husband.

Mr. D and Mrs. D have normal phenotypes however only Mr. D has a normal karyotype.



Document 3

3. Compare the karyotype of Mrs. D to that of Mr. D. (1)
4. Explain the normal phenotype of Mrs. D. (0.5)
5. Make, by limiting yourself to the chromosomes 14 and 21, the factorial analysis that permits us to determine the risk for this couple to have a child affected by trisomy 21. (1.5)

In addition to the previous karyotypes, the doctor asked Mrs. D to repeat the 3 diagnostic tests she did during her last pregnancy. The new results are presented in document 4.

		Measured Values
Fetus echography at week 13	**Thickness of the skinfold of the neck**	2 mm
Mrs. D blood tests at week 13	**AFP**	34 $U.mL^{-1}$
	HCG	29 $U.mL^{-1}$

Document 4

6. Specify, by referring to documents 2 and 4, the state of the fetus of Mrs. D. (0.5)

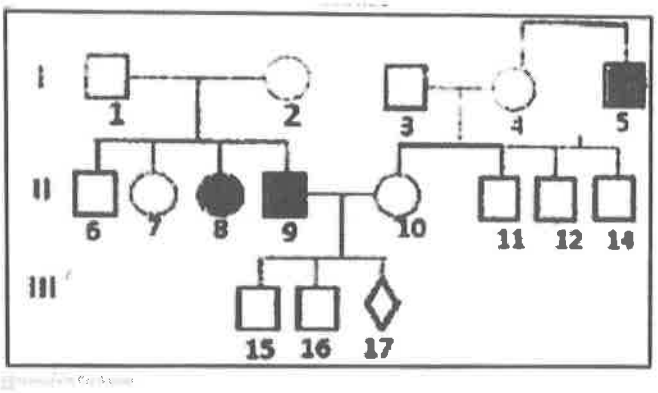
Exercise 2 (7.5 points)

Gaucher disease (GD) or toxic lysosomal storage disease is a genetic disease that affects the GBA gene, responsible for the synthesis of the enzyme glycocerebrosidase. This disease is characterized by bone deformations, kidney and liver damage…

Document 1 shows the pedigree of a family having some of its members affected by this disease.

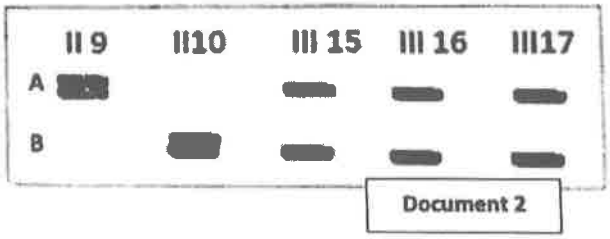
1. Indicate if the allele responsible for this disease is dominant or recessive. Justify your answer. (1)

2. Determine the localization of the gene responsible for this disease. (1)
3. Determine the risk of the fetus III-17 to be affected by this disease knowing that I-3 is homozygous. (1.5)



Document 1

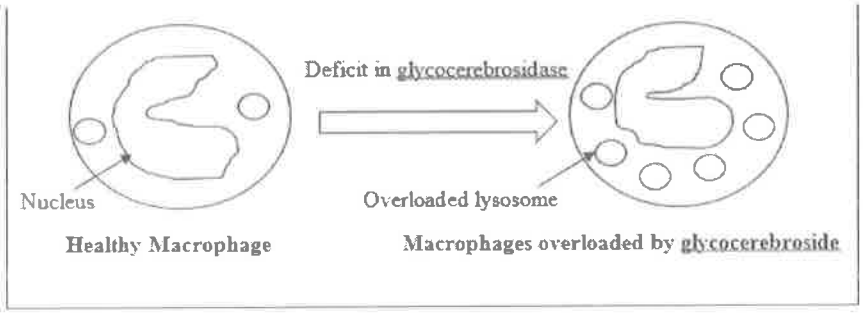
Couple II (9,10) is suspected to have an affected child. They perform a genetic study concerning the GBA gene to the different members of their family. The results are shown in document 2.



Document 2

4. Identify the band corresponding to the normal allele and that corresponding to the mutant allele. (0.5)
5. Indicate the genotype of couple II (9,10) and their descendants. (1)
6. After one year of the birth of child III-17, the symptoms of the disease begin to appear in this child. Formulate 2 hypotheses concerning the origin of this disease in III-17. (1.5)

In order to determine the cause of the symptoms of this disease, a study concerning the enzyme glycocerebrosidase and its effect on the cells rich in lysosomes, especially macrophages, is represented in document 3.



Document 3

7. Justify that GD is a disease of toxic lysosomal storage. (0.5)

The treatment of child III-17 consists of administrating a perfusion of the missing enzyme glycocerebrosidase in order to allow the normal functioning of its metabolism. However, this treatment didn't succeed and the symptoms didn't disappear. The administration of a molecule similar to saponin (protein synthesized in the organism and coded by the gene PSAP) eliminates the symptoms of the disease.

8. Verify which hypothesis is validated. (0.5)

Exercise 3 (5 points)

In order to study the relationship between the pituitary gland, the ovaries and the uterus during a menstrual cycle, 3 series of experiments were performed.

First series of experiments

Several female monkeys were subjected to the ablation of the pituitary gland. Each female was then subjected to different injections of pituitary hormones. The ovaries are examined after 12 then 17 days from the injection. The experimental conditions and the results are shown in document 1.

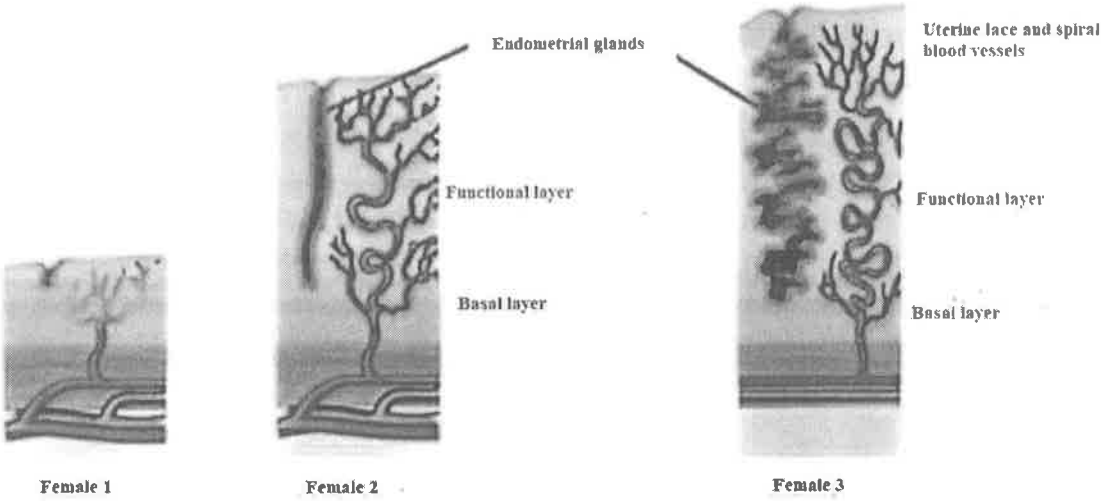
	Experimental conditions		**Results observed in the ovaries**	
Female	**Injection of FSH for 14 days**	**Injection of a high dose of LH**	**Graafian follicle after 12 days**	**Yellow body after 17 days**
1	-	-	-	-
2	+	-	+	-
3	+	Around day 14	+	+
4	+	Before the formation of the Graafian follicle	+	-

(-): absence (+): presence

Document 1

1. Indicate the neuro-hormonal stimulus responsible for the production of FSH and LH by the pituitary gland. (0.25)
2. Interpret the results of document 1. (1)

Document 2 shows, at day 20, the results observed at the level of the endometrium for the first 3 females of document 1.



Document 2

3. Explain the results shown in document 2 based on the results of document 1. (1.25)

To better understand the origin of the cyclic variations seen in a woman, we perform experiments on female monkeys since they have a cycle very similar to that observed in women.

Second series of experiments

We prepare culture media containing fragments of the uterus taken from female 1. The results are shown in document 3.

Culture Medium	Aspect of the endometrium after the culture
Nutritive medium	No secretion of glycogen and mucus
Nutritive medium enriched with estrogen	Presence of glands. No secretion of glycogen and mucus
Nutritive medium enriched with progesterone	Absence of glands. No secretion of glycogen and mucus
Nutritive medium enriched with estrogen and progesterone	Presence of developed glands. Secretion of glycogen and mucus

Document 3

4. Determine, based on document 3, the role of the ovarian hormones in the cyclic variations of the uterus. (1.25)

Third series of experiments

The size of the follicle is measured along with the concentration of LH during a normal cycle. The results are shown in document 4.

5. Identify the 2 types of feedback affecting the production of LH revealed by the results of document 4. (1.25)

Day of the cycle	2	4	6	8	10	12	13	14
Size of the follicle (mm)	5	6	8	10	16	20	22	-
LH concentration (UI/L)	15	12	11	10	10	15	100	25

Document 4

Exercise 4 (6.5 points)

Vaccination is a medical application which consists of injecting the individual with an attenuated antigen, a microbial substance... In this exercise, we are interested in identifying certain characteristics of vaccination.

Experiment 1: we consider 2 lots of macaque monkeys. Only lot 1 is immunized against virus X. Several days later, the 2 lots are exposed to virus X. The proportion of Tc cells specific to virus X is evaluated in the 2 lots. The results are shown in document 1A.

1A

Time (weeks)		0	1	2	3	4	5
Proportion of Tc L specific to virus X (a.u.)	Lot 1	0.5	7	6.5	6	5.5	4
	Lot 2	0	0	0.5	2	1.5	1

↑ Exposure to virus X

1B

Time (weeks)		0	1	2	3	4	5
Proportion of Tc L specific to virus Y (a.u.)	Lot 3	0	0	0.49	2	1.5	0.9
	Lot 4	0	0	0.5	2	1.5	1

↑ Exposure to virus Y

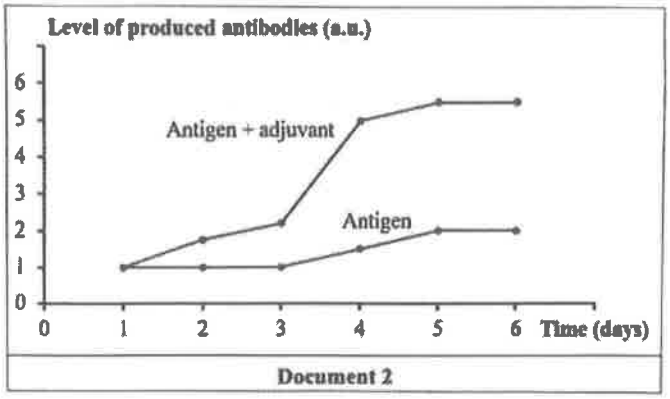
Document 1

1. Draw in the same graph the curves showing the variation in the proportion of TcL specific to virus X in the 2 lots as a function of time (document 1A). (1.5)
2. Draw out the action of vaccination on the immune response against virus X. (0.5)

Experiment 2: We consider 2 other lots of macaque monkeys. Only lot 3 is immunized against virus X. Several days later, the 2 lots are exposed to virus Y. The proportions of TcL specific to virus Y are evaluated in the 2 lots. The results are shown in document 1B.

3. Deduce from document 1B a fundamental characteristic of vaccination. (1.5)

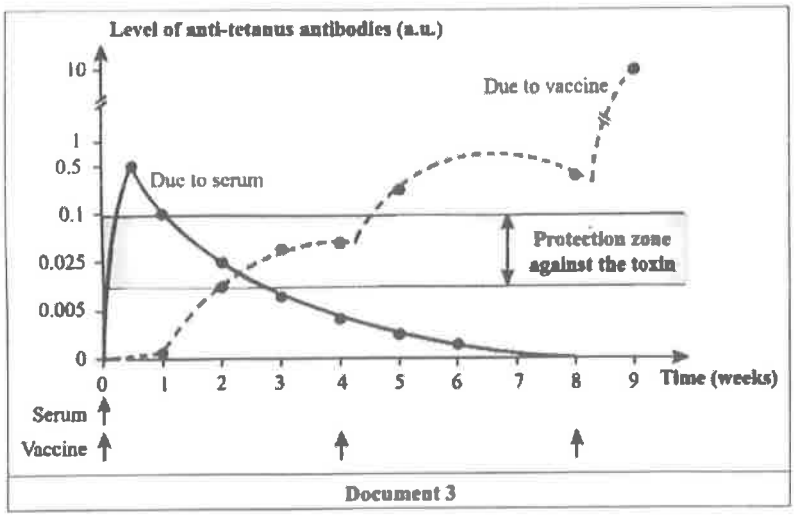
Experiment 3: in 1925, Gaston Ramon showed that diphtheria toxoid has an increased vaccination power if a "tissue irritant" is added to the vaccine. This substance is called an adjuvant. Document 2 shows the variation in the level of antibodies produced after immunization of mice against an antigen with or without adjuvant.



Document 2

4. Interpret the results of document 2. (1)
5. Formulate a hypothesis explaining the mode of action of adjuvants. (0.75)

Experiment 4: if an individual is infected with tetanus toxin, he is injected with a serum containing anti-tetanus antibodies. In addition, an anti-tetanus vaccine can be applied in 3 boosters (doses). Document 3 shows the variation in the level of anti-tetanus antibodies in the plasma of an individual who has undergone an injection of serum and an anti-tetanus vaccine.



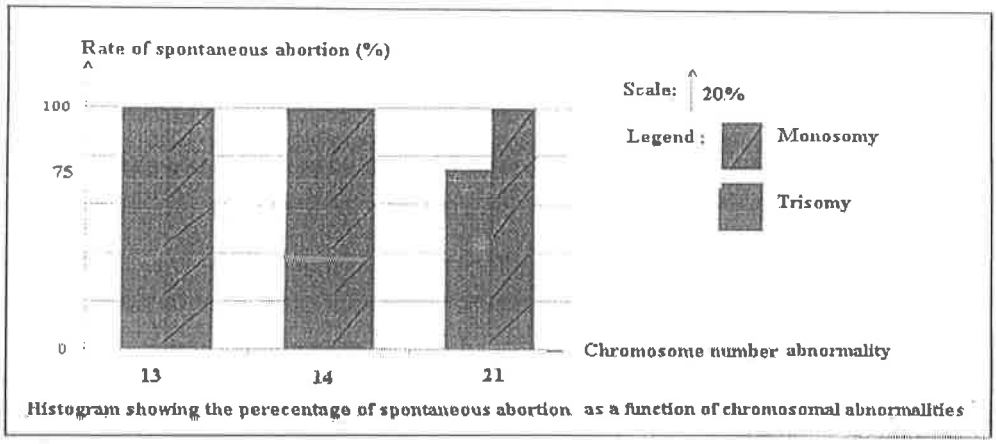
Document 3

6. Name the process of injecting anti-tetanus serum. (0.25)
7. Determine, according to document 3, the aim of applying the vaccine in many boosters. (1)

Answer Key for Mock 2 12E

Exercise 1 (6 points)

1. (1.5)



Histogram showing the perecentage of spontaneous abortion as a function of chromosomal abnormalities

2. The skinfold thickness of the neck of the fetus of Mrs. D at week 13 is equal to 5 mm which is higher than the normal value which is less than 3 mm. The blood levels of HCG of Mrs. D at week 13 are 80 $U.mL^{-1}$ which is higher than the normal values of 35 plus or minus 25 $U.mL^{-1}$. In contrary, the blood levels of AFP at week 13 was 23.6 $U.mL^{-1}$ which is less than the normal values of 33.8 plus or minus 2.7 $U.mL^{-1}$. Hence, the abortion of Mrs. D is caused by a chromosomal abnormality in the fetus. (1)
3. All the chromosomes of both karyotypes are made of 2 chromatids. Chromosomes 13 of Mr. D and Mrs. D have the same number that is 2 for each and they are of the same size also having the same position of the centromere. The number of chromosomes 14 of Mr. D and Mrs. D is the same equal to 2 but one of the 2 chromosomes 14 of Mrs. D is of same size and position of the centromere as both chromosomes 14 of Mr. D while the other is longer and has a different position of the centromere. Moreover, chromosomes 21 of Mr. D and Mrs. D are of same size and position of the centromere, but they have different numbers, Mr. D has 2 while Mrs. D has 1. (1)
4. Mrs. D has complete genetic information without excess nor loss. The missing chromosome 21 has been translocated to one of the 2 chromosomes 14. (0.5)
5. (1.5)

5- Factorial analysis: Genotypes: Mr.D 14 14 21 21 X Mrs.D 14 14^{21} 21

Gametes:	14 21	14 21	14^{21} 21	14	14^{21}
Proportions:	1	¼	¼	¼	¼

Table of cross:

	14 21 ¼	14^{21} 21 ¼	14 ¼	14^{21} ¼
14 21 1/1	14 14 21 21 ¼	14 14^{21} 21 21 ¼	14 14 21 ¼	14 14^{21} 21 ¼
	Normal ¼	Trisomy 21 ¼	Monosomy 21 ¼ lethal	Normal carrier ¼

6. The fetus is normal because the obtained values show a skinfold thickness of the neck equal to 2 mm, and levels of AFP and HCG respectively equal to 34 $U.mL^{-1}$ and 29 $U.mL^{-1}$ which are very close to the normal values. (0.5)

Exercise 2 (7.5 points)

1. The normal allele is dominant over the abnormal recessive allele because individuals I-1 and I-2 who are phenotypically normal have affected children II-8 and II-9 who should have received the affected allele from at least one of their parents who possess the abnormal allele in a masked way. (1)
   N: normal dominant allele d: mutant recessive allele
2. If the allele is located on the non-homologous segment of Y, no girls should be affected. However, girl II-8 is affected therefore this is not the case.
   If the allele is located on the non-homologous segment of X, then female II-8 who is affected and of genotype $X^d X^d$ should have received X^d from her father I-1 who should be of genotype $X^d Y$ and should be affected but this is not the case.
   If the allele is located on the homologous segment of X and Y, female II-8 who is affected and of genotype $X^d X^d$ should have received X^d from her father I-1. Her brother II-9 who is affected and of genotype $X^d Y^d$ should have received Y^d from his father I-1. Their father should be of genotype $X^d Y^d$ and should be affected but this is not the case. Therefore, the gene is autosomal. (1)
3. Individual I-4 is normal and has an affected brother, so her probability to be heterozygous is 2/3.
   Individual II-10 has father NN and 2/3 heterozygous mother thus by making the table of cross, the probability of II-10 to be heterozygous is ½.
   II-9 is affected dd then the risk for II-9 and II-10 to have an affected child is ddxNd= ½
   Total risk 2/3 x ½ x ½ – 1/6 (1.5)
4. Individual II-9 is affected dd in the pedigree and he has 2 bands of allele A then allele A corresponds to the allele of the disease while allele B corresponds to the normal allele. (0.5)
5. II-9: dd II-10: NN III-15, III-16 and III-17: Nd (1)
6. Hypothesis 1: the disease is caused by a mutant allele of an enzyme responsible for lysosomal storage.
   Hypothesis 2: the disease is due to a mutation at the level of another factor that plays a role with this enzyme. (1.5)
7. The normal individual and the individual affected by GD have the same nucleus but the affected one has overloaded lysosomes unlike the healthy individual. This was the result of the deficiency in glycocerebrosidase. Therefore, this disease is due to toxic lysosomal storage. (0.5)
8. Since the treatment of child III-17 with the missing enzyme didn't succeed and the symptoms didn't disappear but the administration of a molecule similar to saponin eliminated the symptoms, therefore, hypothesis 2 is validated where the disease in this child is due to a factor other than the enzyme. (0.5)

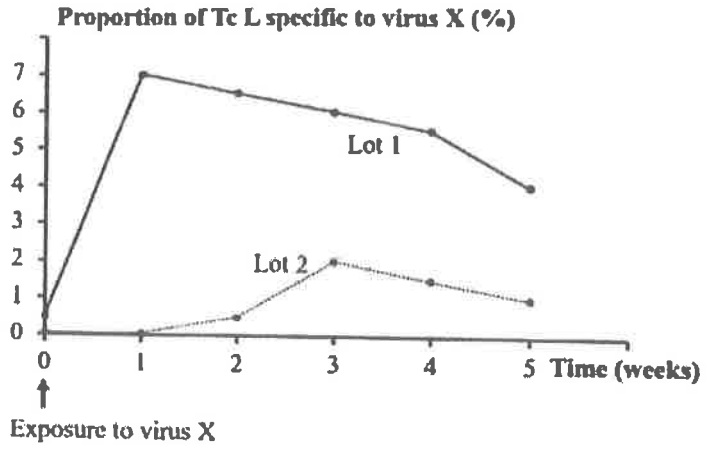
Exercise 3 (5 points)

1. GnRH. (0.25)

2. No Graafian follicle was observed in the ovaries after 12 days and no yellow body after 17 days without FSH and LH injections to the female monkeys that underwent pituitary gland ablation. When these female monkeys received FSH injections for 14 days, a Graafian follicle was observed without a yellow body. This indicates that FSH is responsible for the development of the Graafian follicle. A Graafian follicle and a yellow body are observed when FSH was injected for 14 days and a high dose of LH was injected around day 14. This indicates that LH is responsible for the formation of a yellow body. When FSH was injected for 14 days followed by an injection of LH before the formation of a Graafian follicle, a Graafian follicle was observed without the formation of a yellow body. This indicates that the formation of a yellow body under the effect of LH requires the presence of a Graafian follicle. (1)
3. Estrogen secreted by the follicles and the yellow body is the hormone responsible for the development of the endometrium, the blood vessels and the endometrial glands whereas the presence of progesterone amplifies this development. It allows the blood vessels to become spiral, the endometrial glands will secrete a nutritive substance and the uterine lace will appear. Female 1 has no follicular development therefore its estrogen levels don't increase justifying that the endometrium didn't develop. Female 2 has a more developed endometrium where blood vessels and glands start to appear due to the presence of estrogen secreted by the developed follicle (formation of a Graafian follicle). Estrogen and progesterone are present in female 3 due to the formation of the yellow body. This leads to the development of the endometrial glands, formation of the uterine lace and spiral blood vessels. (1.25)
4. No secretion of glycogen and mucus when the endometrium was cultured in a nutritive medium. However, glands start to appear at the level of the endometrium without secretion of glycogen and mucus when the endometrium was cultured in a medium having nutrients and estrogen. This indicates that estrogen alone stimulates the proliferation of the endometrium to form glands without secretions yet. No glands appear at the level of the endometrium and no secretion of glycogen and mucus when the endometrium was cultured in a medium having nutrients and progesterone. However, secretory glands start to appear in the endometrium with secretion of glycogen and mucus when it was cultured in a medium containing nutrients, estrogen and progesterone. This indicates that progesterone alone has no action on the uterus. However, when estrogen is present, progesterone amplifies the uterine development for better proliferation of the secretory glands to produce glycogen and mucus. (1.25)
5. The LH concentration decreases from 15 to 10 UI/mL when the size of the follicle increases from 5 to 16 mm. As the follicle increases in size, estrogen production increases to reach a moderate amount that exerts a negative feedback on the hypothalamo-pituitary axis decreasing LH production. LH concentration slowly increases to 15 UI/mL and then rapidly increases to 100 UI/mL when the size of the follicle increases from 6 to 12 mm. This variation is due to the transformation of the cavitary follicle into a Graafian follicle that is capable, due to its large size, to secrete a huge amount of estrogen. This amount exerts a positive feedback on the hypothalamo-pituitary complex leading to a LH peak. (1.25)

Exercise 4 (6.5 points)

1. (1.5)



Title: Curves showing the variation in the proportions of TcL specific to virus X in the 2 lots as a function of time

2. Vaccination makes the immune response faster and more amplified. (0.5)
3. Following the exposure to virus Y, the variation in the proportions of TcL specific to virus Y is the same: it is 0 in the 2 lots for one week then increases to 2 a.u. at 3 weeks then decreases to 1 a.u. at 5 weeks. Therefore, vaccination induces a specific immune response against a particular antigen. (1.5)
4. The level of antibodies produced is the same (1 a.u.) in both cases at day 1. It remains constant without adjuvant till day 3 and then increases to 2 a.u. at day 5 and remains constant till day 6. Whereas, with adjuvant, the level of produced antibodies slowly increases to 2 a.u. at day 3 and then rapidly progresses to 5 a.u. at day 4 where it remains almost constant till day 6. This indicates that the adjuvant stimulates antibody secretion. (1)
5. Hypothesis 1: the adjuvant stimulates the differentiation of BL into plasma cells OR Hypothesis 2: the adjuvant stimulates the multiplication of BL. (0.75)
6. Serotherapy. (0.25)
7. Following the 3 doses of the vaccine on days 0,4 and 8, the level of anti-tetanus antibodies increases to 0.0.5, 0.5 and 10 a.u. respectively. This last level becomes greater than the protection zone against the virus (0.012-0.1 a.u.). Therefore, the aim of applying the vaccine in many boosters is to provide immunity above the protection zone. (1)