This is a Class 1 exam

Lebanon Evangelical School for Boys and Girls

Lebanon Evangelical School

for Boys and Girls Loueizeh - Baabda

Stage Exam 06

19, 20, 21JUN2023

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

Subject :	Biology
Class :	12E
Teacher :	Christine Khalil
Duration :	2 hours 30 minutes

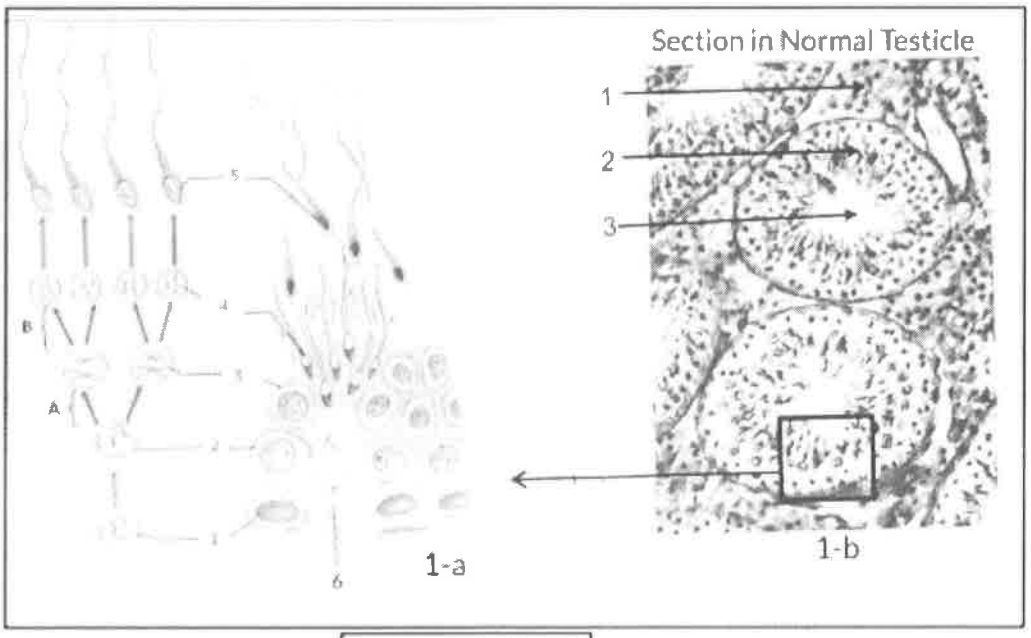
Your name :		*Time taken to complete exam*
Your Class :		
Exam Room :		
How many sheets, including cover sheet :		

- 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 (12 points)

Cryptorchidism (hidden testicles) is a defect in the migration of one testicle (unilateral) or both testicles (bilateral); they do not descend into the scrotum and remain in the abdominal cavity. This abnormality affects 1 in 25 boys at birth, but if we practice a simple surgical intervention before the age of two years, allowing the testicles to be repositioned in the scrotum, the structure of the testicles at puberty is perfectly normal.

Document 1 shows a section in the seminiferous tubule in a normal male.

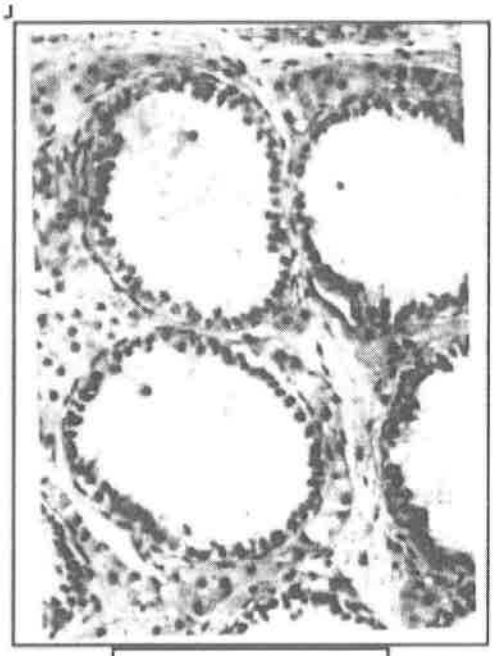


Document 1

1. Label document 1-a and document 1-b. (2.25)
2. Mention the structural modifications happening while the cell transforms from 4 to 5 in document 1-a. (1.25)

Document 2 shows a similar section in the seminiferous tubule of Mr. X who suffers from cryptorchidism.

3. Compare the section of document 2 to that in document 1-b. (2)
4. Based on document 1 and 2, show that:
   a. Mr. X is sterile. (1)
   b. Mr. X has normal secondary sexual characteristics. (1)



Document 2

Factors acting on the fetus in utero are often involved in this condition: significant maternal diabetes during pregnancy, excessive caffeine or regular alcohol intake during pregnancy. These factors triple the risk of cryptorchidism. Toxic substances acting as endocrine disruptors are also implicated like certain pesticides used in agriculture and gardening.

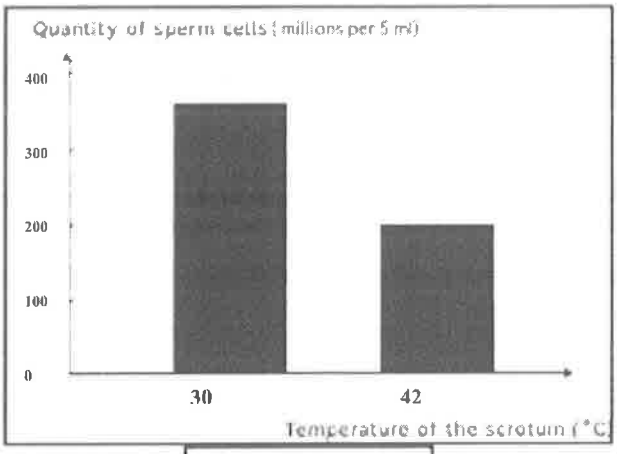
The medical follow-up of the mother during pregnancy and of the newborn during the first months is therefore decisive in preventing or detecting cryptorchidism.

5. Pick out from the text above:
   a. Factors that increase the risk of cryptorchidism. (0.5)
   b. Preventive measures to detect cryptorchidism. (0.5)

The risk of infertility in adulthood increases by 60% for a baby with untreated cryptorchidism. The completion of the development of the testicle and the acquisition of its capacity to produce spermatozoa require a temperature between 30 to 34°C, therefore lower than the normal temperature of the body.

Document 3 shows the effect of temperature on the production of sperm cells.

6. Interpret the results of document 3. (1.5)
7. Explain, based on all what preceded, how untreated bilateral cryptorchidism can cause infertility. (2)



Document 3

Exercise 2 (11 points)

A doctor seeks a remedy to the sterility of a couple: Mrs. X and Mr. X. In order to achieve it, he requests a spermogram for the husband and the results are represented in document 1.

	Spermogram					
	Volume	**pH**	**Viscosity**	**Number of sperm cells**	**Normal shape**	**Mobility**
Mr. X	4.1 mL	7.3	Normal	$9x10^6$/mL	90%	52%
Normal male	>3mL	7.3	Normal	$>60x10^6$/mL	>56%	52%

Document 1

1. Compare the results of document 1. (1.5)

2. Draw out the abnormality of Mr. X. (1)

To achieve pregnancy, the doctor prescribes IVF and ET (*In Vitro* Fertilization and Embryo transfer) technique where sperm cells from the husband and oocytes from the wife will be fertilized *in vitro* and then the selected embryo(s) will be transferred to the uterus of Mrs. X. Three embryos were obtained E1, E2 and E3.

Knowing that Mr. X also suffers from a genetic disease and Mrs. X is normal, the couple needs to choose among the three embryos. For a better choice, the doctor prescribes electrophoresis for the couple and for the three embryos. The results are presented in document 2.

3. Specify the allele of the disease. (1)

4. Determine the mode of transmission of the disease. (The gene is not located on the homologous segment of X and Y). (3)

5. Indicate which of the three embryos is/are most suitable to be implanted. Justify. (1.5)

	Mr X	Mrs X	Embryos		
			E1	E2	E3
A1	2	1	1	2	2
A2	0	1	1	1	0
A1 and A2 are the alleles of the gene					

Document 2

Document 3 represents the karyotypes of two germ cells at the origin of the embryo E2.

- Figure a represents the karyotype of the oocyte II,

- Figure b shows the karyotype of the spermatocyte II.

Document 3

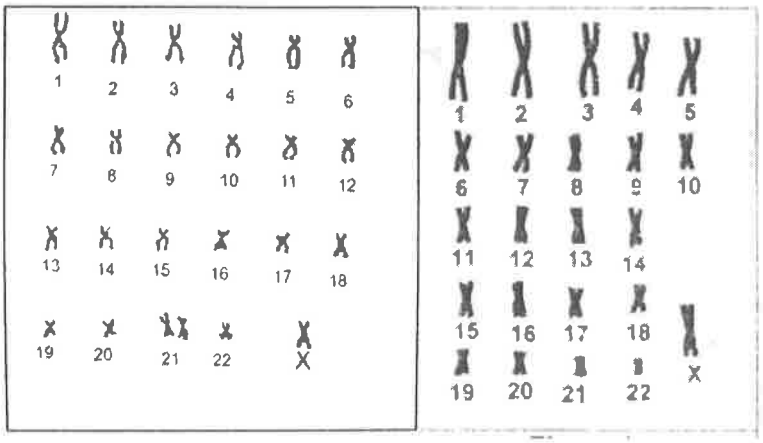


Figure a **Figure b**

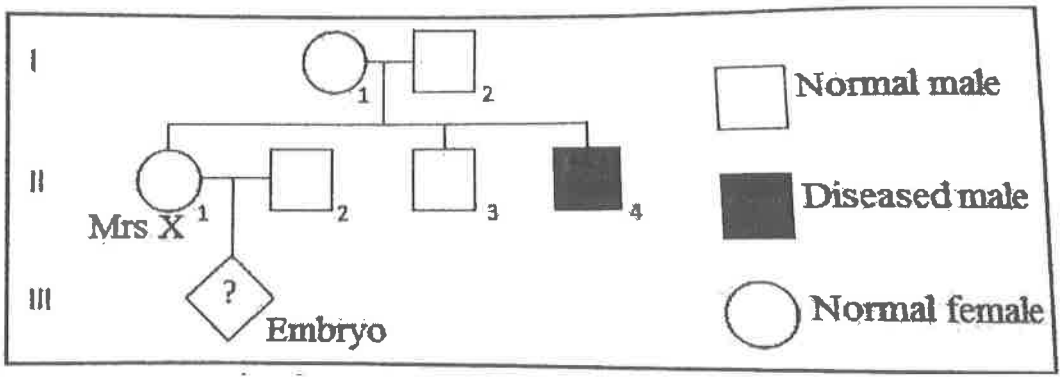
6. Illustrate the behavior of chromosomes in the parent responsible for the error obtained. (Consider pairs 21 and 23 only) (2)

7. Do the karyotypes of document 3 confirm the location of the gene? Justify your answer. (1)

Exercise 3 (11.5 points)

Hemophilia A is a hereditary disease caused by an abnormal blood coagulation factor, factor VIII.

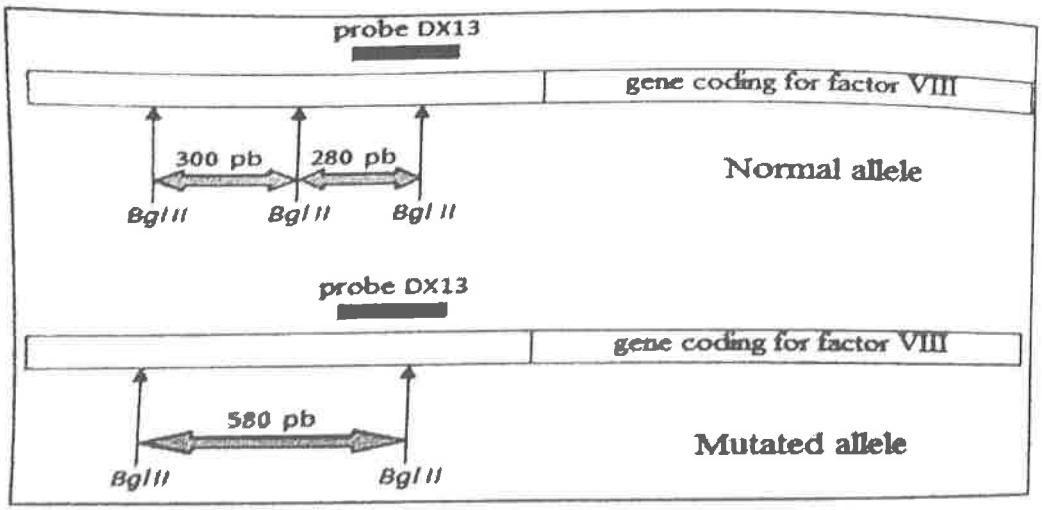
Mrs. X, whose brother has hemophilia, is pregnant and her doctor requested a prenatal diagnosis during her sixth week of pregnancy. Document 1 shows the pedigree of Mrs. X. (Note that her father shows no mutant allele using DNA fingerprinting.)



Document 1

1. Is the allele coding for hemophilia dominant or recessive? Justify. (1)
2. Using logical reasoning, prove that the gene is located on the non-homologous segment of the X chromosome. (1.5)
3. Determine the genotypes of individuals I-1, II-1, II-2 and II-4. (2)
4. Calculate the risk for the embryo to be affected. (2)

Document 2 shows the location of the restriction sites of the enzyme Bgl II and the site of hybridization of the probe DX13. The probe DX13 hybridizes with a complementary sequence near the gene coding for factor VIII. Scientists believe that it characterizes the mutation of this gene.



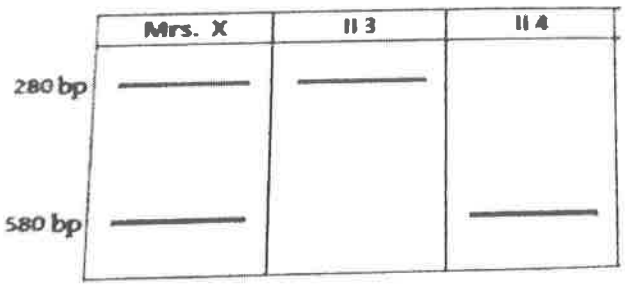
Document 2

The analysis of the karyotype of the embryo carried by Mrs. X indicates that the embryo has the following chromosomal formula: 46,XY.

Moreover, DNA analysis is done for Mrs. X and for individuals II-3 and II-4 according to the following protocol:

- Southern blot technique is used.

- DNA of the individuals mentioned are degraded with the help of the restriction enzyme Bgl II.
- The DNA fragments obtained are separated by electrophoresis after denaturation.
- Radioactive probe DX13 is used.
- Autoradiography is realized and the obtained results are shown in document 3.



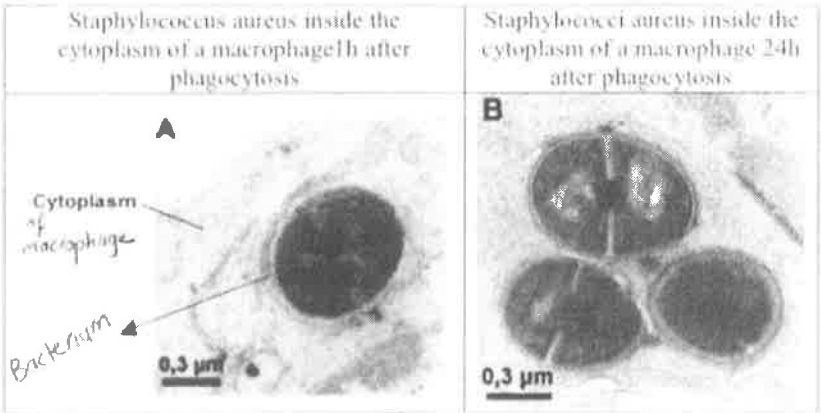
Document 3

5. Use the restriction map to confirm that the gene coding for hemophilia is located on the non-homologous segment of chromosome X. (2)
6. Give the genotypes of the individuals shown in document 3. (1.5)
7. Given all the information above, calculate the new risk for the embryo to be affected. (1.5)

Exercise 4 (10.5 points)

Staphylococcus is a bacterium found on the skin and in the nasal passages of human beings. Sometimes, virulent staphylococcus strains can cause infection, most often local but which can also be generalized and potentially life-threatening when bacteria enter the blood circulation and spread to other organs. To fight against this bacterium, we use antibiotics, chemical substances that can destroy bacteria or prevent their development, but they are sometimes inefficient and recurrence of the infection may occur. Researchers have tried to improve antibiotic treatment by combining them with antibodies.

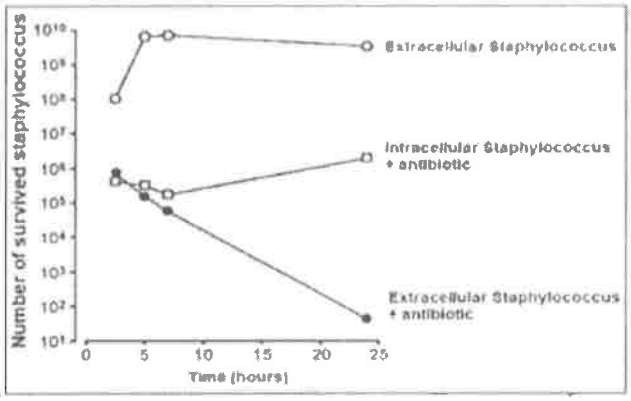
Document 1 shows a micrograph taken under an electron microscope of a macrophage having phagocytozed staphylococcus 1 hour and 24 hours after phagocytosis.



Document 1

1. Determine, based on document 1, the efficiency of the macrophage against staphylococcus. (2)
2. Mention 2 causes that may lead to the result shown in document 1 (B). (2)

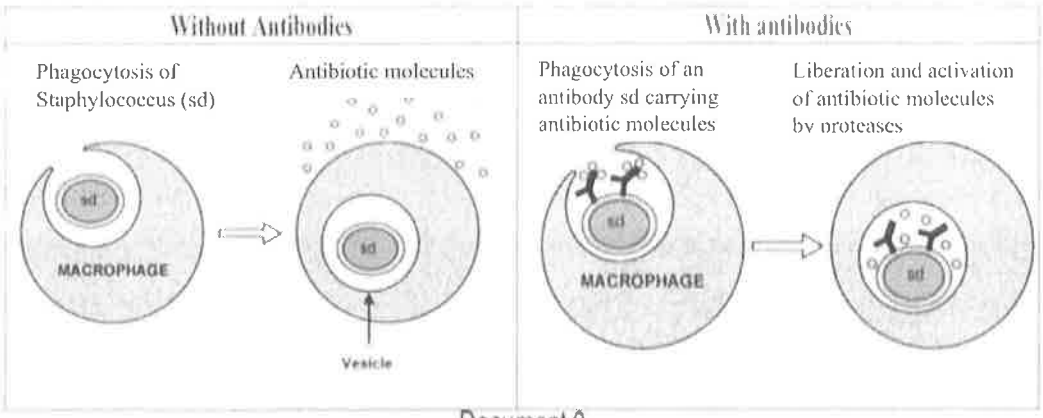
The graph of document 2 shows the action of an antibiotic, injected at t=0 on *Staphylococcus aureus* present outside cells (extracellular medium) or inside cells (intracellular medium).



Document 2

3. Interpret the results of document 2. (2.5)

To improve the antibiotic treatment against intracellular forms of *Staphylococcus aureus*, we associate an inactive antibiotic to an antibody that is specific for these bacteria. This inactive antibody-antibiotic complex recognizes these bacteria (sd) and binds to them. The mechanism is shown in document 3.



Document 3

4. Based on document 3, indicate the steps that lead to the activation of the antibiotic. (2)
5. Show that, based on document 3, the intracellular bacteria can only be destroyed by antibody-antibiotic complexes. (2)

Bareme for Mock 12E

Exercise 1 (12 points)

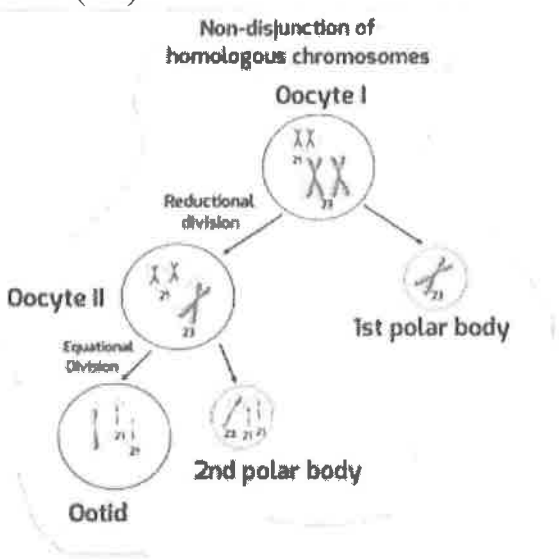
1. Figure 1-a 1- spermatogonium; 2- Spermatocyte I; 3-Spermatocyte II; 4- Spermatid; 5- Sperm cell.; 6- Sertoli cell.
   Figure 1-b: 1- Leydig cell; 2-Seminiferous tubule; 3- Lumen (0.25 each)
2. a. Golgi apparatus gives rise to the acrosome.
   b. Distal centriole gives rise to the tail.
   c. Mitochondria aggregate at the middle forming the mid-piece.
   d. Part of the cytoplasm detaches (residual cytoplasm).
   e. The rounded spermatid becomes an elongated sperm cell. (0.25 each)
3. Similarities: The diameter of the seminiferous tubules in both diagrams is approximately the same. Moreover, both contain Leydig cells. (1)
   Differences: the lumen of the seminiferous tubule in doc 2 is much wider than that of doc 1-b. The seminiferous tubules in doc 1-b contain all germ cells but in document 2 they contain only spermatogonia. (1)
4. a. Document 2 shows that the seminiferous tubule of Mr. X contains only spermatogonia and absence of gametes (sperm cells) in contrary to the normal one where all germ cells are present including the gametes. This means that there is no spermatogenesis occurring in the testes of Mr. X. So, Mr. X is sterile. (1)
   b. Document 2 shows that the section which is similar to Mr. X's case contains leydig cells identical to that in normal section (doc. 1-b). Knowing that leydig cells secrete testosterone, which is a hormone responsible for the appearance of secondary sexual characteristics. So, Mr. X has normal secondary sexual characteristics. (1)
5. a. Significant maternal diabetes during pregnancy, excessive caffeine or regular alcohol intake during pregnancy, toxic substances like pesticides. (0.5)
   b. Medical follow-up during pregnancy and of the newborn during the first months. (0.5)
6. The quantity of sperm cells is 350 million per 5 ml when the scrotal temperature is 30 C which is higher than that obtained when the temperature increases to 42 C which is equal to 200 million per 5 ml. (0.75) This implies that the rise in temperature attenuates the production of sperm cells. (0.75)
7. In case of bilateral cryptorchidism, the two testes remain in the abdominal cavity and do not descend to the scrotum. If not treated, the testes are present under the body temperature 37 C which is greater than the optimum temperature for the production of sperm cells (30 to 34 C) and referring to doc. 3 as the temperature increases, the rate of spermatogenesis decreases. In untreated individuals, there is prolonged exposure to such high unsuitable temperature with respect to spermatogenesis which will finally lead to the cease of the production of sperm cells as shown in document 2. This explains why people with untreated bilateral cryptorchidism are infertile. (2)

Exercise 2 (11 points)

1. The spermogram of Mr. X shows that the volume is 4.1 ml which is greater than 3 ml (the normal value). The pH of both is 7,3 also the viscosity is normal in both. Moreover, the percentage of normal shape in Mr. X is 90% which is greater than 56% (the normal value). In addition, the mobility in both is 52%. However, the number of

sperm cells for Mr. X is $9x10^6$/ml which is much less than that of the normal male: >60 $x10^6$/ml. (1.5)

2. Mr. X suffers from production of low number of sperm cells (oligospermia). (1)
3. Allele of the disease is A1 since Mr. X is affected and document 2 shows that he has only allele A1. (1)
4. Mrs. X is normal and she possesses two alleles A1(allele of the disease) and A2 (the normal allele). This means that the normal allele is expressed in the phenotype and the allele of the disease is masked. So, allele A2 is dominant and allele A1 is recessive. (1) With respect to the location: The studied gene is not located on the non-homologous segment of X or non-homologous segment of Y since in both cases Mr. X must have only one allele but document 2 shows that he has two alleles A1. So, the studied gene is located on an autosome. (2)
5. E1 is the suitable embryo for implantation since E2 has 3 alleles (2 alleles of A1) and one allele of A2 which means that he has trisomy (0.5) and E3 has 2 defective alleles of (A1) which means that he will be born affected by the genetic disease (0.5) but E1 has 2 alleles which means that he does not suffer from chromosomal abnormality. Also, the fetus will be born healthy since he has 2 alleles of A2 which is the normal allele. (0.5)



6. (2)
7. Yes, since document 2 shows that E2 carries three alleles (2 alleles of A1 and one for A2). Each allele must be carried by one chromosome. The karyotypes of document 3 show that after fertilization the zygote will carry 3 copies of chromosome 21 but the other chromosomes are found in pairs. This means that the gene is carried by chromosome 21 which is an autosomal pair. (1)

Exercise 3 (11.5 points)

1. I-1 and I-2 are phenotypically normal but they have an affected child II-4 that should have received the diseased allele from at least one of his parents who have this allele masked by the normal allele. The disease allele is recessive while the normal allele is dominant. (1) N=normal allele h=diseased allele.
2. If it is autosomal then II-4 should be of genotype hh and should receive the h allele from both parents but I-2 doesn't carry any affected allele. Then it is not the case. (0.5)

If it is located on the non-homologous segment of Y, then II-4 should be of genotype XY^h and should have received Y^h from his father I-2 who should then have the same genotype and be affected but this is not the case. (0.5)
If it is located on the homologous segment of X and Y, then II-4 who is affected should be of genotype X^hY^h and should also receive Y^h from his father I-2 but his father doesn't carry any affected alleles therefore this is not the case. (0.5)
The gene is located on the non-homologous segment of the X chromosome.

3. I-1 $X^N X^h$ because she is phenotypically normal but she has an affected child II-4 that should receive X^h from her. (0.5)
II-1 $X^N X^N$ $X^N X^h$ because she is phenotypically normal which is dominant and can be expressed in the homozygous or the heterozygous state. (0.5)
II-2 X^N Y because he is phenotypically normal and expresses the only allele he has on his X chromosome. (0.5)
II-4 X^h Y because he is affected and expresses the only allele he has on his X chromosome. (0.5)
4. II-2 is phenotypically normal and must be of genotype X^N Y. No matter what the mother's genotype is, his daughters will inherit X^N from their father and be normal. The risk for a girl to be affected is 0. (1)
If it's a boy, he will receive Y from his father II-2. His mother will decide if he will be affected or not. The probability for Mrs. X to be heterozygous is ½ and her probability to give X^h to her son is ½. Probability of a boy to be affected =1/2x1/2= ¼ (1)
General probability for a child to be affected is ¼ x1/2=1/8 (probability of being a boy is ½)
5. Since Mrs. X has 2 bands indicating the presence of 2 alleles and each of the males II-3 and II-4 has 1 band indicating the presence of 1 allele only. Therefore, the gene is located on the non-homologous segment of the X chromosome since if it was located on an autosome or the homologous segment of X and Y, there should have been 2 bands for each individual and it was Y-linked then males should present 1 band only and no bands for females. (2)
6. The 580 band corresponds to the affected allele where the restriction enzyme couldn't cut while the 280 band corresponds to the normal allele cut by the restriction enzyme.
Mrs. X $X^N X^h$ II-3 X^N Y II-4 X^h Y (1.5)
7. We are now sure that Mrs. X is heterozygous so her probability is 1. We are sure the baby is a boy. The probability of Mrs. X to give the affected allele to her son if her genotype is $X^N X^h$ is ½. The new probability is ½. (1.5)

Exercise 4 (10.5 points)

1. After one hour of phagocytosis there is one bacterium inside the cytoplasm of the macrophage but after 3 hours there is multiplication of the bacterium to become 3 bacteria. This means that the macrophage did not digest the bacterium. So, the macrophage is inefficient against staphylococcus. (2)
2. a- The bacterium is resistant to lysozymes.
b- The digestive enzymes (lysozymes) are deficient (each is 1).
3. The number of survived extracellular staphylococcus bacteria increases from 10^8 to 10^{10} during the first 5 hours and remains approximately around this value till 25 hours

but when these bacteria are cultured with antibiotic their number decreases from 10^6, at 2 hrs, to $5x10^1$ as the time increases to 25 hrs. This means that antibiotic reduces the multiplication of bacteria. (1.5) On the other hand, when intracellular bacteria are cultured with antibiotic their number decreases slightly from 10^6, at t=2hrs, to 10^5 at t=7.5 hrs but it increases again to reach $5x10^6$ as the time increases to 25 hrs in contrary to what observed when the bacteria are extracellular and cultured with antibiotic. This implies that antibiotic is effective only against extracellular bacteria. (1)

4. Steps of activation of the antibiotic:
   1. Antibodies coupled to antibiotic bind to bacteria.
   2. Macrophage engulfs the bacteria with the bounded antibodies.
   3. Inside the phagosome the antibiotic is liberated.
   4. After liberation, antibiotic is activated by proteases. (each step 0.5)
5. Referring to document 3, without antibodies the antibiotic cannot reach the bacteria inside the phagosome of the macrophage. While when antibiotic is coupled with antibodies, it can reach the bacteria and become active, after liberation, by proteases since macrophage phagocytes the antibiotic with the antibodies that are bound to the bacteria. This means that the intracellular bacteria can only be destroyed by antibody antibiotic complex. (2)