

Exercise I: A condition of fertilization.

(5 pts.)

A- In vitro fertilization is a technique that can be applied in many mammalian species. This technique can be associated with many experiments, which enable us to understand the mechanism of fertilization in mammals.

Experiment 1: Sperms are not able of binding on zona pellucida of oocyte II (at metaphase II) that was previously treated by enzymes extracted from cortical granules vesicles.

Experiment 2: We isolate molecules from zona pellucida of an oocyte II (metaphase II) of a mouse, identified as glycoproteins, and known as Zp3. We treat these molecules with radioactive isotopes then we place them with mouse sperms. The observation by autoradiography technique shows that radioactivity is found at the membrane of the sperms.

- 1- Interpret each experiment.
- 2- Formulate a hypothesis concerning the role of Zp3.

B- The meeting of some male and female gametes may lead to genetic disorder. Figure 1 is a caryotype made using a cell from an embryo expelled during a stopped pregnancy.

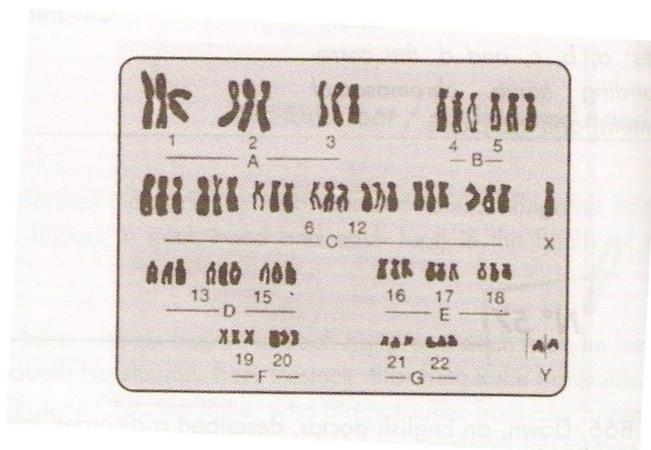


Figure 1

- 1- What disorder is detected in this caryotype?
- 2- This caryotype leads to think that the female gamete was fertilized by two male gametes. Indicate why this hypothesis is in contradiction with your knowledge about fertilization. The examination of the father's gametes shows some abnormal spermatozoa such as the one represented in figure 2.

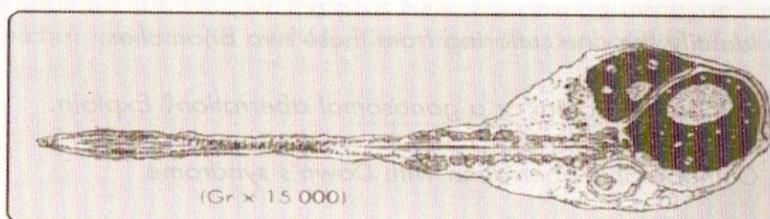


Figure 2

- 3- What abnormality in the sperm cell does this figure show? Indicate the real cause of the abnormal caryotype of figure 1.
- 4- Explain at which stage of spermatogenesis this abnormality in male sex cell takes place.

Exercise II: The effect of irradiation on genetic information.

(3.5 pts)

The drosophilae of wild phenotype have eyes of red color. The mutant drosophilae have yellow eyes (phenotype zest). We want localize the gene zest which plays a role in the character: color of eyes.

Document 1: genetic information, the results of 3 crosses.

The crossed drosophilae are different by the phenotype « color of eyes » which is determined by 2 alleles of the gene zest, allele (+) and allele (z). The phenotype red eye is symbolized {+}; the phenotype yellow eye is symbolized {z}.

	Individuals	Results of cross
1st Cross	Female [+] × male [z] homozygote	50 % males [+] 50 % females [+]
2 nd Cross	Femelle [z] × male [+] homozygote	50% males [z] 50 % females [+]
3rd Cross	Femelle [z] × male [+] Homozygote, irradiated with X rays Non- irradiated with X rays	50 % males [z] 50% : - females [+] -rare females [z]

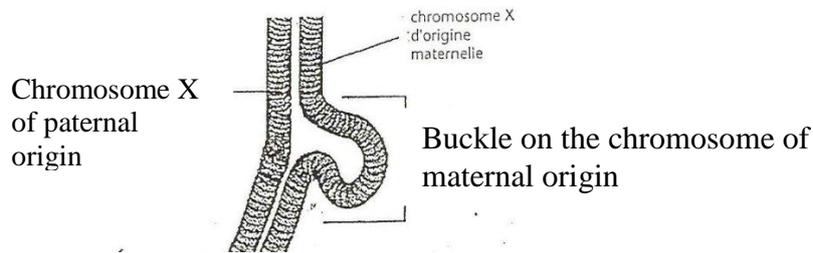
- 1- Based on document 1, determine the mode of transmission and the localization of the gene of the character color of eyes in drosophilae.
- 2- Formulate a hypothesis which explains the appearance of the phenotype « zest » in certain females of the 3rd cross.

Document 2: Consequences of the irradiation

The irradiation (exposure) to X rays causes in certain cases in drosophilae a loss of fragments of chromosomes. In the case of a loss of a fragment on one of the two homologous chromosomes, the chromosome which didn't have any loss makes a buckle in the region where the other chromosome presents a loss.

This diagram presents the observation of two chromosomes X of a rare female [z] of the 3rd cross of document 1:

Chromosome
X of maternal
origin

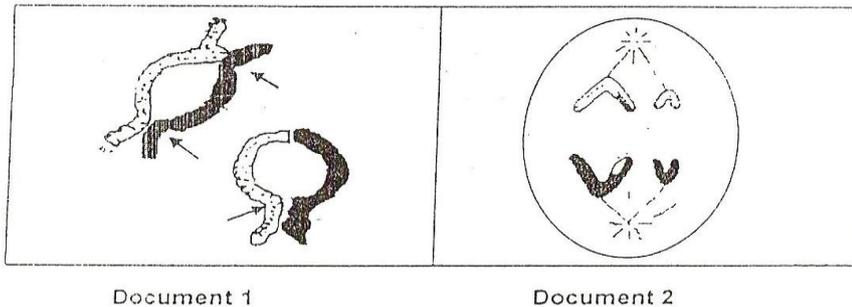


- 3- Use the document 2 to confirm or infirm the hypothesis proposed in question 2.
- 4- Draw the pair of chromosomes implicated in the appearance of the phenotype « color of eyes » in drosophilae of each of the 3 descendants of the 3rd cross.

Exercise III: stages of meiosis.

(3 pts.)

The documents 1 and 2 represent the chromosomes of a cell in meiosis.

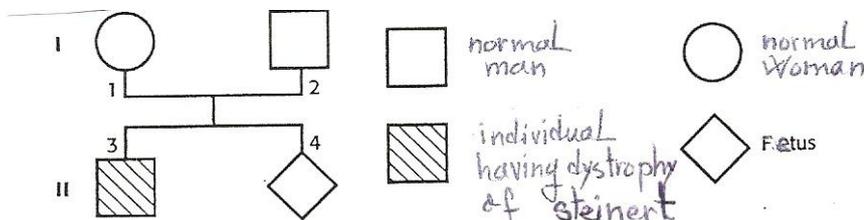


- 1- Precise at which stages of meiosis the chromosomes in each document are found.
- 2- What is the significance of the points marked by arrows in document 1?
- 3- Why some chromosomes are mixed in color (white and black) in document 2?
- 4- Draw the other possibility of distribution of chromosomes in document 2.
- 5- Draw the anaphase of the second division of meiosis in document 2.
- 6- By taking into consideration the responses of the above questions, determine the genetic consequences of meiosis.

Exercise IV: The origin of a dominant disease.

(7 pts.)

The dystrophy of Steinert is a hereditary disease affecting the muscles. The first child of the couple I-1 and I- 2 of the pedigree below is affected by this dystrophy. The parents want to know the risk of having the other child affected.



The gene at the origin of this disease is carried by an autosome and the allele responsible for this disease is dominant. However, in certain families we can see the appearance of this disease in a generation.

- 1- Knowing that the allele of this disease is dominant, formulate a hypothesis which explains the cause of the appearance of this disease in the child II-3.

Document 1: The myopathy of Steinert belongs to the group of "diseases by triplets". The alleles of this gene have a big number of a successive triplets CTG. The phenotype depends on the number of triplets.

alleles	Number of triplets(n)	Phenotype of individual having this number
1-	$N \geq 7$	No symptoms(normal)
2-	$N \geq 36$	No symptoms(normal)
3-	$N \geq 81$	Symptoms of the disease appear and they increase with the number of triplets.

The alleles having a number of triplets bigger than 35 are expressed normally but this number of triplets can increase from a generation to another.

- 2- Draw a histogram that represents the number of triplets in the alleles 1, 2 and 3.
- 3- Analyze the document 1, what can you deduce about the genetic cause of the disease?

Document 2: We use the technique of Southern blot. We digest the DNA of the genome by using two restriction enzymes: BglII and HindIII which cut the repetitive sequence of triplets from its two sides. The restriction fragments are separated by electrophoresis then denatured. The radioactive probe hybridizes with the complementary sequences and reveals the place of the fragment (figure 1).

This technique is done on the members of the family; the results are shown on figure2:

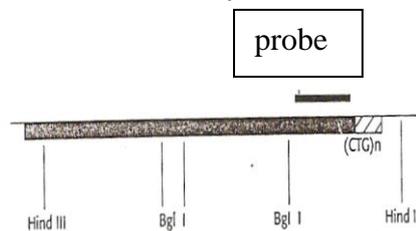


Figure 1: The hybridization of the probe showing the place of the fragment.

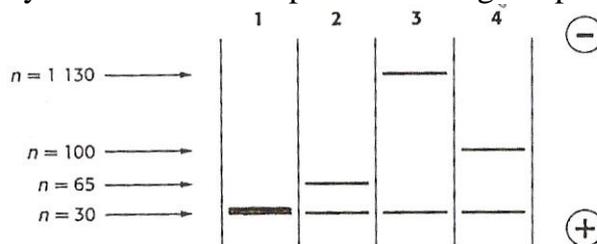


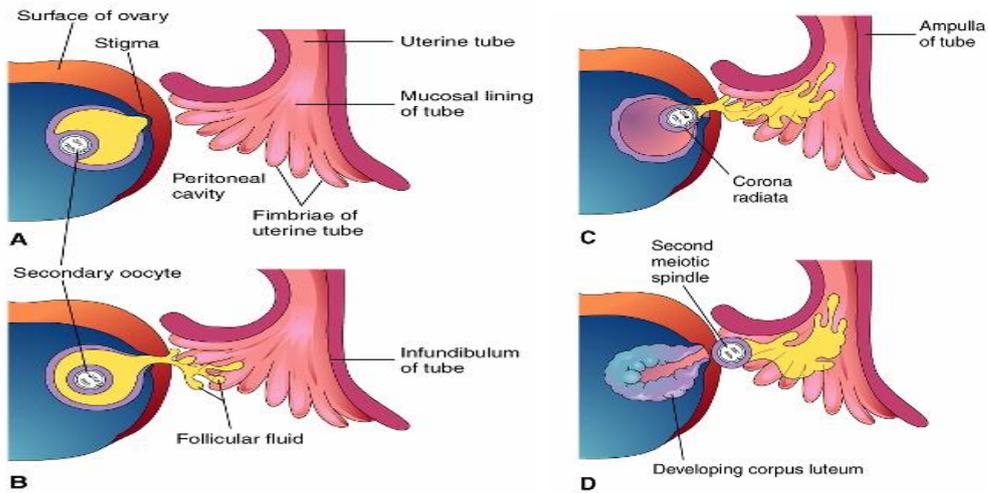
Figure 2: results of the individuals of the pedigree of document 1.

- 4- Determine the origin and the role of the restriction enzymes.
- 5- By referring to the documents 1 and 2, identify the DNA fragments that correspond to each of the alleles 1, 2 and 3 of document 1.
- 6- Indicate the genotypes of the parents 1 and 2 and that of their child 3, justify your answer.
- 7- Does the anxiety of the parents about having another affected child is logical? Justify your answer by precisig the genotype of the fetus4.

Exercise V: A stage of ovarian cycle.

(1.5 pts.)

- 1- Name the process that takes place in the following figures.
- 2- Describe the events represented in these figures.



GOOD WORK