

Question-5-

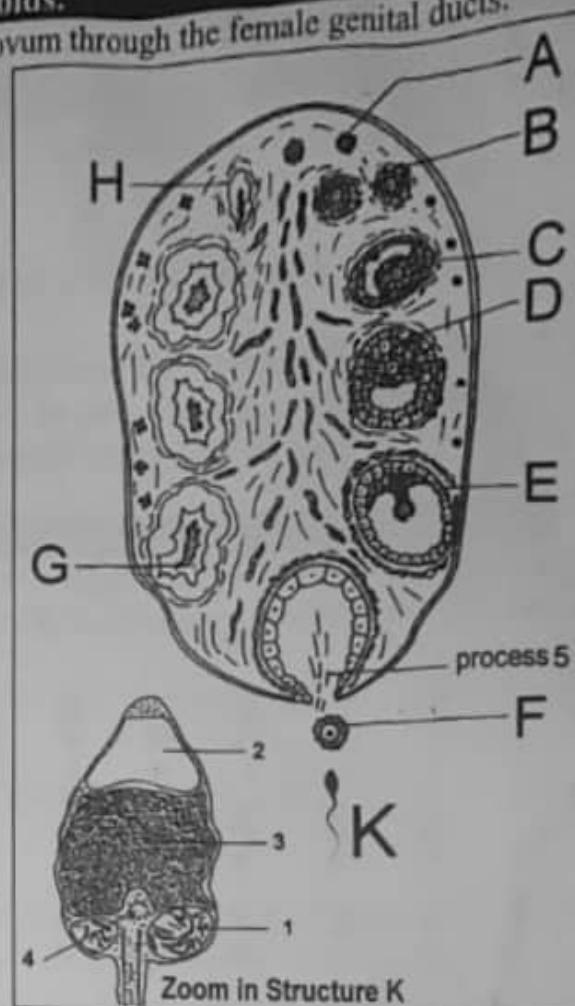
Reproduction and Spermatozoids.

The spermatozoid is a very specific cell capable of moving to meet the ovum through the female genital ducts.

1- Label A to K then 1 to 5.

- ❖ Exp₁. Spermatozoa are removed from the seminiferous tubules and are introduced into the uterus. No significant migration towards the fallopian tubes is observed.
- ❖ Exp₂. The sperm taken from the vas deferens canal is introduced into the uterus; fertilization is normal.
- ❖ Exp₃. The sperm taken from the vas deferens canal is put in a test-tube in the presence of ova. No fertilization was possible.
- ❖ Exp₄. Injection of spermatozoa into an oocyte leads to fertilization. The element 2 of figure 1 remains intact, but it normally opens when in contact with the oocyte.
- ❖ Exps. We separate active spermatozoa from the remaining sperm, before placing them in a physiological solution deprived of fructose. Sperms become immobile & incapable of fertilization.

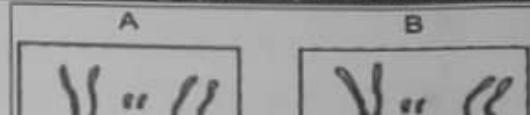
2- Interpret the following experiments; conclude the conditions needed for spermatozoids action.



Question-6-Meiosis in drosophila

The following drawings represent the Karyotypes of two drosophila A and B of different sex of $2n = 8$.

1- Knowing that the male drosophila contains different



Risk in Gonosomal Disease, (non-homologous part of X)

G. Risk = Probability of
the mother to
be heterozygous \times Probability to
have an affected
child

- if the mother have an affected parent
or an affected child \Rightarrow she is 100% heterozygous
- if the mother have an affected brother
 \Rightarrow she is 50% heterozygous.

- If the fetus was a boy he will obligatory receive Y from his father. The risk to receive X⁰ from his mother is $\frac{1}{2}$. So the risk is $\frac{1}{2}$ of boys.

4- In the sterile man X, the number of spermatocytes I is the same as in the fertile man (2 a.u.), but the number of spermatocytes II in the sterile man is much higher than that in the fertile man ($10 \text{ a.u} > 4 \text{ a.u}$). On the other hand, the number of spermatids or sperm cells in the sterile man is abnormally lower than that in the fertile man ($1 \text{ AU} < 8 \text{ AU}$). Therefore, not all spermatocytes II had divided into spermatids during meiosis. Hence, the cause of sterility in man X is an abnormal meiosis, which is blocked at the stage of spermatocytes II leading to an insufficient number of sperm cells (oligospermia).

5- Document 3 reveals one type of sperm cell that has a normal flagellum and a normal head, but the middle piece is larger than in the normal sperm cell. This is due to the non-elimination of residual cytoplasm (abnormal spermiogenesis).

Exercise 1 (5pts.)**Monohybrid dominant**

In drosophilae a pair of alleles determines the color of the eyes: the allele R (red eye color) is dominant over the allele w (white eye color).

1st cross:

If we cross two drosophilae of true breeding lines, a female with white-eyes and a male with red-eyes, we obtain in F1 all the offspring having red eyes.

- 1- Determine the genotypes of the parents and that of the offspring, by locating the alleles on their chromosomes. (1 ½ pts.)
- 2- Give the necessary analysis to verify the above result. (1 ½ pts.) (use a punnet square)

2nd cross:

The cross of the F1 drosophila among themselves gives in F2: 63 drosophilae, 47 drosophilae having red eyes and 16 drosophilae having white eyes.

- 3- Name the type of the mentioned above cross. (1/2 pt.)
 - 4- Find the phenotypic proportions of the result in F2. (1 ½ pts.)
-

Exercise 2 (5 ½ pts.)**Incomplete or intermediate dominance**

In order to study other traits in drosophilae we cross two true breeding lines of drosophila, one has long wings with another one has vestigial wings. All F1 generation gives drosophilae with medium wings.

- 1- Give an explanation of the result obtained from this cross. (2pts.)
 - 2- Identify the genotypes of the parents and that of F1. (1 ½ pt.)
 - 3- Give the necessary analysis to verify the above result. (2pts.) (use a Punnett square)
-

Exercise 3 (2 ½ pts.)**Lethal allele**

Consider a gene responsible for the length of the legs in drosophilae, where the long legs allele whose symbol "M" is dominant over the short legs allele whose symbol is "m". In general, there are no homozygous individuals with long legs.

We cross a heterozygous drosophila with a drosophila having short legs. We obtain ½ drosophila with long legs, ½ drosophilae with short legs.

- 1- Name the type of the mentioned above cross. Justify your answer. (1/2 pt.)

We cross two heterozygous drosophilae with long legs (F1xF1). We obtain surprising results.

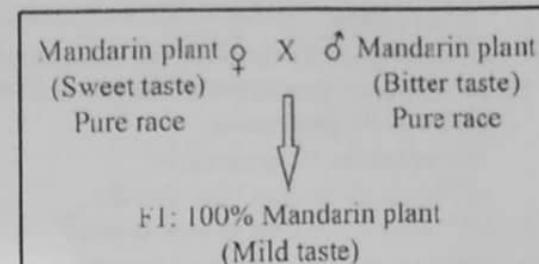
2/3 drosophilae with long legs, 1/3 drosophilae with short wings

- 2- Formulate a hypothesis explaining the surprising results. (1/2 pt.)
- 3- Make a factorial analysis to explain the results obtained. (1 ½ pts.)

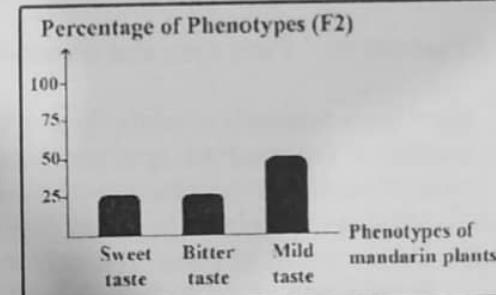
Exercise 1: Transmission of a Hereditary Trait in Mandarin Plants

To determine the type of inheritance of the gene responsible for the taste of mandarin fruits, a cross is performed between two varieties of mandarin plants that differ by one trait only. The cross and its results are represented in document 1.

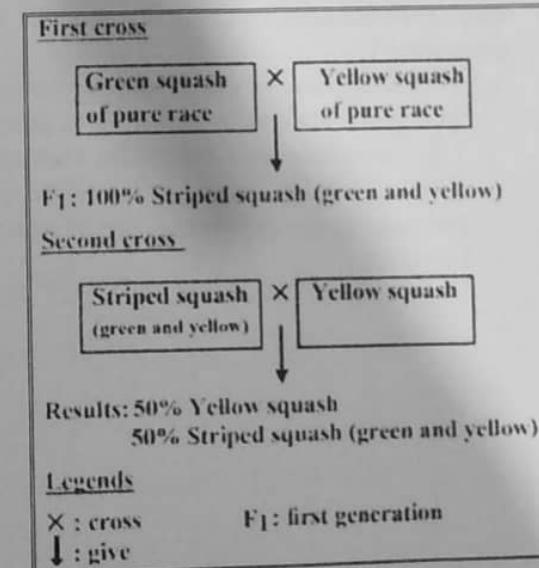
1. Specify the type of inheritance studied in mandarin plants.
2. Designate by symbols the corresponding alleles.
3. Write, by referring to document 1, the genotypes of each of the two parents and their descendants. The descendants of F1 generation are self-crossed (F1 x F1). The phenotypic results of the descendants of this cross (F2) are represented in document 2.
4. Make the necessary factorial analysis to verify the phenotypic results represented in document 2.
5. Verify if it is necessary to perform a test cross to determine the real genotypes of the descendants of the 2nd generation (F2).



Document 1



Document 2

**Exercise 2:**

The gene responsible for the color of squash is located on an autosome. To study the transmission of this gene, two crosses are performed whose results are shown in the adjacent document.

- 1- Describe the first cross using the given legends.
- 2- Is it a case of dominance or codominance? Justify the answer.
- 3- Designate by symbols the corresponding alleles.
- 4- Make the necessary factorial analysis to verify the results in the second cross.

Exercise 1:

1) (specify = answer then justify).

→ The type of inheritance studied is incomplete dominance (Intermediate) since the cross of 2 pure race of 2 different characters leads to descendants of intermediate phenotype (mild is between sweet and bitter).

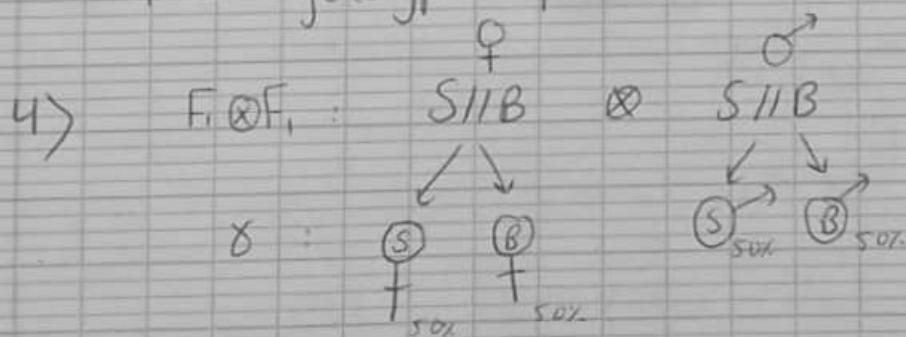
2) Let 'S' be the symbol of the allele coding for sweet taste.

Let 'B' be the symbol of the allele coding for bitter taste.

3) The genotype of the ♀ : S/S

The genotype of the ♂ : B/B

The genotype of the F₁ : S/B.



F ₂	♂	S	B
♀		50%	50%
S	S/S	B/S	
50%	25%	25%	
B	B/S	B/B	
50%	25%	25%	

⇒ The theoretical results confirm the experimental results.

The following document represents cells observed during oogenesis:

1-Identify the cells (and the phase at which the cell is) in figures 1,2,3,4 and 5.

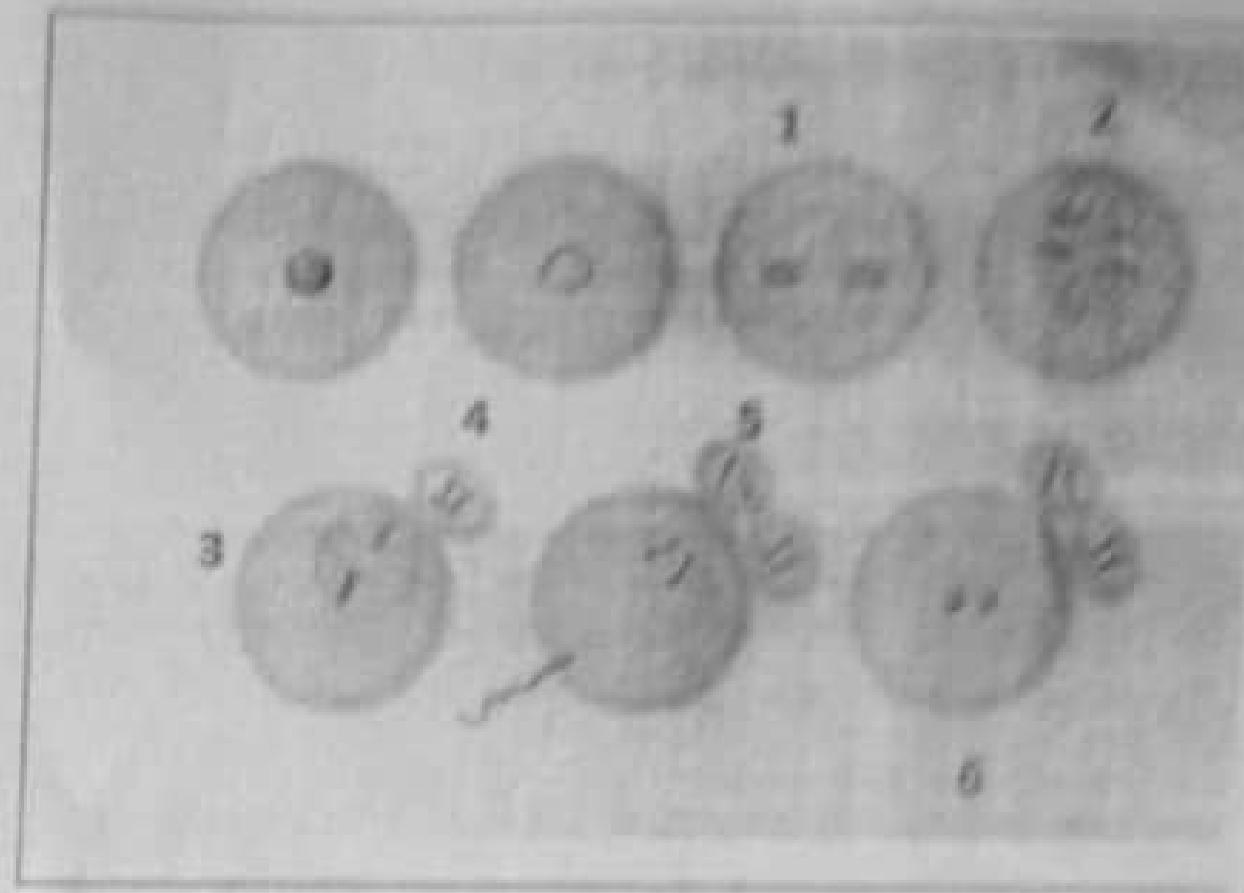
2-Specify the follicle that may contain cells 1,2 and 3.

The amount of DNA in cell 6 is 14.6au.

3-Indicate the amount of DNA in cell 5.

4-List the steps of oogenesis.

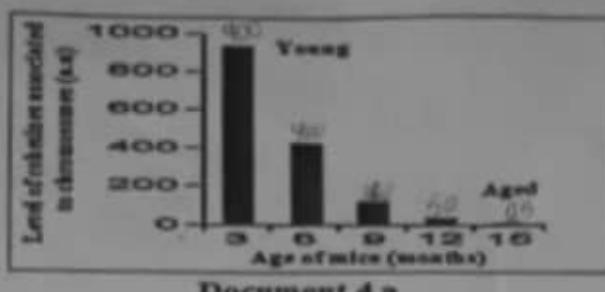
5-Represent by a drawing the phases of Oogenesis before birth ,(use only 4 chromosomes in your representation)



Document 4a shows the rate of cohesines according to the age of the mouse

The oocytes, during their formation, have the same amount of cohesines synthesized only once for their entire life.

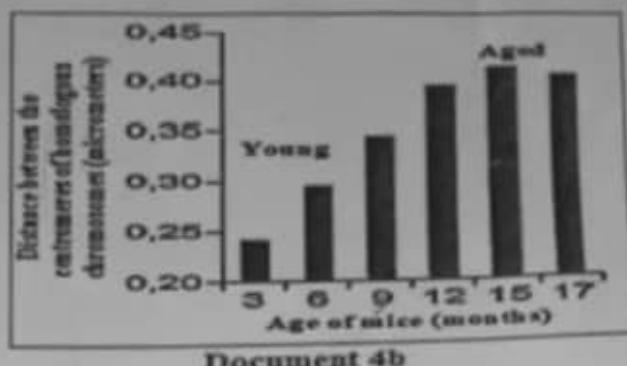
5. Interpret the results of document 4a. (1pt)



Document 4b represents the evolution of the distance between the centromeres in mice.

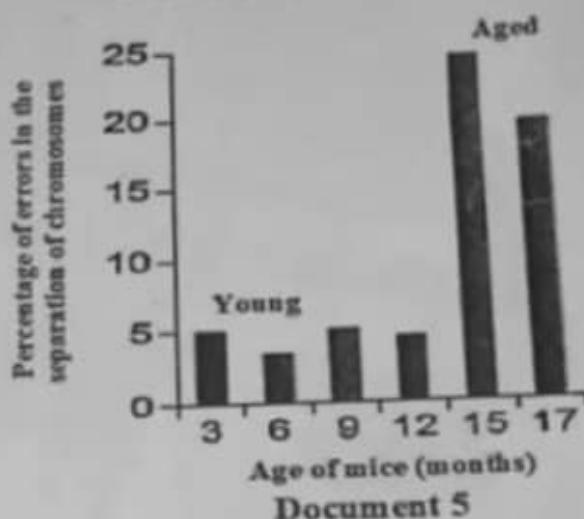
An increase in the distance between the centromeres of the homologous chromosomes increases the probability of their abnormal fixation on the spindle of division.

6. Derive from the results of the document 4b, the influence of age on the distance between the centromeres of the homologous chromosomes. (1pt)



Document 5 represents the percentage of error of separation of chromosomes in mice

7. From the information's issued from the documents and the acquired knowledge, show how meiosis can be at the origin of the increase of trisomy with age. (1½pts)



Mazen Ismail

d: Telophase I, cleavage producing 2 daughter cells each has a chr. of 2 chromatids.
e-Metaphase 2, single chromosomes line up to form the equatorial plate.

b: Anaphase 2, two sister chromatids separate to the opposite poles.

2- $n=5+X$ $n=5+Y$.

3-1. Fertilization.

3-2. 1-Sperm cell; 2-pedunculated follicular cell; 3-cytoplasm; 4-female
nucleus has chr.s arrested at meta-II; 5-oocyte II 6- 1st polar body 7-
cortical granules 8- male & female pronuclei 9- 2nd polar body 10- asters

11- First two cells of the embryo.

3-3.a-e-d-c-b

a-penetration of the sperm. e-Chr.s resume meiosis II d: formation of male &
female pronuclei c- Karyogamy b: Mitosis of the zygote.

1. The scheme X represents an oocyte II blocked at metaphase II after having released the first polar body. Once fertilized by a sperm cell this cell releases the content of its cortical granules forming the fertilization membrane and resumes meiosis II releasing the second polar body. The male and female pronuclei are formed.

2. The % of non-fertilized cells of scheme X is 100% while that of fertilized cells of scheme Y is 0% when the sperm cells are collected from the testicles; however as we proceed through the epididymis the percentage of scheme X decreases to reach 8% while the % of the scheme B increases to reach 92%. This shows that the testicles produce sperm cells with no fertilization capacity & that this fertilization capacity became more & more important as the sperm cells proceed through the epididymis. Therefore, the epididymis is the site of maturation (sperm cells acquire their fertilization capacity).

3. 3-1- Meiosis and fertilization.

3-2- Meiosis allows the reduction of the chromosomal number to obtain haploid cells. This is revealed in scheme a and/or b that show anaphase II and the separation of chromosomes into two haploid sets.

Fertilization restores the diploid state of the species. This is revealed in scheme c that shows the male and female pronuclei before their fusion.

4. Scheme b corresponds to step 3, since there is separation of the two haploid lots of chromosomes each with one chromatid. This corresponds to the second meiotic division where the DNA quantity is reduced from Q to Q/2.

Scheme c corresponds to step 4, since it shows the male and female pronuclei just before their fusion. The female pronucleus undergoes replication of its DNA resulting in an increase of its DNA quantity from Q/2 to Q.

Scheme d corresponds to step 6, since it shows the metaphase of the first mitotic division of the zygote having $2n$ chromosomes with 2 chromatids each and that corresponds to a DNA quantity of $2Q$.

Exercise 4 (7pts.)**monohybrid cross**

We have two true breeding lines of rats which differ by only one character, one constitutes the white color and the other the gray one.

- 1- Justify the following statement: "the purity of a race gives always same type of gametes" (1pt)

The crossing of the given gray male with the given white female gives in F1 only gray rats.

- 2- Specify the dominant and the recessive alleles. (2pts.)
- 3- Make the factorial analysis to find the theoretical phenotypic proportions in each of the following crosses:
 - i- F1 individuals with white rats. (1 ½ pt.)
 - ii- F1 individuals with heterozygous gray rats. (1 ½ pt.)
- 4- Identify the test (i or ii) that reflects the genotype of the parents. (1pt)

Exercise 5 (5pts.)**lethal alleles**

Given the following two crosses, each gives a different result:

$$1^{\text{st}} \text{ cross: } Aa \times Aa \longrightarrow 10 [A] + 5 [a]$$

$$2^{\text{nd}} \text{ cross: } Bb \times Bb \longrightarrow 15 [B]$$

- 1- Calculate the proportions of each result. (2pts.)
 - 2- What can you conclude? (1pt)
 - 3- Explain the result of each cross by using a Punnett square. (2pts)
-



Genetic Risk: Autosomal Recessive disease

G.Risk Probability for
the mother to
be heterozygous

X Probability for
father to be
heterozygous

X Probability
for the
couple to
give birth
to an affected
child

from Pedigree
(family history)

↓
from table
of-cross

if the mother or the father
have no family history:

take the Probability of population

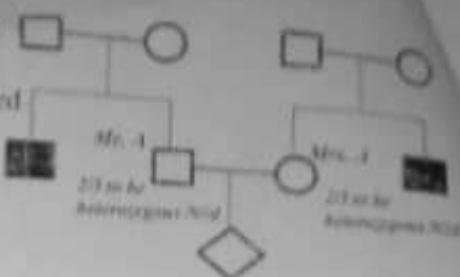
$$\frac{\% \text{ of-population}}{100}$$

3- The two parents are with family history of the disease.

Frequency of Mr. A to be heterozygote is $2/3$ since he has affected brother (his parents are heterozygotes)

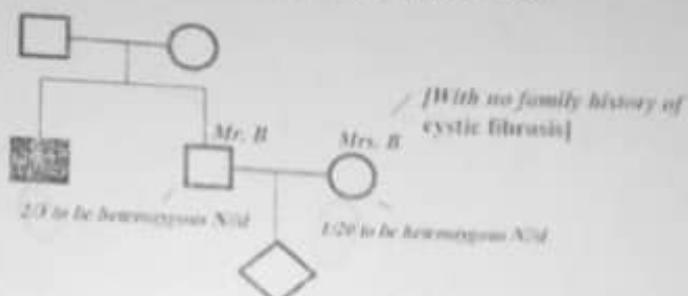
Frequency of Mrs. A to be heterozygote is $2/3$ since she has affected brother (her parents are heterozygotes)

Probability of having affected child is $1/4$



Therefore, the genetic risk for couple A to have a child with cystic fibrosis is: $2/3 \times 2/3 \times 1/4 = 1/9$

4- One of the parents is with family history and the other is not.



The frequency of Mr. B to be heterozygous is $2/3$ (he has an affected brother)

The frequency of Mrs. B to be heterozygous is $1/20$ since there is no family history of the cystic fibrosis;

The Probability of having affected child is $1/4$

Therefore, the genetic risk for couple B to have a child with cystic fibrosis is:
 $1/20 \times 2/3 \times 1/4 = 2/240 = 1/120$

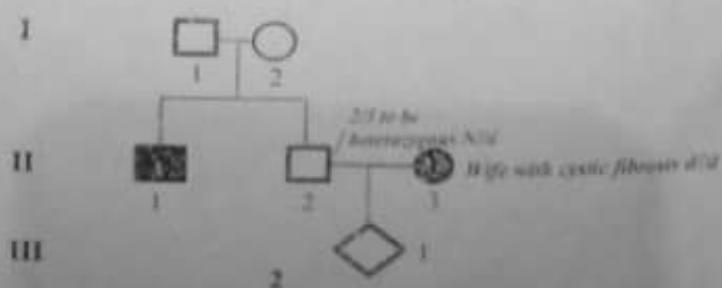
5- The husband is with cystic fibrosis and the wife is with no family history



The wife is with no family history of the cystic fibrosis, the frequency to be heterozygous is $1/20$. The husband is affected by cystic fibrosis, he is necessarily homozygote d/d and surely (probability is 1) gives his child the allele d. The phenotype of the fetus depends on whether he receives the allele N or the allele d from the probable heterozygous mother. This mother if she is heterozygous, there is a chance of $1/2$ to transmit the allele d to her child.

Therefore, the risk for this couple to have a child with cystic fibrosis is: $1/20 \times 1/2 = 1/40$.

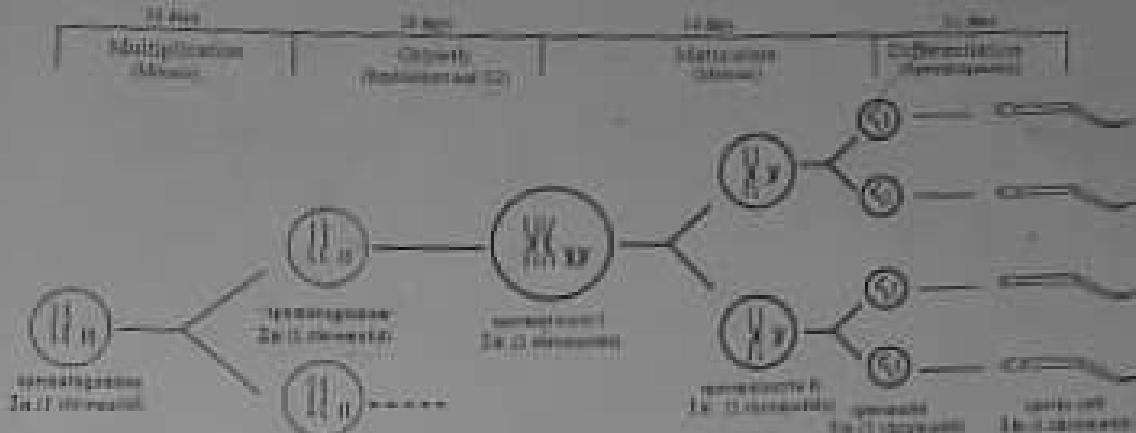
6- One of the parents is affected and the other with family history of the cystic fibrosis.



Ch-I-no.II-

- 1- c-a-d-e-b
- c: prophase I, since homologous pair form tetrads.
- a: Metaphase I, since homologous pair lines up to form the equatorial plate.

- Growth phase**: characterized by cellular development. Spermatogonia become spermatocytes I (2n chromosomes with 2 chromatids each).
- Maturation phase**: characterized by meiosis. Spermatocytes I → Spermatocytes II (n chromosomes with 2 chromatids each) → Spermatozoa (n chromosomes with 1 chromatid each).
- Differentiation phase of spermatogenesis**: characterized by the transformation of spermatids into mature spermatozoa which are finally released into the lumen of the seminiferous tubules.



Spermatogenesis

- Stalk cells are found in the seminiferous tubules between the reproductive cells. Their role is to sustain and nourish these cells.

Documents 5: Oogenesis (P27-29)

- The ovary contains many follicles at different developmental stages. Each follicle is made up of a single germ cell surrounded by epithelial (follicular) cells. (Doc. a)
- Oogenesis is the formation of the female gametes, the ova. It occurs in the peripheral zone of the ovaries. Oogenesis starts during the fetal life of the mammalian female and continues till menopause in a discontinuous manner.
- Oogenesis includes 4 phases (Doc. d):
 - Multiplication phase**: during embryonic life, this phase is characterized by successive mitotic divisions of the germ stem cells, the oogonia (2n chromosomes with 1 chromatid each) to give a total number of 700 million oogonia.
 - Growth phase**: during embryonic life, this phase is characterized by the transformation of oogonia into oocytes I (2n chromosomes with 2 chromatids each). The oocyte I is surrounded by few flattened follicular cells to form the primordial follicle. A large number of follicles degenerate during the fetal life (follicular atresia). Oocytes I in the remaining follicles remain blocked at prophase I until puberty.
 - Maturation phase**: this phase starts during embryonic life and resumes few hours before ovulation. Follicular degeneration (atresia) continues during childhood so that, at puberty, the ovaries will contain only 400000 primordial follicles.

4 months before each cycle, about 100 primordial follicles resume their growth activity but only one of these follicles will reach maturity. All the others will degenerate.

Few hours before ovulation, oocyte I divides by reductional division giving rise to one oocyte II and first polar body. Oocytes II enter equational division and blocks at metaphase II. It continues

Exercise 2 (5 points)

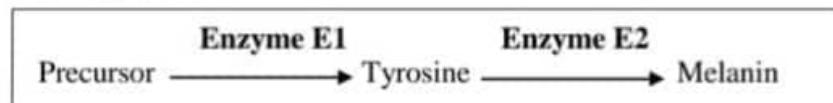
Albinism is a hereditary autosomal and recessive disease caused by the absence of melanin, a pigment responsible for hair color. Document 1 represents the pedigree of a family where some members are affected by the disease.

- Indicate the genotypes of individuals I-1, II-1, and III-6.

In the population where this family lives, among each 100 normal individuals 15 are heterozygous.

- Determine the genetic risk for the fetus IV-3 to be albino.

Researchers have identified gene E1 coding for enzyme E1 and gene E2 coding for enzyme E2. These enzymes are essential for the synthesis of melanin according to the following reactions:



To find the exact origin of albinism in this family, the researchers determined the nucleotide sequences of specific parts of the non-transcribed DNA strand of gene E1 (document 2a) and gene E2 (document 2b) for a normal individual and an albino individual of this family.

Non-transcribed DNA strand of gene E1									
	1	2	3	4	5	6	7	8	9
Normal individualACG	AGG	CCT	ACG	GGC	TTA	TGG	GGC	GAA...
Albino individualACG	AGG	CCT	ACG	GGC	TTA	TGG	GGC	GAA...

Document 2a

Non-transcribed DNA strand of gene E2									
	1	2	3	4	5	6	7	8	9
Normal individual	...ATC	ATG	CGA	ACC	GGC	TGC	TCA	AAC	CCA...
Albino individual	...ATC	ATG	CGA	ACC	GGC	TGC	TGA	AAC	CCA...

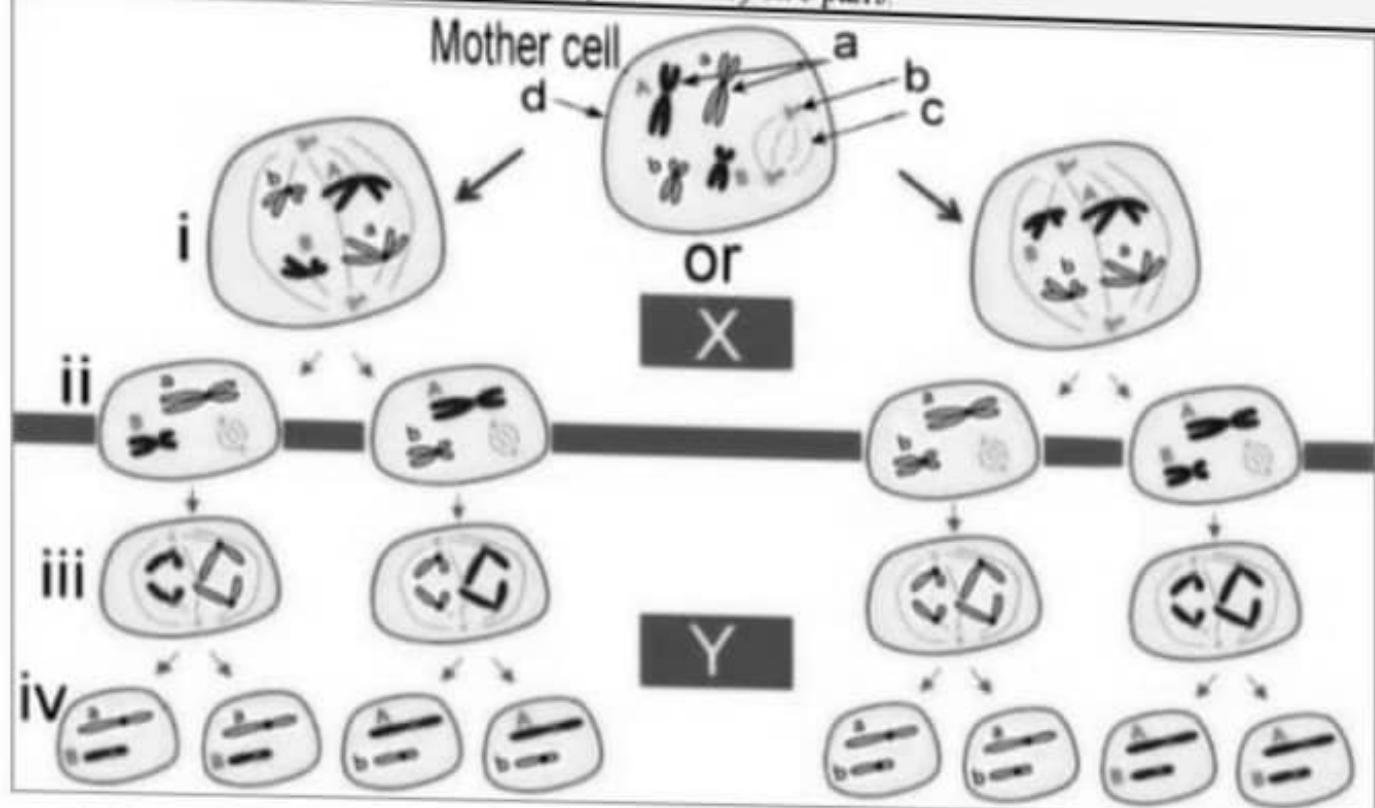
Document 2b

		Second letter				Third letter
		U	C	A	G	
First letter	U	UUU Phe UUC UUA UUG	UCU Ser UCC UCA UCG	UAU Tyr UAC UAA STOP UAG STOP	UGU Cys UGC UGA STOP UGG Trp	U C A G
	C	CUU Leu CUC CUA CUG	CCU Pro CCC CCA CCG	CAU His CAC CAA Gln CAG	CGU Arg CGC CGA CGG	U C A G
	A	AUU Ile AUC AUA AUG Met	ACU Thr ACC ACA ACG	AAU Asn AAC AAA Lys AAG	AGU Ser AGC AGA Arg AGG	U C A G
	G	GUU Val GUC GUA GUG	GCU Ala GCC GCA GCG	GAU Asp GAC GAA Glu GAG	GGU Gly GGC GGA GGG	U C A G

- Show that the gene responsible for albinism in this family is gene E2.
- Determine, using the genetic code (document 3), the amino acid sequence of enzyme E2 that corresponds to each of the two individuals, the normal and the albino.
- Explain how the modification in the nucleotide sequence of the allele coding for enzyme E2 leads to albinism in this family.

Document 3

Meiosis process takes place in both male & female gonads to ensure haploid gametes for fertilization otherwise, the fertilization is not successful. For this reason, consider these schemes that represent this division in a human cell, for simplicity we represent only two pairs.



- a- Pick out the name of the represented division.
- b- Pick out the name of the phenomenon that follows this division.
- c- Indicate the objective of such division.
- d- Using your acquired knowledge, name each of the male & female gametes.
- e- Explain the term "haploid".
- f- Label the mother cell.
- g- Compare the number of chromosomes between the mother cell & cell ii.
- h- Conclude the name & importance of division X.
- i- Name the phases of cell i & cell iii, then compare them.
- j- Cell ii & iv have the same number of chromosomes. Is it true? Conclude the name of division Y.
- k- Compare the initial cell to that of gamete iv.
- l- Conclude concerning the genetic information.
- m- Indicate the number of the possible types of the produced gametes.

Question -11:

The document below represents some important phases of meiosis that are observed during spermatogenesis.

1. Classify these different phases of meiosis in chronological order. Justify your answer.

A spermatocyte ($2n=6$) undergoes meiosis.

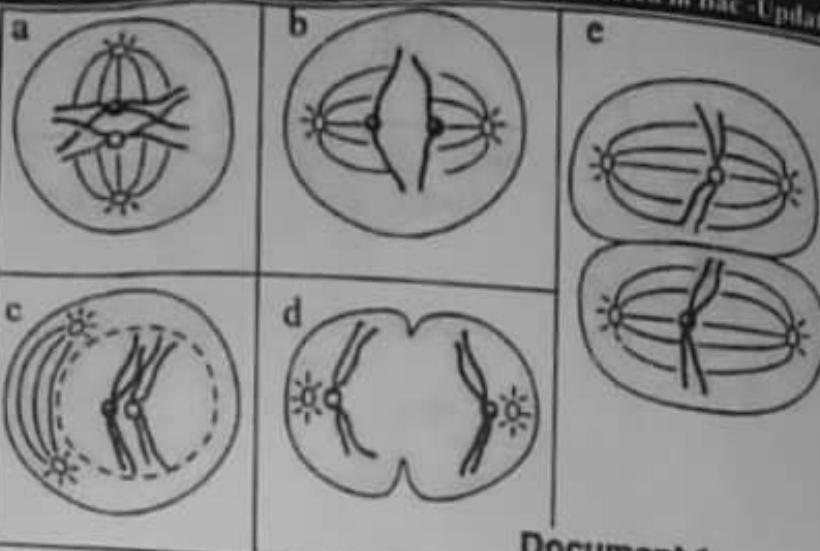
2. Indicate the different possible chromosomal formulas of spermatids obtained in human species.

The following document shows certain stages of a biological phenomenon in human reproduction.

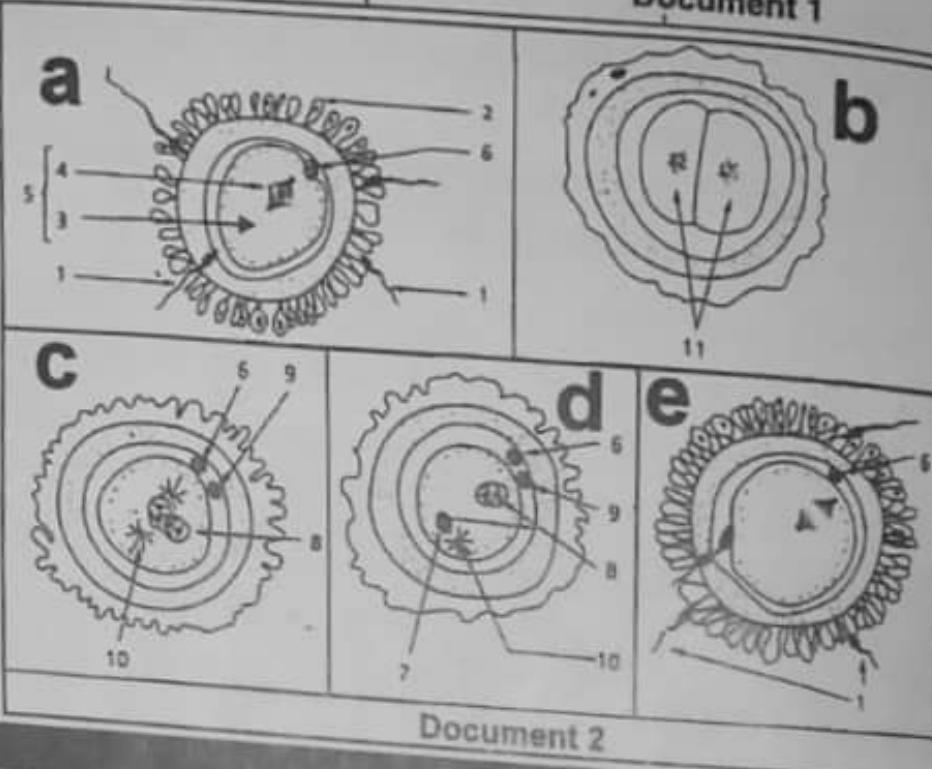
- 3-1. Name this phenomenon.

- 3-2. Label structures 1 to 11 in document 2.

- 3-3. Give the chronological order of the different stages of this phenomenon. Justify.



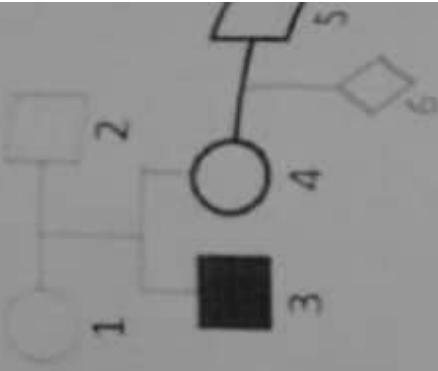
Document 1

**Question -12:**

During follicular development, oocytes increase in size due to:

Case 3:

- Mother ($X^N X^N$) or ($X^N X^d$). Thus, the risk for mother 4 to be hybrid is $\frac{1}{2}$ since she has hybrid mother (1) who gave a diseased boy. The father is healthy of genotype $X^N Y$.
 - If the fetus is a girl, she will obligatory receive X^N from her father and normal allele is dominant regardless of gamete received from mother. So, the risk to be affected is null
 - If the fetus is a boy, then he will obligatory receive Y from his father. The **risk for his mother to hybrid is $\frac{1}{2}$** and the **risk for this hybrid mother to give X^d is $\frac{1}{2}$** . So the risk to have affected boy = $\frac{1}{2} \times \frac{1}{2} = \frac{1}{4}$ of boys.



Case 4:

If the risk to be hybrid in population is $1/8$.



- The father is normal of genotype $X^N Y$ and the mother is healthy and the risk to be hybrid is $1/8$ since no family history is revealed.
 - If the fetus is a girl, she will obligatory receive X^N from her father and normal allele is dominant regardless of gamete received from mother. So the risk to be affected is null
 - If the fetus is a boy, he will obligatory receive Y from his father. The **risk for his mother to hybrid is $1/8$ (no family history)** and the **risk for this hybrid mother to give X^d is $\frac{1}{2}$** . So, the risk to have affected boy is $1/8 \times \frac{1}{2} = 1/16$ of boys.

Exercise 11 An enzymatic deficiency

- 1.1. It is linked to deficiency of aldolase B. This enzyme, located in the liver, small intestine and kidneys, cleaves the fructose-1-phosphate to DHAP and glyceraldehyde in order to allow further glycolysis for the
- 1.2. This results in an accumulation of fructose-1-phosphate in the liver and kidneys. Since this molecule cannot follow another metabolic pathway, it becomes toxic to these organs.
- 2.1. Since the couple III (3, 4) is normal and gave birth to affected children (boys 1 and 7 and a girl 3: Generation IV) then the allele that determines the disease was masked or hidden in at least one of the parents. Thus, it is recessive.
 N : normal allele a:Abnormal allele
- 2.2. The allele responsible for this disease is linked on an autosome because:
 If the gene that determines the disease is bound to the proper part of Y every affected boy must have an affected father but the boy 7 (IV) is affected but his father 3 (III) is normal then this is not the case.
 If the gene that determines the disease is bound to the proper part of X then the affected daughter 3 (IV) should be of genotype X^aX^a , must receive X^a from her mother and X^a from her father 3(III), who will be affected (X^aY), but he is normal.
 If the gene that determines the disease is bound to the common part of X and Y, the affected daughter 3 (IV) of genotype X^aX^a must receive X^a from her mother and X^a from her father and the affected boy 1 (IV) X^aY^a , should receive Y^a from his father who will then of genotype X^aY^a (affected) but he is normal.
- 3.1. mRNA (normal sequence): AAU GGA CUG GUA CCU AUU GUU GAA.....
 Peptide Sequence: asn- gly- leu- val- pro -Ile -val -glu.....
 mRNA (abnormal sequence): AAU GGA CCU AUU GUU GAA
 Peptide Sequence: asn- gly- pro -Ile- val -glu.....
- 3.2. A mutation by deletion modified non-functional protein
- 4 The probability of the individual 9 (III) being heterozygous (no family history) is equal to that of the population: 1/5000. The mother is sick and the probability of this couple having a sick child is $\frac{1}{2}$ (fusion of parental gametes that would give birth to an affected child) then the risk = $1/5000 \times 1/2 = 1/10000$. The genetic risk for his cousins to be affected is 1/4 (parents III 3-4 are heterozygous). This big difference is explained by the fact that consanguineous marriage (parents III 3-4 are close relatives) increases the risk of genetic diseases.

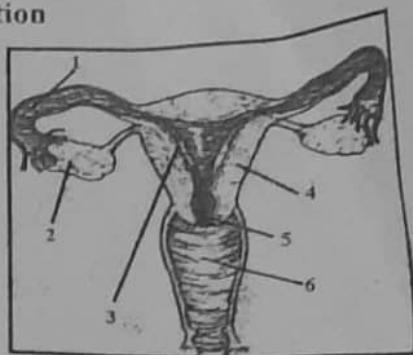
Exercise 1 (10 points)**Mechanisms of Sexual Reproduction**

Reproduction is the process that ensures the survival of the species. Sexual reproduction involves two individuals of opposite sexes and belonging to the same species.

1. Name the two principal mechanisms of sexual reproduction in mammals and state the importance of each.

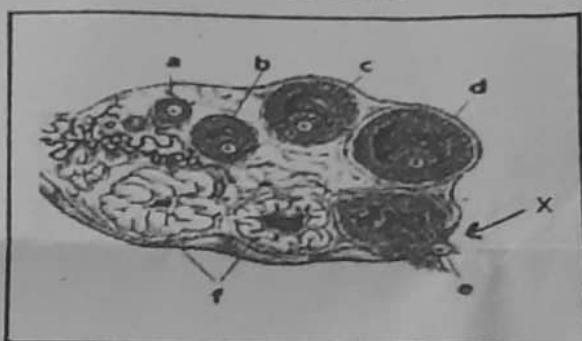
Sexual reproduction necessitates the presence of specialized and organized reproductive systems. Document 1 represents the human female reproductive system.

- 2.1. Annotate document 1.
- 2.2. Indicate the role of organs 1 and 2.



Document 2 represents a schematic section of an organ in the female reproductive system.

- 3.1. Name this organ.
- 3.2. Annotate the structures (a) to (f) and phenomenon X.
- 3.3. Indicate the phase of the sexual cycle during which structures (a) to (d) of document 2 can be seen.
- 3.4. State the importance of phenomenon X.
- 3.5. Explain the fate of structures (e) and (f) during the sexual cycle.



Document 3 reveals a photograph accompanied by an interpretation diagram of the female sex cell during oogenesis.

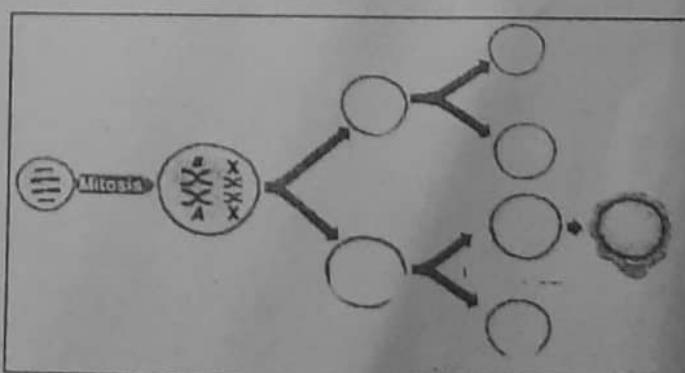
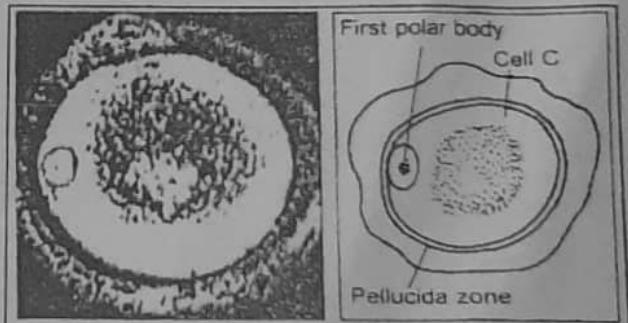
4. Verify that this sex cell is an oocyte II and not an oocyte I.
5. Describe the karyotype of this oocyte.

Document 4 shows the process of oogenesis of a mother cell with $2n=4$.

6. Reproduce this diagram and complete it with all the details of the process.

Note that your diagram should include the chromosomal

behaviour with the names of the cells and the phases of oogenesis.

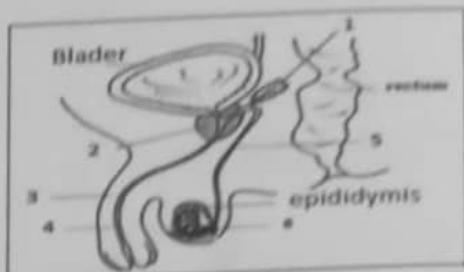


Answer the following exercises:

Male sterility

Exercise 1: (6 pts) **Male Reproductive System**
Diagram 1 shows the organs of the male reproductive system.

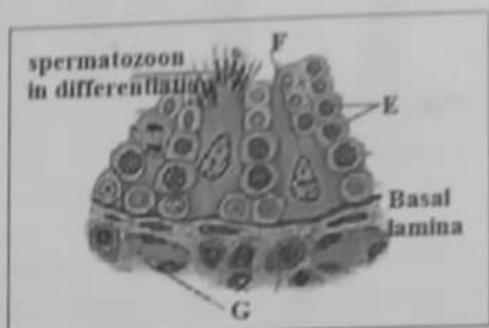
- 1. Label the figure of document 1.**



Document 1

Document 2 shows a cross-section of a part of a structure of a fertile man.

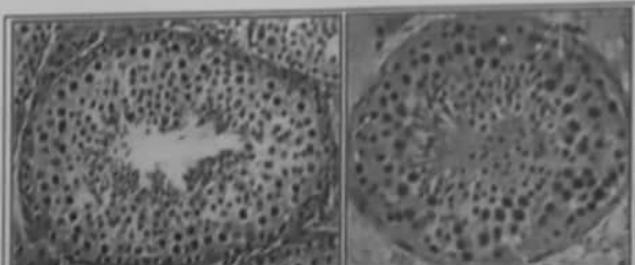
- Identify the cells E, F and G of this structure.
 - Explain, referring to your acquired knowledge, the variation in cell size as it passes towards the center of the structure shown in document 2.



Document 2

Microscopic observation was performed for a seminiferous tubule of a normal man and another sterile. The obtained microphotographs are shown in document 3 (a and b) respectively.

4. Compare the two seminiferous tubules
 5. Formulate a hypothesis that explains the sterility of this man.



Document 3

b

Exercise 2: (4.5 points)

Testosterone and Spermatogenesis

The testes continuously produce sperm and the male hormone 'testosterone' from puberty until death. Testosterone is essential for spermatogenesis and the maintenance of primary and secondary sexual characteristics.

Document 1 shows the variation in the number of sperm in the testes as a function of the concentration of testosterone in the fluid of the seminiferous tubules.

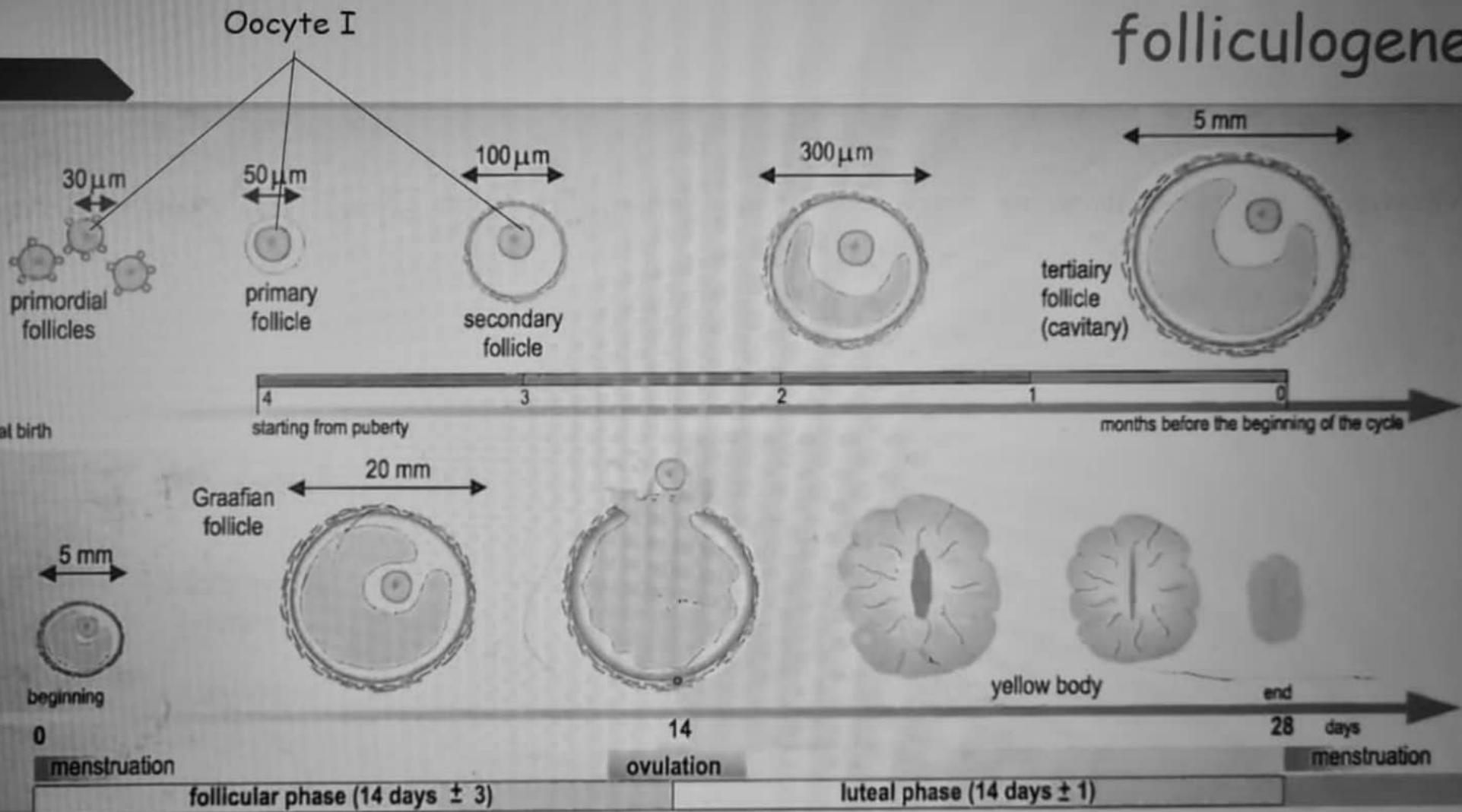
Testosterone concentration (ng/mL)	5	10	15	20
Number of spermatids per testis x 10 ⁶	40	150	210	250

Document 1

1. Doc 1 → Graph

2. Interpret Doc.

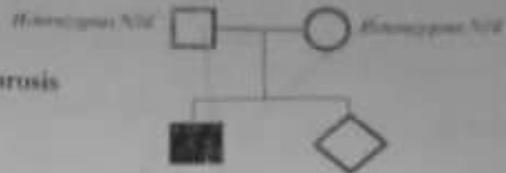
folliculogenesis



© Dr. Jan

***Determining the genetic risk for a couple to have a child affected by autosomal disease**

II- Autosomal recessive disease: Cystic fibrosis



***Genetic risk = Frequency of father to be heterozygous \times Frequency of mother to be heterozygous \times probability of having affected child.**

1- The couple already has an affected child.

The couple has already an affected child, the two parents are surely heterozygotes N/d.

Frequency of father to be heterozygous is 1

Frequency of mother to be heterozygous is 1

Probability of having affected child is $\frac{1}{2} \times \frac{1}{2} = \frac{1}{4}$ (probability to receive two alleles d, one allele d from each parent)

$$\text{Genetic risk} = 1 \times 1 \times \frac{1}{4} = 1/4$$

Male Female	% N	% d
$\frac{1}{2} N$	$\frac{1}{4} N/N$	$\frac{1}{4} N/d$
$\frac{1}{2} d$	$\frac{1}{4} N/d$	$\frac{1}{4} d/d$ [Sick]

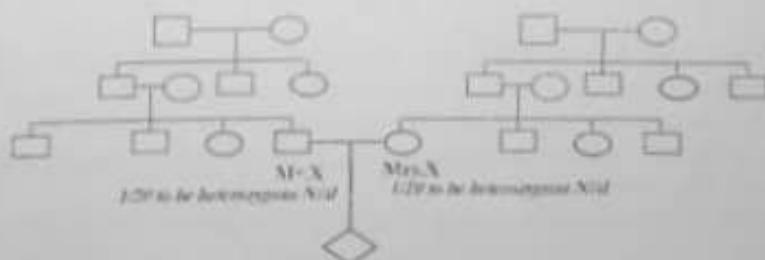
Don't use Punnett square if you are asked for determining the risk.

It's introduced here for explanation only.

2-Both parents are without family history of the disease.

*** In France, heterozygotes form 5% (=1/20) of French population.**

- A study was done in French population: in 800 persons, 40 persons of them are found to be heterozygous for cystic fibrosis. (Frequency of French man or woman to be heterozygous is $40/800 = 1/20$).



Since each of the two parents has no family history of cystic fibrosis, the frequency of each of Mr.X and Mrs.X to be heterozygous is $1/20$ (frequency in French population)

The Probability of having affected child is $1/4$

Therefore, the genetic risk for this couple to have a child with cystic fibrosis is $1/20 \times 1/20 \times 1/4 = 1/1600$

Case 2

Specify if the allele responsible for the disease is dominant or recessive.

The allele responsible for the disease is dominant with respect to normal allele. Since The affected couple, I1 and I2 had normal children II3 and II4. This means that both affected parents carry the normal allele. Thus, the allele of the disease masks the normal allele.

Determine the risk of the fetus II-6 to be affected. By logical reasoning.

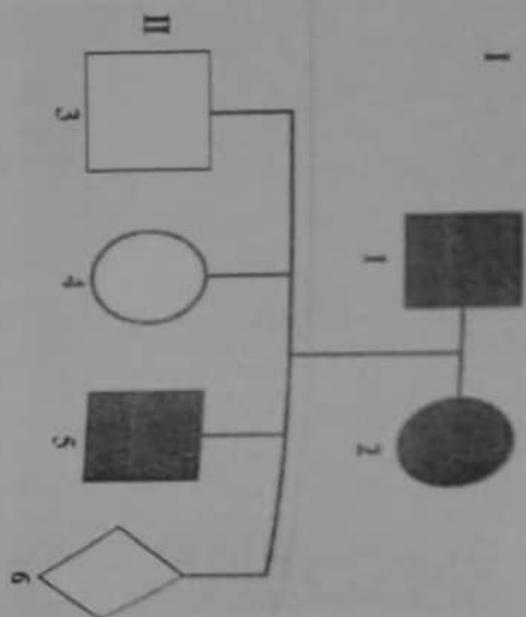
First step:

Probability of fetus to be normal= probability of mother to be hybrid x probability of father to be hybrid x probability of fetus to take normal allele from each parent.

- Probability of mother to be hybrid = probability of father to be hybrid = 1 (since the two parents are affected having normal child with genotype n/n that must take the normal allele from each parent)
- Probability of child to take the normal allele from each parent= $\frac{1}{2} \times \frac{1}{2} = \frac{1}{4}$
- ($\frac{1}{2}$ to take normal allele from mother since $\frac{1}{2}$ to take normal allele and $\frac{1}{2}$ to take diseased allele)
- ($\frac{1}{2}$ to take normal allele from father since $\frac{1}{2}$ to take normal allele and $\frac{1}{2}$ to take diseased allele)
- Probability of fetus to be normal= $1 \times 1 \times \frac{1}{4} = \frac{1}{4}$

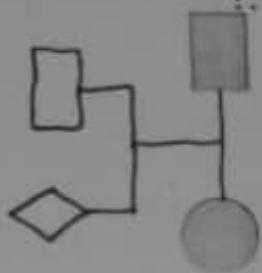
Second Step:

Risk of fetus to be affected = $1 - \text{Probability of fetus to be normal} = 1 - \frac{1}{4} = \frac{3}{4}$



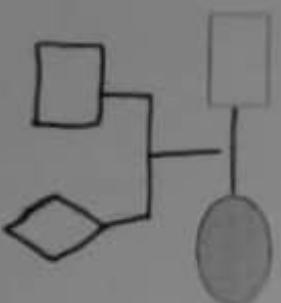
B. Dominant sex linked diseases (Gene carried on non- homologous part of X).

Case 1:



- The genotype of the father is $X^D Y$ since he is affected by the disease. The mother is hybrid of genotype $X^D X^d$ since she is affected by dominant disease and has normal boy who received X^d from his mother.
- If the fetus is a girl then she will obligatory affected since she will always receive X^D from her father and the allele of disease is dominant.
So, risk = 1 (all girls)
- If the fetus is a boy he will obligatory receive Y from his father. The risk to receive X^D from his mother is $\frac{1}{2}$ since his mother is hybrid and then the risk to be affected is $\frac{1}{2}$ of boys.

Case 2:



- The father is healthy of genotype $X^n Y$ and the mother is hybrid of genotype $X^D X^d$ since she is affected by dominant disease and has healthy son.
- If the fetus was a girl then she will obligatory receive X^d from her father. The risk to receive X^D from her mother is $\frac{1}{2}$. Then the risk is $\frac{1}{2}$ of girls.

Question-14-**Reproductive function of the testicles.**

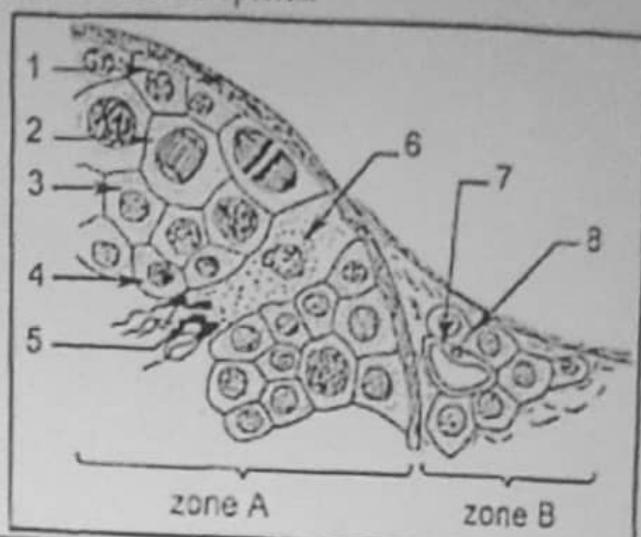
A study is done on certain aspects of reproductive functions in human species.

Document-1 represents a simplified cross section in a testis of a human male. Doc-1.

a. What do the zones A and B doc-1 refer to?

b. Annotate the given figure.

To confirm the role of the testicles in reproductive function, experiments were realized on two lots of mice: lot-1 and lot-2. The experiments and their results are summarized in Document-2



	Experiments	Results
Lot-1	Destruction by X-rays the cells of zone-B in doc-1.	-Regression of the secondary sexual characteristics. -Sterility.
Lot-2	Destruction by X-rays the cells of zone-A in doc-1	-Maintenance of the secondary sexual characteristics -Sterility.

c. Interpret the experiments done.

d. Based on your acquired knowledge, explain the role of zone-A and that of zone-B.

f. Deduce two possible causes of sterility.

Exercise 2 (8.5 pts)

1. Label the cells indicated by numbers 1 to 5

2. what is the number of chromosomes in each cell?

Exercise 1: (1.5 points)

Circle the correct answer(s) in each of the following items:

1. Many events occur during meiosis including:

- a. The separation of sister chromatids during anaphase I leads to a reduction in the number of chromosomes in the daughter cells.
- b. At the end of meiosis I, each chromosome is made up of 1 chromatid, while at the end of meiosis II, chromosomes are double.
- c. At the end of meiosis II, the quantity of DNA decreases from Q to Q/2 while the number of chromosomes is conserved.
- d. During prophase I, the formation of tetrads followed by the exchange of genetic information between sister chromatids, favors the diversity of gametes.

2. In spermatogenesis, the phases of maturation involve

- a. formation of spermatids from primary spermatocytes through meiosis
- b. Growth of spermatogonia into primary spermatocytes
- c. formation of spermatogonia from germ cells through mitosis
- d. formation of oogonia from spermatocytes through meiosis

3. During meiosis:

- a. there is reduction of the number of chromosomes but not the quantity of DNA
- b. the first division is equational
- c. the first division is reductional
- d. there is duplication of DNA just before prophase 2

Exercise 3

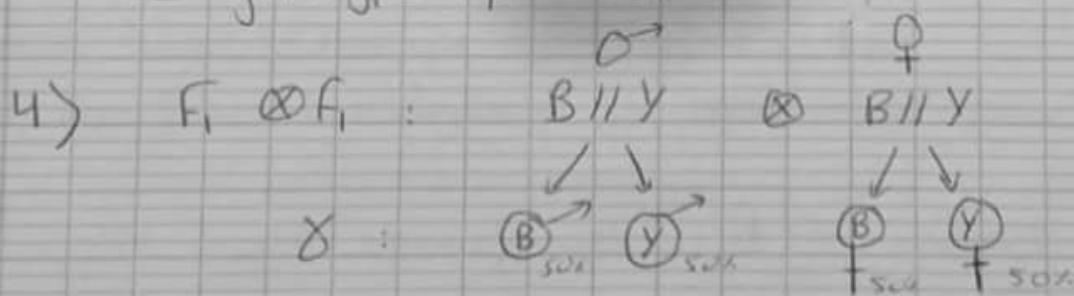
1) The descendants of the first generation inherit the yellow flower allele from one pure parent and the blue flower allele from the second pure parent and they express a third color (violet) which means that the alleles of the genes that determines the color of the flowers are incompletely dominant.

2) Let "y" be the symbol of the allele coding for the yellow color of the flower.

Let "B" be the symbol of the allele coding for the blue color of the flower.

3) The genotype of the parents are : $B\text{II}B$ & $Y\text{I}y$

The genotype of : F_1 is $B\text{II}\cdot Y$



F_2 :

♂	B	Y	phenotype:
♀	50%	50%	$[BY] : 50\%$
B	$B\text{II}B$	$B\text{II}Y$	$[B] : 25\%$
50%	25%	25%	$[Y] : 25\%$
y	$B\text{II}Y$	$Y\text{I}Y$	the theoretical results confirmed the experimental results.
50%	25%	25%	

Document 3- Sex-linked diseases

Now to calculate the genetic risk in case of Sex-linked diseases (gene carried on non-homologous part of X)

In case of gene carried on non-homologous part of X, we have to find the risk of girls to be affected and the risk of boys to be affected using logical reasoning.

Determine the risk for the fetus to be affected in the following cases:

A- Recessive sex-linked diseases and Gene carried on non-homologous part of X.

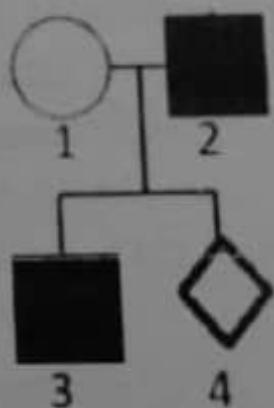
Case 1

- The fetus has a heterozygous mother of genotype $X^N X^d$ since she has an affected boy whom receives X^d from his mother and normal father of genotype $X^N Y$.
- If the fetus was a girl, she will obligatory receive X^N from her healthy father regardless of gamete received from mother (N is dominant). So, the risk will be null (zero)
- If the fetus was a boy, he will obligatory receive Y chromosome from his father. The risk of fetus to receive chromosome X^d from his mother is $\frac{1}{2}$. So, the risk to have affected boy is $\frac{1}{2}$.



Case 2:

- The fetus has a diseased father of genotype $X^d Y$ and obligatory hybrid mother of genotype $X^N X^d$ since the mother gave birth to affected boy.
- If the fetus were a female, she will obligatory receive X^d from father. The risk to receive X^d from her mother is $\frac{1}{2}$. So, the risk is $\frac{1}{2}$ of girls.
- If the fetus is a boy, he will obligatory receive Y from his father. The risk to receive X^d from his mother is $\frac{1}{2}$. So, the risk is $\frac{1}{2}$ of boys.



- 1.1-TP53 which plays a role in transcription and therefore intervenes in multiple important cellular functions such the regulation of the cell cycle and apoptosis (programed cell death)
- 1.2- The inactivation or this gene is marked in 50% of cases of sporadic cancer (no family history) and the inactivation results rather in an abnormal and rapid multiplication of cells and the development of a tumor mass
- 1.3- exposure to ultraviolet rays (UV studies rays), smoking and chronic alcoholic consumption. (1.5)

2- The sequences of nucleotides in both individuals are the same except in the third nucleotide in codon 5334 where G in normal individual is replaced by T in the affected one. then, this is a point mutation by substitution.

(1)

3- Normal DNA: CCT TCA GTC AGG AAA
 Normal mRNA: CCU UCA GUC AGG AAA
 Normal peptide: pro-ser-val-arg-lys

Normal DNA: CCT TCA GTC AGT AAA
 Normal mRNA: CCU UCA GUC AGU AAA
 Normal peptide: pro-ser-val-ser-lys

mRNA is the same as DNA but T is replaced by U since DNA is non-transcribed. (2)

4- The mutation by substitution in the third nucleotide in codon 5334 is transcribed at the level of mRNA by a new codon which is AGU. This is translated at the level of the peptide by a new amino acid (ser instead of arg). The new amino acid sequence leads to the modification of the three-dimensional structure of the protein TP53 which plays a role in the regulation of the cell cycle and apoptosis. The inactivation of TP53 leads rapid multiplication of cells and the development of a tumor mass. (1.5)

5- The variation of the AFP level in blood and the mass of the liver in two individuals a normal and an affected one. (1.5)

Individuals	Individual 1	Individual 2
AFP level in blood (ng/ml)	7	500
Mass of the liver (g)	800	1300

6- the AFP in individual 1 is 7 which is between the normal range (5 and 10 ng/ml) this shows that individual one is normal. However, the AFP level in individual 2 is 500 which is greater than the normal range (5 and 10 ng/ml). therefore, individual 2 is affected. (1)

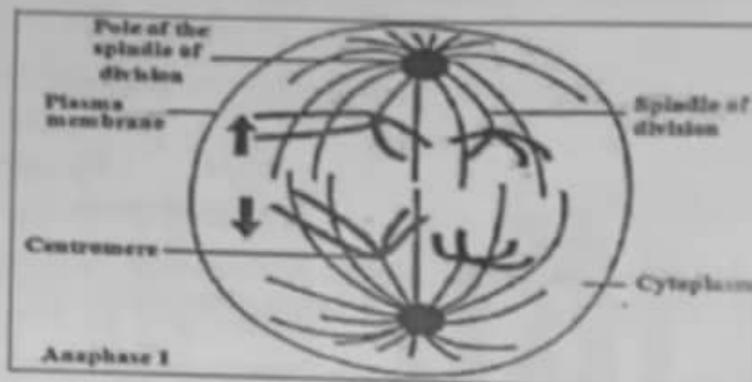
Exercise II: (7½pts)

Meiosis and Trisomy

In order to study the influence of meiosis on the increasing of trisomy's with age, several analysis were made with the results found in the documents below.

- What is the studied problem? (5pt)

Document 1 describes the behavior of spindle of division. The spindle of division appears during prophase of each of the two divisions of meiosis. Its shortening allows the migration to the poles of the chromosomes in anaphase I and the chromatids in anaphase II.



Document 1

Document 2 represents the frequency of chromosomal abnormalities during gametes formation in a father and a mother that will lead to Down's syndrome (trisomy 21).

	Father	Mother
Anaphase I	5 %	70 %
Anaphase II	5 %	20 %
Total	10 %	90 %

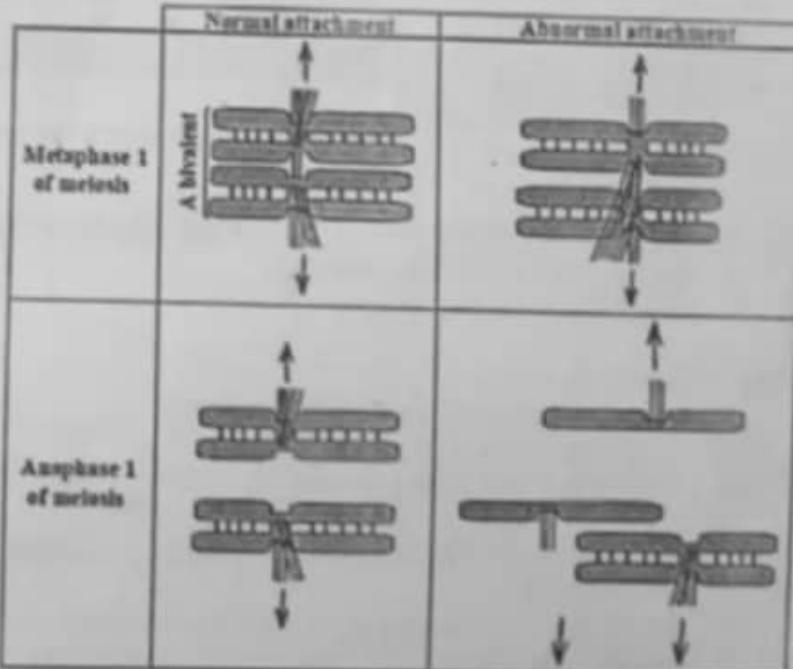
Document 2

- What information can you draw out from the results of document 2? Justify. (1½pts)

During meiosis, the cohesion between the homologous chromosomes is provided by proteins called cohesines. During metaphase I of an abnormal meiosis, each chromosome can be attached by the spindle fiber to the 2 opposite poles of the cell.

Document 3 shows normal & abnormal attachment of the chromosomes to the spindle of division in the first division of meiosis.

- By referring to document 3, compare the separation of chromosomes in normal and abnormal attachment during anaphase I of meiosis. (1pt)
- Formulate a hypothesis, concerning the cause of the abnormal separation of chromosomes during anaphase I. (1pt)



Document 3 \equiv Fibers of the spindle of division \equiv Cohesines

The frequency of father II2 to be heterozygous is $\frac{2}{3}$ (has affected brother).

Mother II3 is affected by cystic fibrosis, she is necessarily homozygote d/d and surely transmits to her child the allele d . The phenotype of the fetus depends on whether he receives the allele N or the allele d from the probable heterozygous father II2. This father II2 if he is heterozygous, there is a chance of $\frac{1}{2}$ to transmit the allele d to his child.

Therefore, the genetic risk for this couple to have a child with cystic is $\frac{2}{3} \times \frac{1}{2} = \frac{1}{3}$

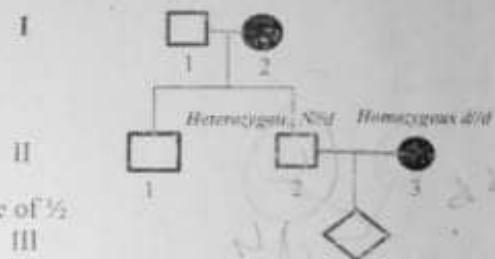
7

- Both parents are affected.



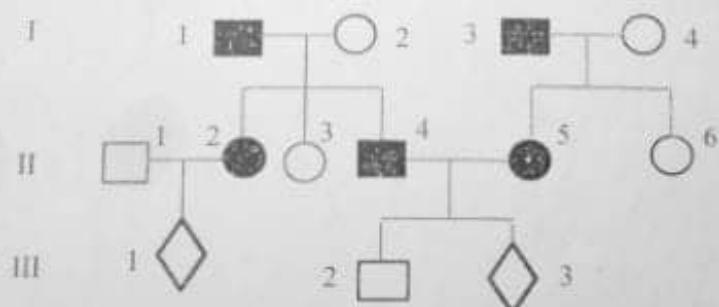
The parents are affected with cystic fibrosis which is recessive. The fetus is necessarily homozygous d/d , he would receive 2 mutant alleles d , one from each affected homozygous parent. *Then there is 100% risk for the fetus to be affected*

8- Individual II2 is heterozygous N/d since he is healthy and surely inherited the allele of disease d from his homozygous affected mother I2. Woman II3 is affected by cystic fibrosis, she is necessarily homozygote d/d and surely gives her child the allele d . The phenotype of the fetus depends on whether he receives the allele N or the allele d from the probable heterozygous father II2. There is a chance of $\frac{1}{2}$ for father II2 to transmit the allele d to his child.



Therefore, the genetic risk for this couple to have a child with cystic is $1/2$

B- Autosomal dominant disease:



*Determining the genetic risk for fetus III 3 to be affected:

Father Mother	$\frac{1}{2} D$	$\frac{1}{2} n$	$\frac{1}{4} Sick$	$\frac{1}{4} Healthy$
$\frac{1}{2} D$	Sick $\frac{1}{4} D/D$	Sick $\frac{1}{4} D/n$		
$\frac{1}{2} n$	Sick $\frac{1}{4} D/n$	$\frac{1}{4} n/n$ [Healthy]		

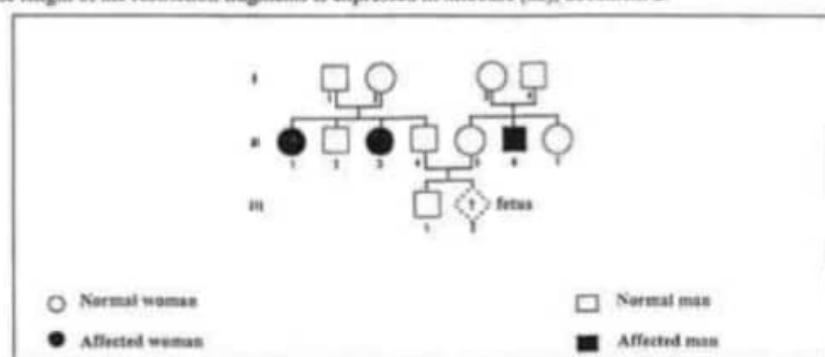


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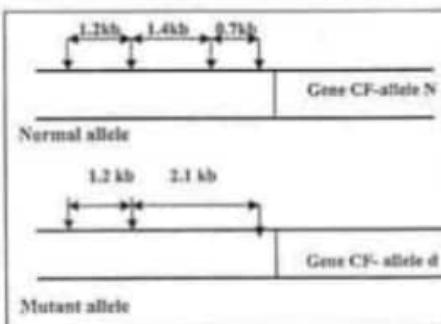
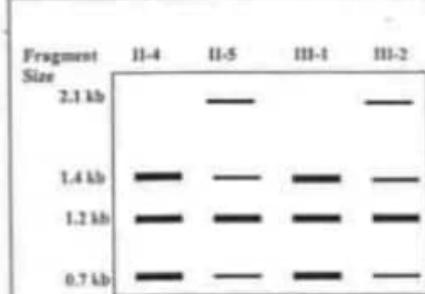
**Question III (5 pts)**

Document 1 represents the pedigree of a family of whom some members, figured in black, are affected by a disease called cystic fibrosis, a hereditary disease manifested by respiratory and digestive troubles. This disease is determined by a mutant allele of a gene called CF. This gene is located on chromosome 7, and very close to a non-coding region that has restriction sites recognized by the restriction enzyme Taq I.

The non-coding region close to the functional dominant allele N has four restriction sites for enzyme Taq I, while the non-coding region close to the mutated recessive allele d has three restriction sites. The length of the restriction fragments is expressed in kilobase (kb), document 2.

**Document 1**

- Indicate the possible genotypes of individuals II-4 and II-5. Justify the answer.
- Determine the genetic risk of couple II-4 and II-5 to have a sick child.

**Document 2****Document 3**

- Specify the site at which mutation took place, document 2. Justify the answer.

Document 3 shows the results of electrophoresis of the restriction fragments obtained by Southern Blot technique for individuals II-4, II-5, and their children.

- After analyzing the obtained results, indicate the real genotype of each of individuals II-4, II-5, and the fetus.
- Based on the above analysis, is this couple in risk of having affected children? Justify the answer.



Exercise 2 (5 points)		Albinism
1	N; normal dominant a: affected recessive Genotype I-1 N//N or N//a Genotype II-1 N//a Genotype III-6. a//a	1
2	Genetic Risk= frequency of father to be hetero X frequency of mother to be hetero X probability of having an affected child frequency of father III-4 to be hetero = 15/100 (no family history) frequency of mother III-3 to be hetero = 2/3 (having affected brother or sister) probability of having an affected child = 1/4 Genetic Risk= $15/100 \times 2/3 \times 1/4 = 10/400 = 1/40$	1
3	Since mutation is observed only at the level of gene E2, where in the seventh triplet the second nitrogenous base C in the normal individual is substituted by T in the albino individual. Therefore, the gene responsible for albinism in this family is gene E2 only.	1
4	The transcribed strand of the normal individual TAG TAC GCT TGG CGG ACG AGT TTG GGT... The m RNA of the normal individual: AUC AUG CGA ACC GGC UGC UCA AAC CCA... The amino acid sequence of the normal individual: Ile – Met – Arg – Thr – Gly – Cys – Ser – Asn – Pro – The transcribed strand of the albino individual TAG TAC GCT TGG CGG ACG ACT TTG GGT... The m RNA of the albino individual: AUC AUG CGA ACC GGC UGC UGA AAC CCA... The amino acid sequence of the normal individual: Ile – Met – Arg – Thr – Gly – Cys	1
5	The mutation by substitution at the level of the seventh triplet where the second nitrogenous base C is substituted by G in the mutant allele leads to a stop codon in the sequence of nucleotide in the transcribed m RNA. The translation of this mRNA results in an incomplete/truncated/ peptide of abnormal 3D structure. The non-functional E2 enzyme is not able to transform tyrosine into melanin, thus leading to albinism.	1

Exercise 3:

Incomplete Dominance

In order to study the transmission of the hereditary trait, the color of flowers in plants, we perform a cross between two pure lines of plants, one with yellow flowers and the other with blue flowers. All plants that are obtained in the first generation F1 have flowers of violet color.

- 1- Show that the alleles of the gene that determines the color of the flowers are incompletely dominant.
- 2- Designate by symbols the corresponding alleles.
- 3- Write the genotypes of the plants of each of the parents and that of F1 generation. We cross hybrid plants of F1 generation.
We obtain in F2 generation:
 - 25% plants having blue flowers
 - 25% plants having yellow flowers
 - 50% plants having violet flowers.
- 4- Make a factorial analysis that permits to verify the obtained result in F2 generation.

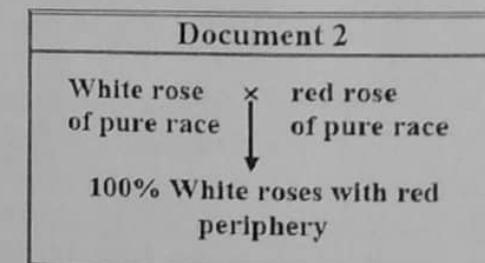
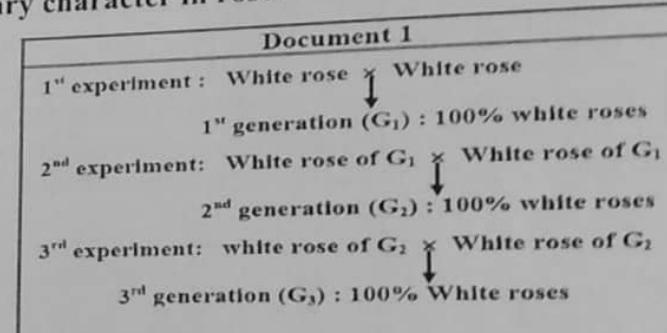
Exercise 4: Pure race and transmission of a hereditary character in roses

Sami has a rosebush of white flowers in his garden. He wonders if this rosebush is of pure race. To be sure, he realized several crosses as shown in document 1.

- 1- What is the problem posed by Sami?
- 2- Do the results of these crosses (document 1) solved the problem posed by Sami ? justify the answer.

Sami realized another cross Document 1 as shown in document 2.

- 3- 1- Is it the case of dominance or codominance?
Justify the answer by referring to document 2.
- 3-2- Designate by symbols the corresponding alleles.
- 3-3- Write the genotype of the white roses having red periphery and precise the different types of gametes produced by these roses.



- 2. Differentiate only the differences
 - 3. Pick out directly from the text / doc
 - 4. List \equiv (\rightarrow yes & if)
 - 5. Analysis (the better to start with result)
 - variable \rightarrow Re solt.
 - be \rightarrow death
 - (x) \rightarrow survive

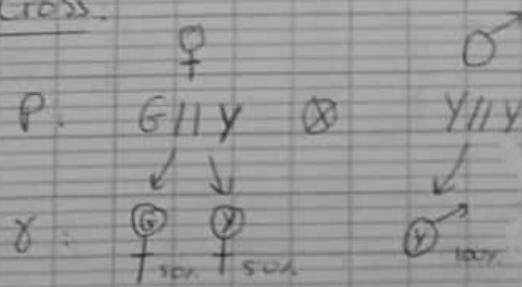
use while / where as / but to contrast.
- 6. Interpret: Analysis + Significance
 - $\rightarrow R^+ S^-$ (for studied the variable)
 - 7. Analyse \rightarrow This show that (variable) stimulate, enhances, inhibit responsible, promote, indispensable
 - 8. conclusion: it comes after
 - Analyse or interpret or comparison
then write a general conclusion
 - 9. Deduce: logical reasoning (analyse \rightarrow comparison) then reach a conclusion (with acquired knowledge)
 - 10. Draw out without writing the logical reasoning just give the conclusion directly.
 - 11. Explain: collect the necessary information to clarify the results of the experiment of phenomena - (acquisition acquired knowledge of knowledge)
 - 12. Justify like Explain.

5) No, since the test cross determines the genotype and the descendants are of incomplete dominance, So the sweet and bitter taste must be of pure race and the mild taste must be hybrid.

Exercise 2:

- 1) The cross of green squash of pure race with yellow squash of pure race gives the first generation of 100% striped squash (green & yellow).
- 2) It is the case of codominance since the 1st generation presents both phenotypes of the parents (green & yellow).
- 3). Let (G) be the symbol of the codominant allele coding for green squash.
Let (Y) be the symbol of the codominant allele coding for yellow squash.

4) 2nd cross.



phenotype: [GY] 50%

[Y] 50%

$F_2:$		$\text{♀ } \text{♂}$	$\text{Y } 100\%$
$\text{G } 50\%$	$\text{G/Y } 50\%$	$\text{G/Y } 50\%$	$\text{Y/Y } 50\%$
$\text{G } 50\%$	$\text{G/Y } 50\%$	$\text{G/Y } 50\%$	$\text{Y/Y } 50\%$
$\text{Y/Y } 50\%$	$\text{Y/Y } 50\%$		

→ The theoretical result confirms the experimental results.

Ch-1 -no.1

LEb Bac-09II

1- During the first meiotic division (reductional division), spermatocyte I produces two spermatocytes II that are subjected to the second meiotic division (equational division), each producing two spermatids. Then, the spermatids differentiate into sperm cells (spermiogenesis).

2- Population 1 corresponds to spermatocytes I because the quantity Q is duplicated during the S phase of interphase and becomes $2Q$ in spermatocyte I that has $2n$ chromosomes of 2 chromatids each.

Population 2 corresponds to spermatocytes II because after the reductional division of meiosis we obtain spermatocytes II that have n chromosomes each of 2 chromatids corresponding to the quantity Q of DNA.

Population 3 corresponds to spermatids or sperm cells because after the equational division of meiosis, we obtain 4 cells (spermatids) each having n chromosomes of one chromatid each corresponding to the quantity $Q/2$ of DNA. This same quantity remains constant after spermiogenesis that gives sperm cells.

In the fertile man, the number of germ cells is doubled from 2 to 4 then to 8 passing from population 1 to population 3 because the number of cells is doubled after each mitotic division. Each spermatocyte I produces 2 spermatocytes II and each spermatocyte produces 2 spermatids (1-2-4).

Sheet

Bruton disease

III_4 and I_2 are phenotypically normal couple, they gave III_4 diseased boy.

1. III_4 and I_2 are phenotypically normal couple, they gave III_4 diseased boy. Then the Bruton allele is masked by normal allele in at least one of the parent.

Thus, Bruton allele is recessive symbolized by "b" and normal allele is dominant symbolized by "N".

2. In the previous pedigree only boys are affected by Bruton, so the disease is sex-linked.

If the gene is located on nonhomologous part of Y then Father and all his sons must have the same phenotype but I_2 is normal and his son III_3 is diseased so it is not the case. Therefore the gene is located on nonhomologous part of X chromosome.

$$3. \text{III}_1 : X^N Y \rightarrow \text{IV}_1 X^N Y \\ \text{III}_2 : X^b X^b$$

4. Risk ($\frac{\text{Affected}}{\text{Expected}}$) = ??

Known genotypes from 3. In the pedigree we can see 1 male with disease and 1 female with disease. So we can assume that there is no normal female in pedigree.

Genotypes	III_1	$X^N Y$	\times	III_2	$X^b X^b$
gametes	X^N 50%	Y 50%		X^b 50%	X^b 50%

Table

$\frac{0}{2}$	X^N	Y
X^N	$X^N X^b$	$X^N Y$
X^b	$X^N X^b$	$X^b Y$

$\Rightarrow 25\% \text{ of all children will be affected}$

or
 $50\% \text{ of boys are affected}$

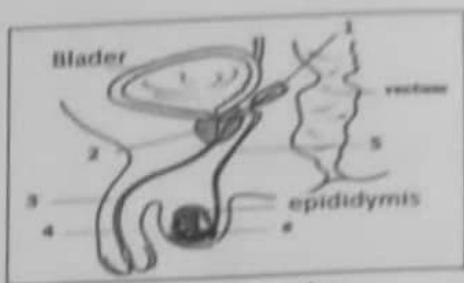
Answer the following exercises:

Exercise 1: (6 pts)

Male sterility

Document 1 shows the organs of the male reproductive system.

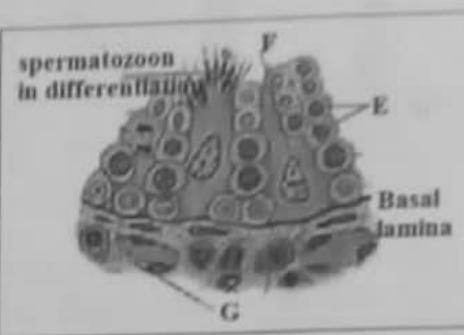
1. Label the figure of document 1.



Document 1

Document 2 shows a cross-section of a part of a structure of a fertile man.

2. Identify the cells E, F and G of this structure.
3. Explain, referring to your acquired knowledge, the variation in cell size as it passes towards the center of the structure shown in document 2.

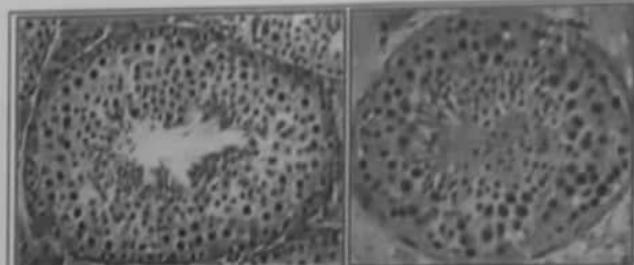


Document 2

Microscopic observation was performed for a seminiferous tubule of a normal man and another sterile. The obtained microphotographs are shown in document 3 (a and b) respectively.

4. Compare the two seminiferous tubules
5. Formulate a hypothesis that explains the sterility of this man.

*↑
don't
have
capacity to have
children*



a b

Exercise 2: (4.5 points)

Testosterone and Spermatogenesis

The testes continuously produce sperm and the male hormone 'testosterone' from puberty until death. Testosterone is essential for spermatogenesis and the maintenance of primary and secondary sexual characteristics.

Document 1 shows the variation in the number of sperm in the testes as a function of the concentration of testosterone in the fluid of the seminiferous tubules.

Testosterone concentration (ng/mL)	5	10	15	20
Number of spermatozoa per testis $\times 10^6$	40	150	210	250

Document 1

1. Doc 1 → Graph

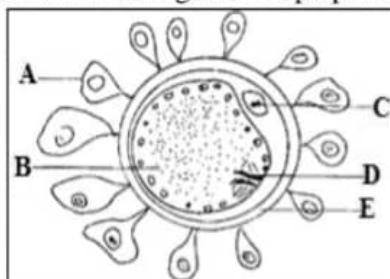
2. I interpret DOC

Document 1 represents a female gamete taken from the ovary of a woman. This gamete is prepared to be fertilized in vitro.

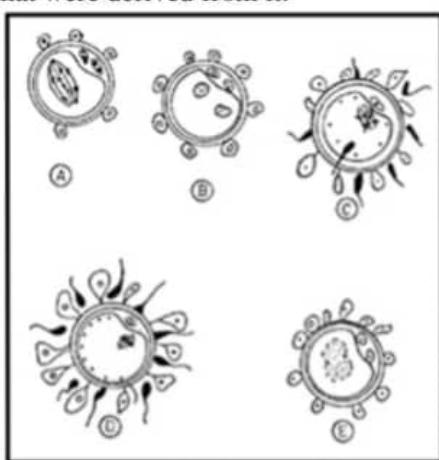
- 1 Label this document.
- 2 Show that this scheme is scientifically incorrect.

This gamete is placed in a container with a big number of human sperm cells where fertilization occurs. Document 2 represents some steps of this fertilization observed under optical microscope.

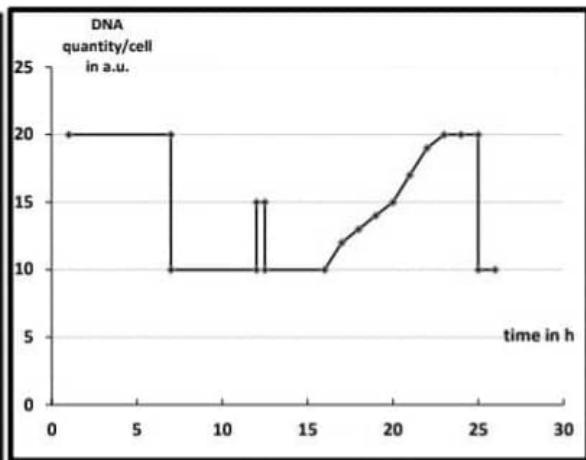
The graph of the document 2 shows the variation of the DNA quantity in the cell that was submitted to fertilization and in the cells that were derived from it.



Document 1



Document 2



Document 3

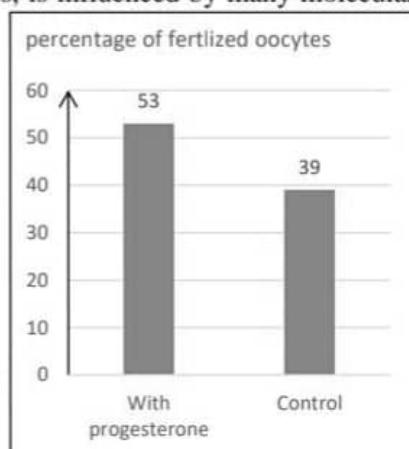
- 3 Specify for the cells in document 2, those that had accomplished meiosis I and those that had accomplished meiosis II.
- 4 Show that the increase in the amount of DNA from 10 to 15 a.u. mentioned in document 2, corresponds to the entry of the sperm cell.

Mammalian spermatozoa have to undergo a series of controlled molecular processes in the female reproductive tract. Capacitation, as a complex biological process, is influenced by many molecular factors, among which the hypothetical role of progesterone female hormone.

- 5 Justify the choice of the hypothesis mentioned above.

An experiment carried out to verify this hypothesis; spermatozoa are incubated in a progesterone medium with progesterone concentration of 20 ng/ml similar to that observed during ovulation period. The percentage of fertilized oocytes is evaluated and compared with that of the control spermatozoa. The results are shown in Document 4.

- 6 Show that the results obtained validate the hypothesis proposed above.

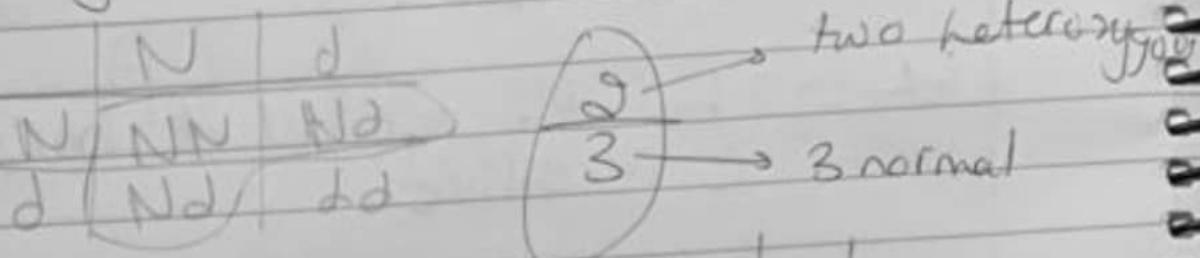


Document 4

Constants:

1. If the parent has a sick brother or sister.

Probability to be heterozygous = $\frac{2}{3}$

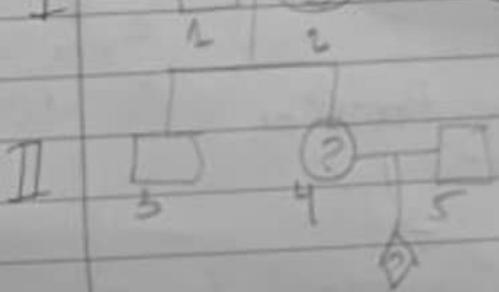


2. If there is no family history, the probability to be heterozygous is the same as the given probability to be heterozygous of population.

3. If the parent has an affected parent then, probability to be heterozygous = 1

I

disease is recessive



II₄:

N	d
d	Nd dd

& she is normal so Nd
100% heterozygous

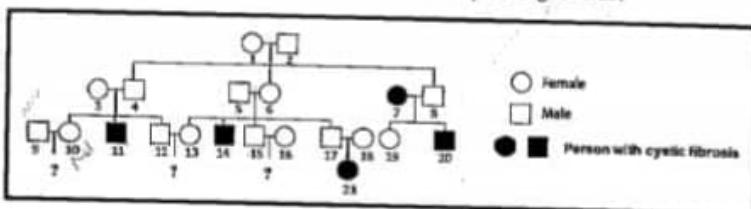
Another group of experiments was performed to study the function of cells 6 and 8

	Result
Experiment 1: Destruction of cell 6 by radiation	<ul style="list-style-type: none">-Spermatogonia couldn't be transferred to sperm-No nutrition and care of cells
Experiment 1: Destruction of cell 8 by radiation	No testosterone was produced

J. Identify the function of cells 6 and 8

**EXERCISE 2 Risk to be affected by cystic fibrosis**

The following pedigree shows the transmission of cystic fibrosis in a family. This disease is recessive. It has the gene located on the chromosome number 7. The proportion of heterozygous persons in the population to which this family belongs is 1/25.



1. Pick out from the text the statement that indicates that cystic fibrosis is an autosomal disease.
2. Evaluate, based on a punnet square the risk for the individuals 17 and 18 of the pedigree to have another child affected by cystic fibrosis.
3. Indicate the risk for the couple (7-8) to have another affected child by cystic fibrosis.
4. Determine the risk for the couple (15-16) to have an affected child by this disease.

Knowing that the individual 9 is from another population where there is no recording of cases of cystic fibrosis.

5. Specify the risk for the couple (9-10) to have an affected child.

Solution:

1. It has the gene located on the chromosome number 7.
2. Let N be the symbol of the normal allele and d be the symbol of the allele of cystic fibrosis. The two individuals 17 and 18 have already an affected child, they are heterozygous of genotypes Nd. Starting from the opposite punnet square the risk for this couple to have another affected child is 1/4.
3. In order to have an affected child, the two parents should be of genotype Nd, and they should both give the allele d to the child to be of genotype d/d and to be affected. The male 15 has two heterozygous parents since his brother is affected of genotype dd and had taken an allele d from each of the parents, then this male 15 has a risk of 2/3 to be heterozygous N/d (1/4 to be N/N, 1/2 to be N/d, and 1/4 to be d/d that is rejected since he is normal one). The risk for the female 16 to be heterozygous is equal to the proportion of heterozygous persons in the population since she has no family history. Then her risk to be heterozygous is 1/25. If the parents are heterozygous, each has a risk of 1/2 to give the allele d to the child. The risk for the child to be affected is $2/3 \times 1/2 \times 1/25 \times 1/2 = 1/150$.
4. The risk for the couple (9-10) to have an affected child is null since the male 9 has no risk to be heterozygous since the proportion of the heterozygous in its population is null knowing that there are no affected persons in it, then he will give surely an allele N to each of its children and make them all normal.



Ch-1-no.5-

1- Head of the sperm cell: 1: Mitochondria 2: Acrosome 3: Nucleus 4: Proximal centriole 5: Ovulation.

Section of the ovary: A: primary follicle; B: Secondary follicle; C, D: Cavitary (tertiary) follicle; E: Graafian (mature)follicle; F: Oocyte II (surrounded by corona radiata); G: Corpus luteum (yellow body); H: Corpus albicans (white body). K: sperm cell

2- E1: The spermatózoa removed from the seminiferous tubules and inserted into the uterus do not show any significant migration towards the fallopian tubes. This indicates that, the sperm cells in the seminiferous tubules have not yet acquired their motility.

E2: The sperm cells taken from the vas deferens canal and introduced in the uterus has normal fertilization ability. This indicates that, the sperm cells acquire their motility during their passage from the seminiferous tubes to vas deferens canal (in the epididymis).

E3: The sperm taken from the vas deferens canal and placed in the presence of the oocyte outside the uterus was not capable of fertilizing the oocyte, but it had the ability of normal fertilization in the uterus (E2). This indicates that sperm cells acquire their fertilization power (or capacity) while passing in the uterus.

E4: Fertilization can occur by injecting the sperm cells in the oocyte but the acrosome remains intact unlike during normal fertilization process where the acrosome opens during contact with the oocyte. This indicates that the acrosome could be involved in the entrance of the sperm into the oocyte during fertilization.

E5: Upon placing active sperm cells in a medium deprived of fructose, sperm cells become immobile and incapable of fertilization. This shows that fructose is a necessary element for the motility of sperm cells and its fertilization ability.

Therefore, the conditions necessary for spermatozoa action are:

- Passage in the male (epididymis) & female (uterus) genital ducts that assure motility and fertilization power or capacity.
- Presence of fructose (energy source).
- Presence of functional acrosome.

Exercise 1 (10 points)**Mechanisms of Sexual Reproduction**

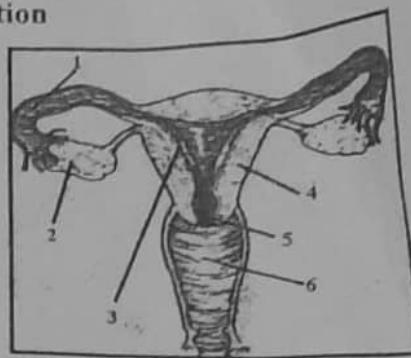
Reproduction is the process that ensures the survival of the species. Sexual reproduction involves two individuals of opposite sexes and belonging to the same species.

1. Name the two principal mechanisms of sexual reproduction in mammals and state the importance of each.

Sexual reproduction necessitates the presence of specialized and organized reproductive systems. Document 1 represents the human female reproductive system.

- 2.1. Annotate document 1.

- 2.2. Indicate the role of organs 1 and 2.



Document 1

Document 2 represents a schematic section of an organ in the female reproductive system.

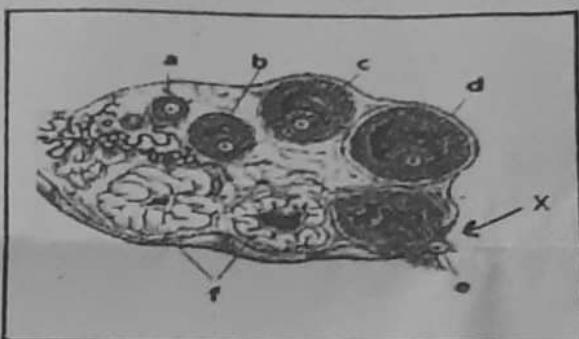
- 3.1. Name this organ.

- 3.2. Annotate the structures (a) to (f) and phenomenon X.

- 3.3. Indicate the phase of the sexual cycle during which structures (a) to (d) of document 2 can be seen.

- 3.4. State the importance of phenomenon X.

- 3.5 Explain the fate of structures (e) and (f) during the sexual cycle.

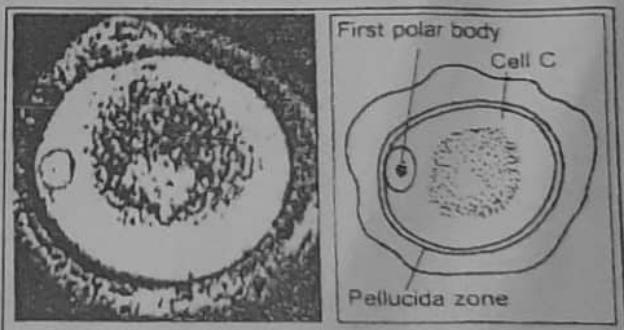


Document 2

Document 3 reveals a photograph accompanied by an interpretation diagram of the female sex cell during oogenesis.

4. Verify that this sex cell is an oocyte II and not an oocyte I.

5. Describe the karyotype of this oocyte.

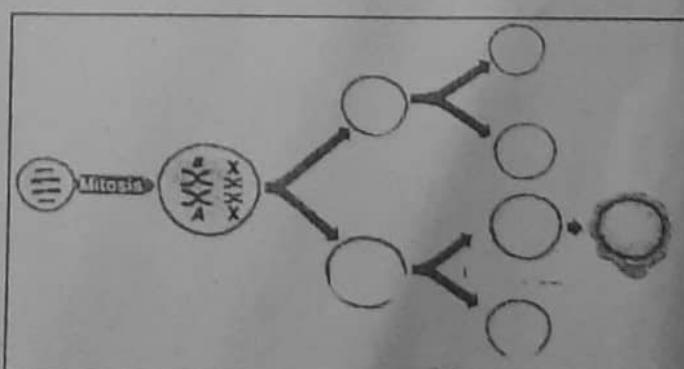


Document 3

Document 4 shows the process of oogenesis of a mother cell with $2n=4$.

6. Reproduce this diagram and complete it with all the details of the process.

Note that your diagram should include the chromosomal behaviour with the names of the cells and the phases of oogenesis.



Document 4

Exercise 3 (5pts)

Hemochromatosis

Hereditary hemochromatosis is a disease caused by an abnormality in the intestinal absorption of iron. The disease is manifested after 40 years in the form of liver, heart, and skin, joint and endocrine complications. This disease is linked to a protein, called "Hepcidin", secreted by the liver in the blood. Blood analysis of two individuals, one healthy and the other suffering from this disease, gave the results presented in document 1.

	Hepcidin	Amount of iron absorbed per day in the intestines (mg)	Amount of iron stored in the organs (g)
Healthy individual	Normal	1 to 2	5
Unhealthy individual	Abnormal	5 to 8	10 to 30

Document 1

1.1 Compare the amount of iron absorbed and stored in the organs between the healthy individual and the sick individual.

1.2 What conclusion can we draw out concerning the role of hepcidin?

- The synthesis of hepcidin is controlled by a gene located on chromosome 6. This gene exists in two allelic forms: the allele responsible for the synthesis of normal hepcidin and the allele responsible for the synthesis of abnormal one.
 Document 2 presents a pedigree where some members of the family are affected by hemochromatosis.

2- Referring to Document 2, show that the allele responsible for this disease is recessive and carried by an autosomal chromosome.

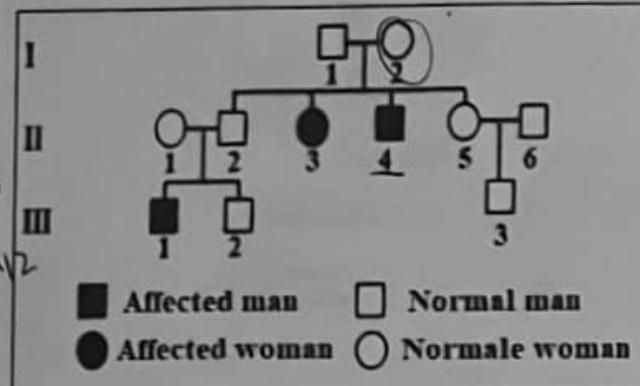
3- Specify the genotypes of individuals: I2, II4 and II5. (Use the symbols "H" to indicate the normal allele and "h" to designate the allele responsible for the disease).

Couple II1 and II2 expecting a third child,

4- determine the risk for this child to be diseased.
Document 3 shows a fragment of the transcribed DNA strand for each of the two alleles responsible for the synthesis of hepcidin in a healthy individual and in a sick individual.

5- Determine the sequence of the mRNA and that of the peptide chain that correspond to the two alleles of the studied gene.

6- Explain, based on all the above, the iron overload in the blood of a hemochromatosis patient.



Document 2

Number of nucleotide : 1060	1069	1074
Normal individual : ATA-CGT-GCC-AGG-TGG.....		
Affected individual : ATA-CGT-ACC-AGG-TGG.....		
Reading direction		

Document 3

		Second letter					
		U	C	A	G		
First letter	U	UUU Phe UUC UUA UUG	UCU UCC UCA UCG	UAU Tyr UAC UAA Stop UAG Stop	UGU Cys UGC UGA Stop UGG Tri	U C A G	
	C	CUC CUC CUA GUG	CCU CCC CGA CCG	CAU His CAC CAA Cln CAG	CGU CGC CGA CGG	U C A G	
A	AUU AUC AAA AUG Met	ACU ACC ACA ACG	ACG Tyr AAC AAA	AAU Asn AAC AGC AAG Lys	AGU Ser AGC AGA Arg AGG	U C A G	
G	GUU GUC GUA GUG	GCU GCC GCA GCG	GAU Asp GAC GAA Glu GAG	GGU GGC GGA GGG	Gly	U C A G	

Genetic code table

Exercise 2: (5pts)**Duchene muscular dystrophy**

Duchenne muscular dystrophy is a degenerative disease of muscle fibers. The boys with myopathy only synthesize no, or incorrectly synthesize a muscle protein, dystrophin. In order to determine the mode of transmission of this disease, we performed an electrophoresis of the DNA of the gene responsible for this disease in certain subjects of two families M and N.

The results obtained in the two families M and N are shown in document 1.

		Famille M		Famille N		
diseased father	Child a	Child b	Normal Mother	Child c	Child d	
A1	█	█	A1	█	█	█
A2	█	█	A2	█		█

Document 1

- Specify the allele corresponding to Duchenne muscular dystrophy.
- Show that the disease allele is recessive.
- Determine if the gene responsible for this disease is autosomal or carried by the X sex chromosome.

Recent techniques make it possible to determine the nature and the number of alleles from the analysis of chromosomes of one chromatid in diploid cells. The results obtained for some individuals of these families are shown in document 2.

	Diseased boy	Normal girl	Normal girl	Normal boy
Number of normal alleles	0	2	1	1
Number of alleles Responsible for the disease	1	0	1	0

Document 2

- Show that the results obtained in document 2 confirms the location of the gene for this disease.
- Indicate the genotypes of children b and d.

In order to check the effectiveness of the genetic treatment against this disease. Gene therapy is applied for the first time in mice with myopathy similar to that of Duchenne muscular dystrophy in human. This technique consists of injecting the dystrophin gene into a diseased organism by means of a virus vector which is harmless to mice and human species. Following this treatment, transversal sections of the diaphragm muscle

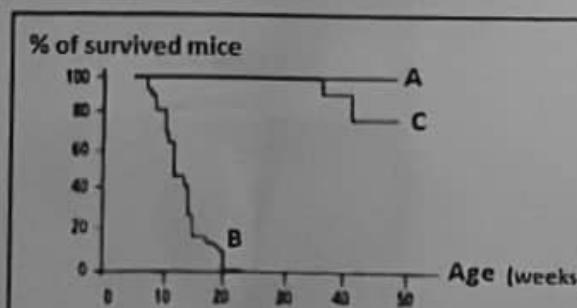
(respiratory muscle) of 3 groups of mice (A,B and C), then incubated with a fluorescent anti-dystrophin antibodies, the sections are observed under a fluorescence microscope.

The results obtained after 16 to 18 weeks are listed in document 3.
"anti-dystrophin anti bodies can bind to dystrophin"

Document 4 reveals the percentage of survival of the three groups of mice in function of time.

- Verify by referring to documents 3 and 4 that this gene therapy is effective.

Mice	Results
A: normal	Presence of fluorescence
B: myopathic, non-treated	Absence of fluorescence
C: myopathic, treated by injecting the dystrophin gene through a virus vector	Presence of fluorescence

Document 3**Document 4**

Exercise 48

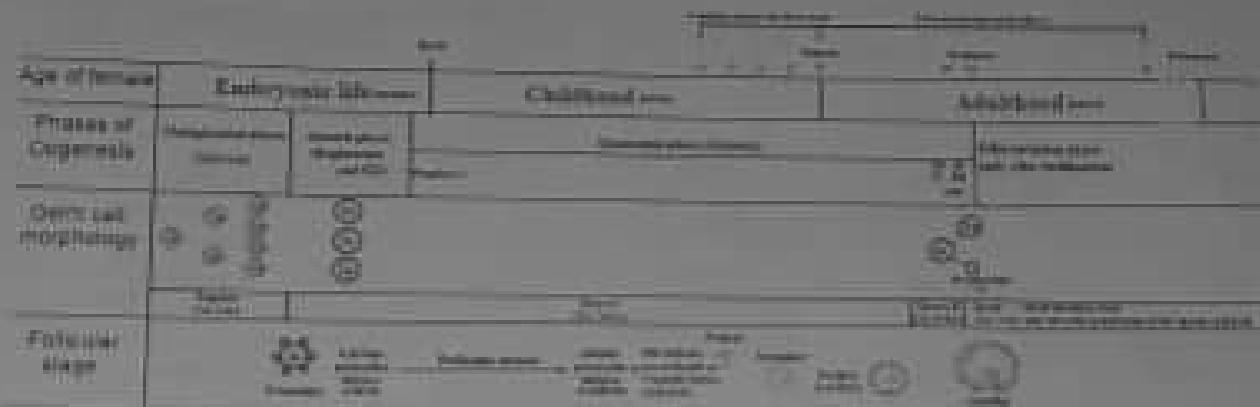
- 1) Is the rose bush of pure race?
- 2) Yes, since in each generation the same white allele is expressed; indicating that there's no hidden allele.
- 3>1) It's the case of codominance since the cross of white pure rose with pure red rose gives 100% white roses with red periphery indicating that the descendants inherit the white and red alleles and express them both.
- 3>2) Let (W) be the symbol of the allele coding for white rose (codominant).
Let (R) be the symbol of the allele coding for outside red rose (codominant).
- 3>3) $W \parallel R$. The gametes are $W_{50\%}$ and $R_{50\%}$.

Remark:

- 1- To check if the race is pure, we apply self-cross (3 generations) (to identify the purity of the race).
- 2- To identify the real genotype of the dominant, we apply test-cross.

~~additional division~~, only ~~of fertilization takes place producing ootid and the second polar body, otherwise, it degenerates after 24 hours.~~

- Differentiation phase: it is characterized by the transformation of ootid into mature female gamete (ovum). In a very short period of time before karyogamy (union of male and female pronuclei). Differentiation occurs only just after fertilization.



Oogenesis versus Folliculogenesis

Documents 6: Fertilization (P30-31)

- Fertilisation is the joining of male and female gametes giving rise to a diploid cell called zygote.
- Spermatozoa get their fertilization capacity in the female's reproductive system. This process is known as capacitation. Only 1% of deposited spermatozoa can reach the uterus and only few hundred reach the oviduct which is the site of fertilization. Other spermatozoa degenerate. (Doc. a)

Steps of fertilization: (Doc. a)

- **Adherence:** sperm cells approach oocyte II where they are entrapped in the viscous substance that links the prehunculated follicular cells which in turn retract and become diffuse. The head of the spermatozoon is now in contact with the zona pellucida.
- **Penetration:** acrosomal enzymes start to digest the zona pellucida surrounding oocyte II. The propulsion of the flagellum ensures the tangential penetration of the sperm's head and the fusion of cell membranes and hence, the entrance of the head into the cytoplasm of the oocyte. This penetration induces the release of the contents of the cortical granules into the pericaryal space thus forming the fertilization membrane which blocks the entrance of other spermatozoa (polyspermy).

Formation and fusion of pronuclei: after the absorption of the sperm's head, the oocyte II is activated and resumes equational division where the second polar body is released. The nucleus of the ovum is reorganized and then it swells and becomes the female pronucleus. The spermatozoon loses its flagellum and mid-piece, its nucleus swells and forms the male pronucleus. Each of the female and male pronuclei contains 16 chromosomes with 1 chromatid.

The membranes of both pronuclei fuse with each other where the two maternal and paternal haploid sets

are mixed leading to the formation a diploid cell which undergoes replication of genetic material preparing itself for the first mitotic division.

Life cycle: (Doc. d)

The life cycle consists of two phases, diploid and haploid.

The diploid phase is ensured by fertilization while the haploid one is ensured by meiosis.

8. Identify the mutant allele.
 9. Verify based on document 4 :
 - 9.1 if the fetus is affected by Tay Sachs disease. Normal of genotype Nd
 - 9.2 the genetic risk of couple (7,8) of having affected child by Tay Sachs.

Mother 8 is homozygous of genotype NN and surely gives her children the normal allele and since it is dominant I, it will be expressed .Thus this couple has no risk for having affected child

Exercise 2 (12 points)

Tay Sachs Syndrome

Tay-Sachs disease is a rare inherited disorder due to accumulation of a type of lipid, called GM2 ganglioside that progressively destroys nerve cells (neurons) in the brain and spinal cord, which causes the symptoms of Tay Sachs disease. Beta-hexosaminidase is an enzyme that breaks down GM2 ganglioside and prevents its building up in the cells. Beta-hexosaminidase enzyme is coded by HEXA- gene. The frequency of heterozygotes for HEXA- gene is 1/30 in a given population.

Document 1 represents partial sequence of nucleotides of non-transcribed DNA strand of Hexa -gene of the normal allele and of the equivalent region of the diseased allele

Non-transcribed DNA strands of normal and mutant alleles	
Normal allele	¹ CGTATATCCTATGCCCTGAC ²¹
Mutant allele	¹ CGTATATC TATC C ATGCCCTGAC ²¹

4 bases inserted

Document 1

1. Compare the nucleotide sequences of the two alleles.
2. Indicate the type of mutation. [Insertion]
3. Write the partial sequence of amino acids of Beta-hexosaminidase enzyme coded by each allele of HEXA gene. [Genetic code table is on last paper]
4. Explain how the modification in the nucleotide sequence of mutant allele can lead to Tay Sachs syndrome.

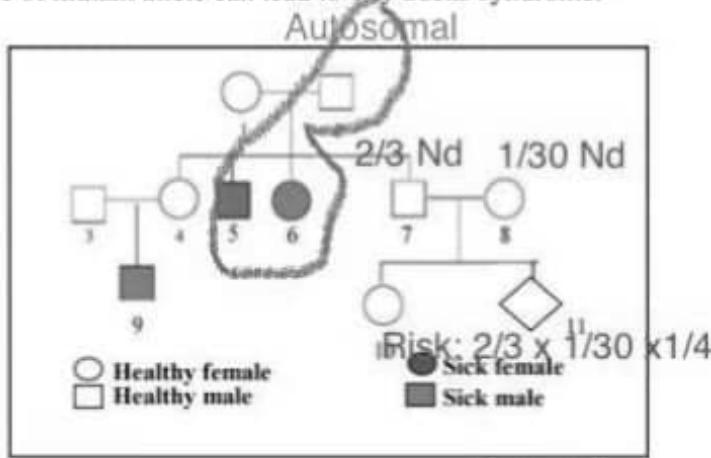
Document 2 represents a family that belongs to the given population by which some of its members are affected with Tay Sachs disease.

5. Specify if the allele of Tay Sachs is dominant or recessive. Recessive
6. Determine the chromosomal location of HEXA gene. Autosomal proved by elimination
7. Determine the genetic risk for couple (7, 8) to have an affected child (Fetus 11) with Tay Sachs

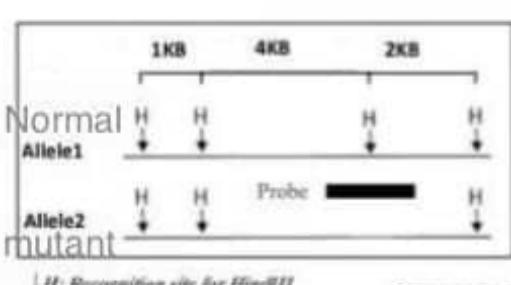
Risk : $2/3 \times 1/30 \times 1/4$

Couple (7, 8) is worried if the fetus is affected with Tay Sachs. For that, they demanded prenatal diagnosis of this fetus.

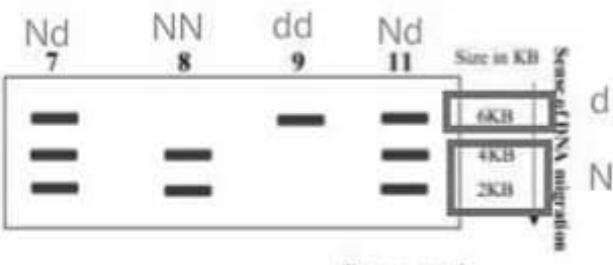
The part (fragment of 7KB) of the normal allele and that of mutant allele have restriction sites for Hind III enzyme. The molecular radioactive probe can hybridize to its sequence or part of it on the restriction DNA fragments that result from digestion of the two alleles by Hind III enzyme (document 3). Document 4 shows the DNA analysis of some members of this family as well as the fetus.



Document 2



Document 3

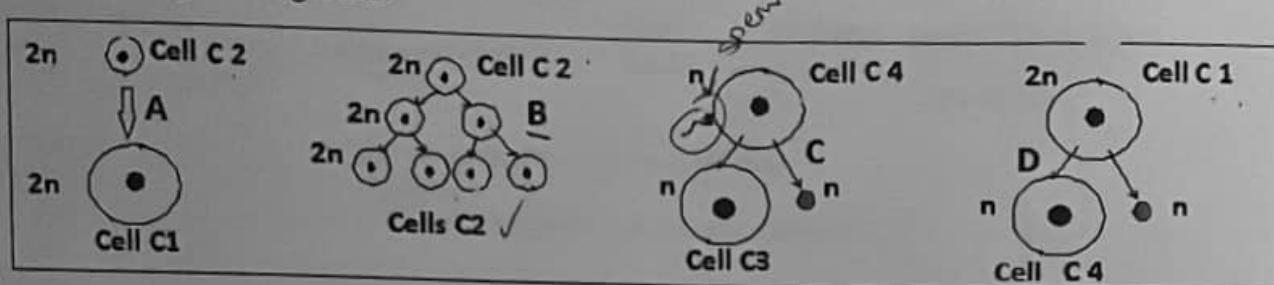


Document 4



Exercise 1: (5pts) Oogenesis in mammals

Oogenesis is the process that leads to the formation of haploid female gametes from diploid stem cells. It is a discontinuous process. It begins during fetal life and resumes its activity at puberty. Document 1 shows the disordered stages of oogenesis.



Document 1

1: name the cells C1, C2 , C3 and C4 as well as the processes A,B,C and D..

2: classify these processes in chronological order.

3: justify the cause of reduction in the number of chromosomes during process D.

Biosphenol A (BPA) is an industrial chemical used in the production of polymeric plastics. Under harsh conditions these plastics may release BPA, which then can seep into the environment. BPA affects the nuclear behavior in meiosis and oocyte development. According to the "developmental Origin of Health and Disease" (DOHaD) proposed that the exposure of pregnant woman to BPA may alter the oogenesis of the female fetus, especially if the exposure occur during critical periods of fetal development.

Document 2.

4- Pick out, from document 2, the hypothesis proposed by the DOHaD which explains the effect of BPA on the female fetus.

In order to study the effect of BPA on the variation in the number of chromosomes in the chimpanzee sex cells ($2n=48$) experiment is performed on two female chimpanzees; female A has been exposed to BPA during her fetal life, and female B which has never been exposed to BPA. The results are shown in document 3

	Sex cell starting from Fetal life and at puberty	Oogonia	Oocyte 1	Oocyte 2
Number of Chromosomes In each cell (a.u.)	The female chimpanzee (B) has never been exposed to BPA during her fetal life development. The female chimpanzee (A) has been exposed to BPA during her fetal life development.	48 48	48 48	24 48

Document 3

5- Construct a histogram that represents the results presented in document 3.

6- Show that BPA affects the nuclear behavior during Oogenesis.

Documents 3: Meiosis (P22-23)

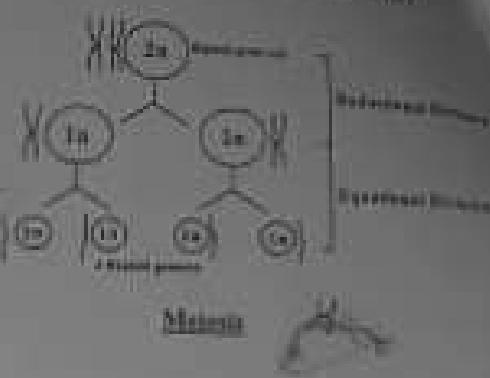
- Meiosis is a type of cell division during which diploid germ cells (2n) give rise to haploid gametes (n). It takes place in the testes and the ovaries. It is the process by which the genetic material is conserved in a species.
- Meiosis takes place in two successive divisions: reductional (2n → n) and equational (n → n).
- During interphase (in the S phase) and prior to reductional division, each chromosome duplicates itself by DNA replication. DNA replication does not take place during meiosis.

Stages of Meiosis

1. Reductional division (Meiosis I)

Prophase I

- Duplicated chromosomes condense and become visible.
- Nuclear membrane disappears.
- Centrosome divides into two centrioles (satellites) in animal cells.
- Akinetotic spindle (spindle fibers) forms.
- Homologous chromosomes pair, synapse with each other forming tetrads. Each homologous pair binds to one fiber of the spindle apparatus.
- **Metaphase I:** chromosomes arrange themselves at the equator of the cell forming the equatorial plate (metaphase plate).
- **Anaphase I:** homologous chromosomes separate and move to opposite poles of the dividing cell. This movement is called "polar ascension".
- **Telophase I:** in this phase, first cytokinesis takes place giving rise to two daughter haploid cells in which each chromosome is made up of two chromatids.



2. Equational division (Meiosis II)

- **Prophase II:** similar to prophase I, but the chromosomes exist only in one copy and hence they do not synapse forming tetrads. Each chromosome binds to one spindle fiber.
- **Metaphase II:** chromosomes arrange themselves at the equator of the cell forming the equatorial plate (metaphase plate).
- **Anaphase II:** sister chromatids of each chromosome separate and migrate to opposite poles of the dividing cell. This movement is called "polar ascension".
- **Telophase II:** in this phase, second cytokinesis takes place giving rise to four daughter haploid cells in which each chromosome is made up of one chromatid.

Documents 4: Spermatogenesis (P24-26)

- In the testicle, there are about 200 to 300 lobules, each of which contains one to four coiled tubules, the seminiferous tubules which produce sperm cells or spermatozoa. (Doc. a)
- Spermatogenesis is a continuous process by which male gametes (n) are produced in the wall of the seminiferous tubules in a centripetal direction from diploid germ stem cells. It starts at puberty and continues till death. (Doc. b, Doc. c)
- Each spermatogenic cycle lasts around 74 days in humans.
- Between the seminiferous tubules, there are groups of interstitial cells, called Leydig cells, which secrete the male hormone, testosterone.
- Spermatogenesis includes 4 phases (Doc. d):
 1. **Mitigation phase:** characterized by successive mitotic divisions of the germ stem cells; the spermatogonia (2n chromosomes with 1 chromatid each)

1-2- Specify on which sites can these enzymes act.

The following document 2 represents the sequence of 2 alleles of same gene where allele "s" correspond to normal allele while allele "m" correspond to the affected allele.

partial sequence of the 2 alleles of studied gene	
allele "s"	1 GCCACAGAACATCAGATTCCGCACGACTC 26
allele "m"	1 GCCACTGAATTGATTCCGCACGACTC 26

Doc.2

2- Describe the effects of the enzymes studied in doc. 1 on the allele "s" and on allele "m".

3- Evaluate the length (in nucleotides) of the obtained fragments.

4- Schematize the result of electrophoreses, of obtained fragments after digestion with the enzyme(s).

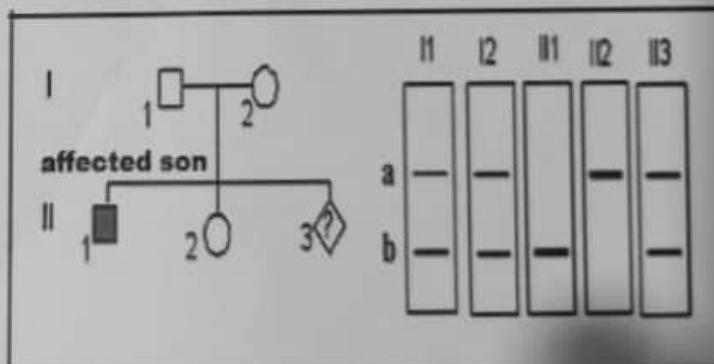
5- Explain by referring to your acquired knowledge, how the mutation of a given gene can cause disease.

The following pedigree (doc.3) belongs for a family where normal parents given birth to one affected child with recessive genetic disease and other normal child. During the 3rd pregnancy, all the members are tested by southern method.

6- Specify which of the two bands a or b corresponds to the affected allele.

7- Establish the prenatal diagnostic for child II3. Justify the results from the autoradiographs.

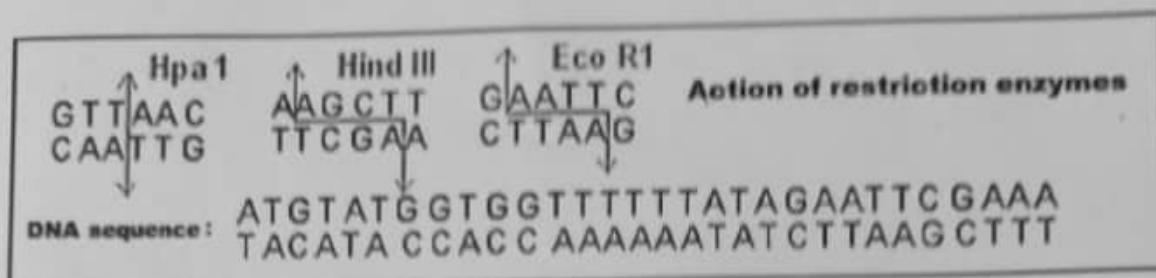
2



Doc.3

Exercise: 2 (7pts) Prenatal diagnosis

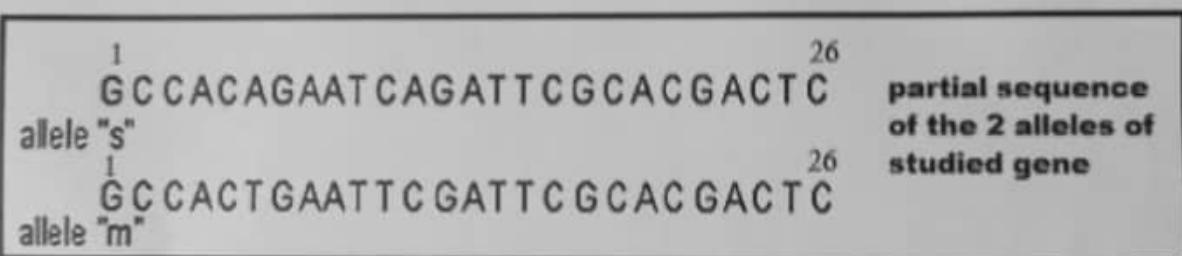
Restriction enzymes are biological scissors used by the bacterium to digest the DNA of bacteriophage that infect it by cutting the DNA molecules in specific sites called restriction sites, three of these enzymes are studied in document 1.



Doc.1

- 1-1-Determine which restriction enzyme(s) can act(s) on the given sequence.
- 1-2- Specify on which sites can these enzymes act.

The following document 2 represents the sequence of 2 alleles of same gene where allele "s" correspond to normal allele while allele "m" correspond to the affected allele.



Doc.2

- 2- Describe the effects of the enzymes studied in doc. 1 on the allele "s" and on allele "m".
- 3- Evaluate the length (in nucleotides) of the obtained fragments.
- 4- Schematize the result of electrophoreses, of obtained fragments after digestion with the enzyme(s).
- 5- Explain by referring to your acquired knowledge, how the mutation of a given gene can cause disease.

The following pedigree (doc.3) belongs for a family where normal parents given birth to one affected child with recessive genetic disease and other normal child. During the 3rd pregnancy, all the members are tested by southern method.

- Specify which of the two bands a or b corresponds to the affected allele.
Establish the prenatal diagnostic for child II3. Justify the results from



Part I: Reproduction and Genetics

Chapter 1: Basic mechanisms of sexual reproduction

Documents 1: Male and female reproductive systems (P18-19)

- In humans, reproduction involves a male and a female reproductive system:
 - The male reproductive system includes: (Docs. a,b)
 - ✓ Two gonads (testes) that produce spermatozoa by meiosis.
 - ✓ Genital tracts (1 epididymis, 2 vasa deferentia, 1 urethra) for maturation, circulation and emission of spermatozoa through the penis. Urethra also conducts urine.
 - ✓ Accessory glands (seminal vesicles, prostate gland, two bulbourethral glands (Cowper's glands)) that secrete a fluid rich in nutrients necessary for survival and motility of spermatozoa.
 - ✓ Penis or copulatory organ.
 - The female reproductive system includes: (Docs. d,e)
 - ✓ Two gonads (ovaries) that produce alternately an oocyte (ovum) by meiosis during each cycle.
 - ✓ Genital tracts (two oviducts or Fallopian tubes) that receive male gametes and constitute the site of fertilization.
 - ✓ Uterus (womb) that houses and nourishes the embryo.
 - ✓ Vagina or copulatory organ.
 - ✓ External genitalia.
- Semen is a fluid that contains 10% spermatozoa and 90% seminal fluid which is secreted by accessory glands to ensure survival and motility of spermatozoa.
- The normal count of spermatozoa is 100 million/ml of semen.
- Both males and females have sexual characteristics which differentiate them from each other:
 - Primary sexual characteristics represented by gonads.
 - Secondary sexual characteristics represented by morphological, physiological and behavioral differences between a male and a female.

Documents 2: Diploid and haploid cells (P20-21)

- Karyotype: a characteristic arrangement of the chromosomes of an individual, according to their size, their form and the distribution of their bands.
- Karyotyping is a test used to identify chromosome abnormalities as well as, the cause of malformation or disease. This test can be performed on a sample of blood, bone marrow, amniotic fluid, or placental tissue. This test can:
 - Count the number of chromosomes.
 - Look for structural changes in chromosomes.
- Diploid cells ($2n$): cells in which each chromosome exists in two copies constituting a pair of homologous chromosomes that share the same structural characteristics. All somatic and germline cells are diploid. (Doc. b)
- Haploid cells ($1n$): cells that have only one copy of each chromosome. These cells are involved in reproduction and they are known as gametes. (Doc. c)
- " n " is the number of different chromosomes.

($\frac{1}{2}$ to take diseased allele from mother since $\frac{1}{2}$ to take normal allele and $\frac{1}{2}$ to take diseased allele)

($\frac{1}{2}$ to take diseased allele from father since $\frac{1}{2}$ to take normal allele and $\frac{1}{2}$ to take diseased allele)

$$\text{So, Risk} = \frac{1}{20} \times \frac{2}{3} \times \frac{1}{2} = \frac{2}{240} = \frac{1}{120}$$

Case 4: both parents have history: couple III3-III4

- Risk mother to be hybrid = risk father to be hybrid = $\frac{2}{3}$ (same explanation as in case 3)
- Risk for fetus to inherit diseased allele from each parent = $\frac{1}{4}$ (same explanation as in case 3)
- So, Risk = $\frac{2}{3} \times \frac{2}{3} \times \frac{1}{4} = \frac{4}{36} = \frac{1}{9}$

Case of parents without family history

If parents have no family history for a certain disease, the risk for parents to be hybrid is equal to risk to be heterozygous in the population which must be mentioned in the given

Example: Risk for an individual to be heterozygous or carrier for cystic fibrosis allele in France = $\frac{1}{20}$

- Risk for mother to be heterozygous = $\frac{1}{20}$ (no family history takes the risk of population)

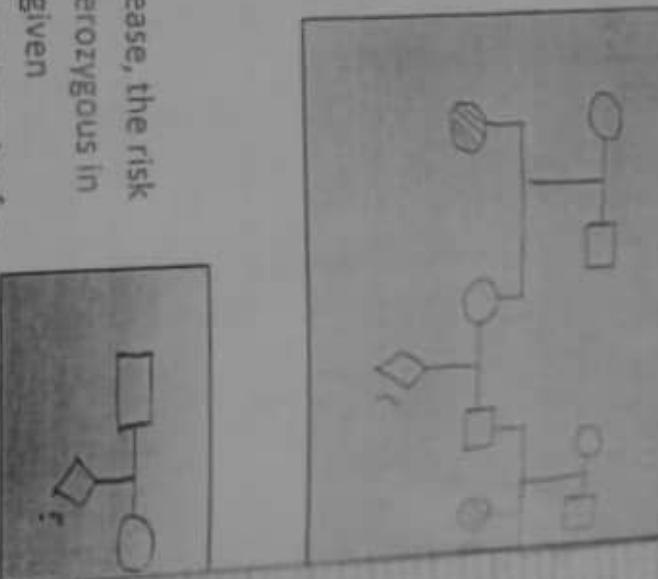
Risk for father to be heterozygous = $\frac{1}{20}$ (no family history takes the risk of population)

Risk for child to take diseased allele from each hybrid parent = $\frac{1}{2} \times \frac{1}{2} = \frac{1}{4}$

($\frac{1}{2}$ to take diseased allele from mother since $\frac{1}{2}$ to take normal allele and $\frac{1}{2}$ to take diseased allele)

($\frac{1}{2}$ to take diseased allele from father since $\frac{1}{2}$ to take normal allele and $\frac{1}{2}$ to take diseased allele)

$$\text{So, Risk} = \frac{1}{20} \times \frac{1}{20} \times \frac{1}{4} = \frac{1}{1600}$$

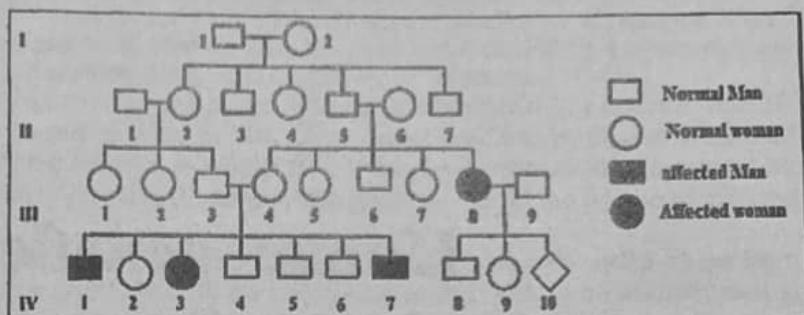


EXERCISE II An enzymatic deficiency

Fructose, or hereditary fructose intolerance, was clinically demonstrated in 1956. It is a congenital (hereditary) disease that prevents the absorption of fructose in the form of polymers (it is linked to a deficiency in aldolase B). This enzyme, found in the liver, small intestine and kidneys, cleaves fructose-1-phosphate to DHAP and glyceraldehyde in order to allow further glycolysis for ATP production by the cell resulting in an accumulation of fructose-1-phosphate in the liver and kidneys. This molecule, which cannot follow another metabolic pathway, becomes toxic to these organs in the long term.

It is a rare disease that affects 1 / 20,000 births, and it is estimated that the % of heterozygotes in the human population is 1/5000.

- II. Pick out from the text:**
- III. The probable cause of fructose intolerance.**
- IV. the consequences of this long-term impairment.**



Document 1

The study of the transmission of this disease in a family made it possible to make the following genealogical tree:

- 2. Specify according to the pedigree:**

2.1. If the allele responsible for this disease is dominant or recessive.

2.2. If the allele responsible for this disease is linked to an autosome or on a gonosome.

Document 2 below shows the DNA-base sequence of a fragment of the gene (non-transcribed strand) coding for normal and mutated aldolase B:

Normal Sequence	AAT GGA CTG GTA CCT ATT GTT GAA
Abnormal Sequence	AAT GGA CCT ATT GTT GAA

Document 2

- 3. Determine, using the table of the genetic code, the amino acid sequences of each fragment of the aldolase coded by each of two alleles, and draw out the cause of the disease.**

The couple III (8, 9) awaits a 3rd child (10) and suspects that this child will be affected as his cousins 1, 3 and 7 (IV).

- 4. Evaluate the risk of this child being affected.**
- 5. Explain why this risk is much lower compared to his cousins.**

	U	C	A	G				
U	UUU UUC UUA UUG	phényl-alanine leucine	UCU UCC UCA UCG	sérine	UAU UAC	tyrosine	UGU UGC	cystéine
					UAA UAG	STOP	UGA UGG	STOP tryptophane
					CAU CAC	histidine	CGU CGC CGA CGG	arginine
					CAA CAG	glutamine		
C	CUU CUC CUA CUG	leucine	CCU CCC CCA CCG	proline	CAU CAC	histidine	CGU CGC CGA CGG	
					CAA CAG	glutamine		
					AAA AAG	lysine	AGA AGG	arginine
					AAU AAC	asparagine	AGU AGC	
A	AUU AUC AUA AUG	isoleucine méthionine	ACU ACC ACA ACG	thréonine	AAA AAG	lysine	AGA AGG	arginine
					AAU AAC	asparagine	AGU AGC	
					AAA AAG	lysine	AGA AGG	
					AAU AAC	asparagine	AGU AGC	
G	GUU GUC GUA GUG	valine	GCU GCC GCA GCG	alanine	GAU GAC	acide aspartique	GGU GGC GGA GGG	glycine
					GAU GAC	acide aspartique	GGU GGC GGA GGG	
					GAU GAC	acide glutamique	GGU GGC GGA GGG	
					GAU GAC	acide glutamique	GGU GGC GGA GGG	

Document 3

5. $\text{X}^{\text{N}}\text{Y}$ XX
Referring to the pedigree only boys are affected by
Bruton
Referring to karyotype Fetus IV_2 is a girl with sex
chromosome is XX
Thus IV_2 will not be affected by Bruton

6.

	III_1	III_2	IV_1	IV_2
A	—	—	—	—
B	—	—	—	—

- specify the band that correspond to diseased allele.
 III_1 genotype is $\text{X}^{\text{D}}\text{Y}$ and he has only band A, so band A
corresponds to normal allele.
Thus band B corresponds to diseased allele.

7. Draw out the genotype and phenotype of the fetus IV_2
genotype : XX^{D}
phenotype : ♀ ~~girl~~ [X] normal girl

Exercise 1: (10 pts)**The p53 Gene and cancer**

The P53 gene is located on chromosome 17. It codes for a protein TP53 which plays a role in transcription and therefore intervenes in multiple important cellular functions such as the regulation of the cell cycle and apoptosis (programmed cell death). The inactivation or this gene is marked in 50% of cases of sporadic cancer (no family history) and the inactivation results rather in an abnormal and rapid multiplication of cells and the development of a tumor mass. Epidemiological studies show a correlation between the inactivation of the p53 gene and the exposure to ultraviolet rays (UV studies rays), smoking and chronic alcoholic consumption.

- 1- Pick out from the text
 - 1.1-The role of TP 53.
 - 1.2-The effect of p53 gene inactivation.
 - 1.3-The factors that cause the inactivation of p53 gene.

In the framework of studying the various factors that lead to the development of liver cancer and its symptoms, the following studies have been carried out as follows:

Study 1: the sequence of a fragment of the p53 gene in two individuals: healthy individual 1 and an affected individual 2 by liver cancer is determined.

Document 1 shows the results of a part of the non-transcribed strand of the DNA

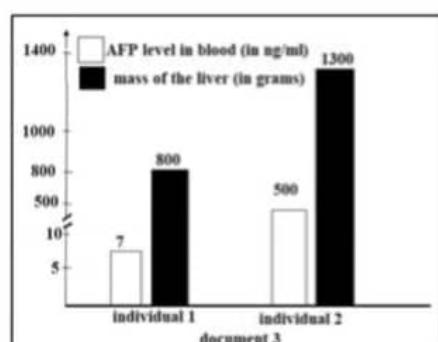
- 2- Identify the type of mutation in the part of the sequence of the affected individual.
- 3- Determine the amino acid sequence for each of the above parts of genes in document 1 by using document 2.
- 4- Explain how the modification in the gene p53 leads to the appearance of the liver cancer in individual 2.

Number of codon	5331	5335
Normal individual	CCTTCAGTCAGGAAA	
Affected individual by liver cancer	CCTTCAGTCAGTAAA	
Document 1		

		Second letter						
		U	C	A	G			
First letter	U	UUU Phe UUC UUA UUG	UCU Ser UCC UCA UCG	UAU Tyr UAC UAA STOP UAG STOP	UGU Cys UGC UGA STOP UGG Trp	U C A G		
	C	CUU Leu CUC CUA CUG	CCU CCC CCA CCG	CAU His CAC CAA Gln CAG	CGU CGC CGA CGG	U C A G		
	A	AUU Ile AUC AUA AUG Met	ACU ACC ACA ACG	AAU Asn AAC AAA Lys AAG	AGU Ser AGC AGA Arg AGG	U C A G		
	G	GUU Val GUC GUA GUG	GCU GCC GCA GCG	GAU Asp GAC GAA Glu GAG	GGU GGC GGA GGG	U C A G		
Third letter								

Study 2: liver mass and activity are measured in two individuals. Liver activity is reported by referring to the blood level of protein AFP produced by the liver. Normally, the level of this protein is in the range of 5 ng/mL to 10 ng/mL. The results are shown in document 3.

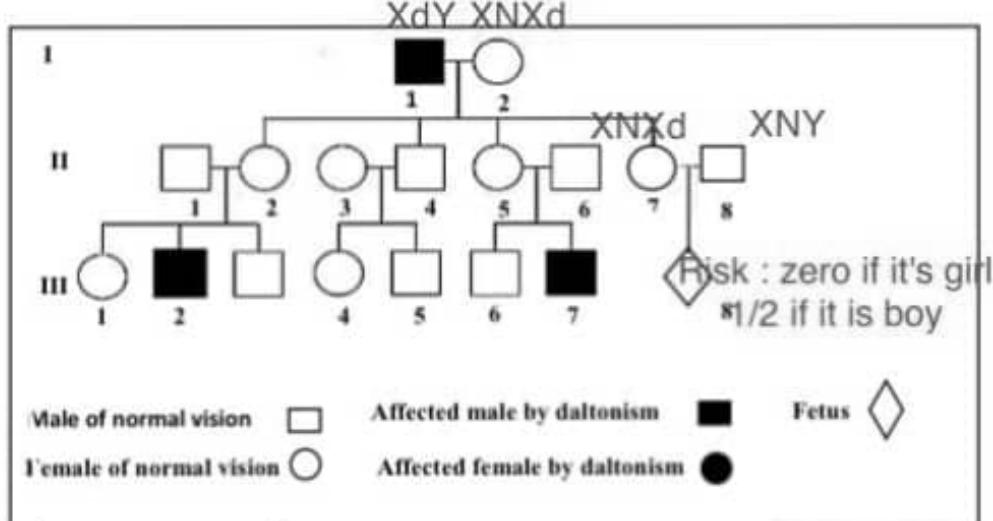
- 5- Transform document 3 into a table.
- 6- Determine from document 3 the individual who is affected by liver cancer.



Only boys are sick then the gene is sex linked :not in Y: father and son different pheno therefore gene is on X

Exercise 1 (8 points) Daltonism

Color blindness or Daltonism is a hereditary disease, which is characterized by difficulty in distinguishing certain colors. Document 1 shows the genealogical tree of a family where some of its members suffers from this anomaly



Document 1

- Indicate if the allele responsible for daltonism is dominant or recessive. Justify the answer.
 - Show that the gene responsible for daltonism is localized on non-homologous part of chromosome X.
 - Specify the genotypes of the individuals I-1, I-2, II-7 and II-8.
 - Determine the genetic risk for fetus III-8 to be affected by daltonism.
- The parents are worried if their expect child is affected by daltonism;
They demanded diagnosis of the fetus. The doctor prescribed different tests:

Test 1: The karyotype of the fetus was prepared in order to study the case, document 2.

5. Show, based on documents 1 and 2, that the karyotype of the fetus

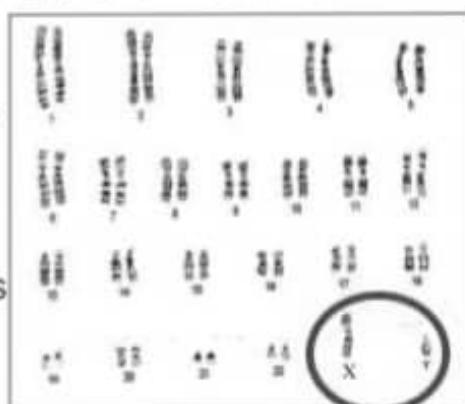
Fetus is a boy (XY) he may be inherited X^N or X^d from his

mother and thus the phenotype couldn't be known

Test 2: DNA analysis of different members of the family (Southern blot).

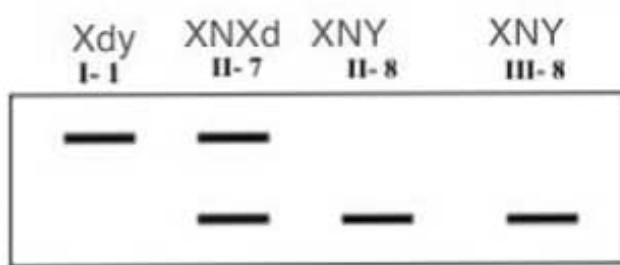
The obtained results are presented in document 3

A1 and A2 are the two different alleles of the studied gene, normal and mutant.



Document 2

6. Identify the mutant allele of daltonism. A1



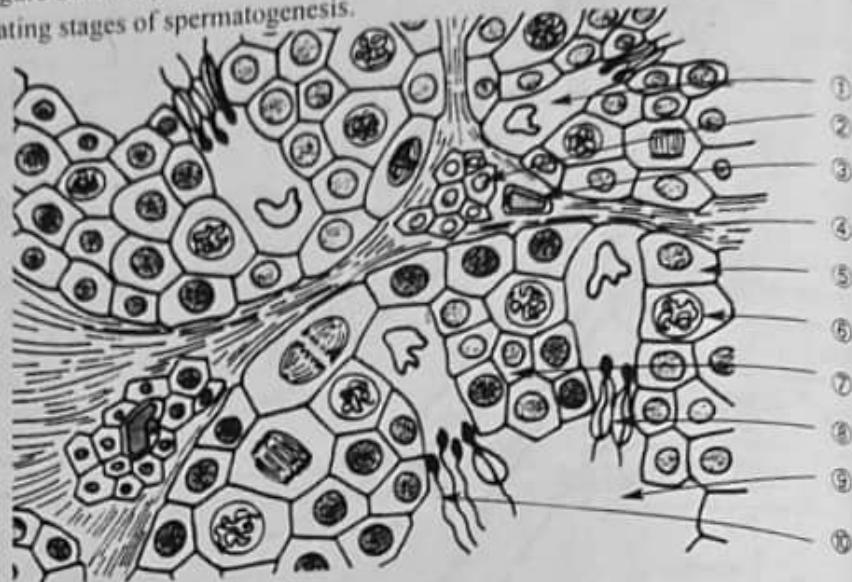
Document 3

7. Draw out the genotype and phenotype of the fetus.

X^Y Healthy

Extra Exercises

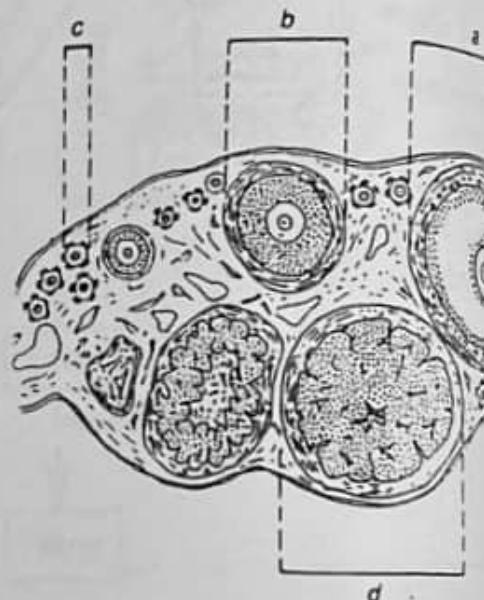
1. The figure below represents a cross-section of the seminiferous tubules of testicles, illustrating stages of spermatogenesis.



- Label the figure starting from № 1 → 10.

2. The adjacent illustration represents a longitudinal section of a female gonad observed using a photomicroscope.

- Give the name of every stage in the evolution of the ovarian follicle: a, b, c, and d.
- Indicate when does structure (a) become mature in a girl's life?
- Determine the origin of structure (d).



causes the activation of meiosis at puberty.

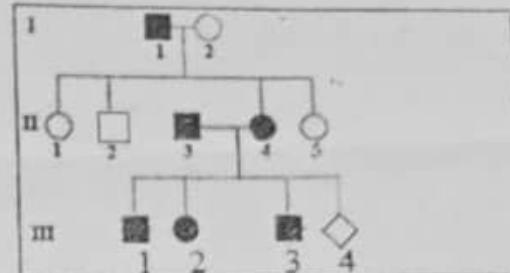
Exercise 3:6pts

Marfan syndrome

Marfan syndrome is a genetic disorder characterized by several symptoms affecting connective tissue. The genetic defect is in the FBN1 gene, which codes for the glycoprotein fibrillin-1 which is an important component of connective tissue, whose role is to provide organ support.

Document 1 shows the transmission of this disease in a family where some members are affected.

1. Show that the allele responsible for this disease is dominant.
2. Determine the chromosomal location of the gene responsible for Marfan disease.



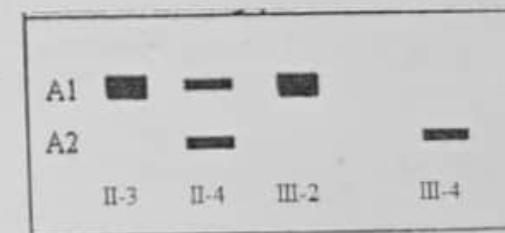
Document1

Document 2 shows a part of the nucleotide sequence of the non-transcribed strand of the two alleles of the FBN1 gene.

3. Identify the type of mutation that is the cause of marfan disease.

Normal allele	CCA CGT AAG TAT CAA TAG
	⁴²⁶ ₁₀₃
Mutant allele	CCA CGT AAG TAC AAT AG
	¹²⁶ ₁₄₂

Document 2



Document3



Document4

Document 3 represents the results of electrophoresis of the normal and mutant alleles of some family members.

4. Specify the band corresponding to the mutant allele.

At birth, child III4 is found to be normal for Marfan's disease; this result is unexpected.

Document 4 shows the karyotype of child III-4.

Knowing that the gene responsible for Marfan disease is carried by chromosome 15:

5. Explain based on documents 3 and 4 the normality of child III-4 concerning the Marfan disease.

4. Determine gene location

If the gene is located on non homologous part of Y then none of the girls would be affected but 2 and 4 are affected
 so it's not the case.

If the gene is located on non homologous part of X, then girl 9 which is normal has genotype $X^A X^A$. She takes X^A from each parent. Then her father's genotype must be $X^A Y$ but her father 3 is affected so it's not the case.

If the gene is located on homologous part of X and girl 3 is a normal girl her genotype $X^A X^A$ and she takes X^A from each parent and her brother 8 is an abnormal boy his genotype $X^a Y$ he takes X^a from his mother and Y from his father. Then the father's genotype must be $X^a Y^A$ (normal) but the father is affected so it's not the case. Therefore it's an X-linked trait.



5. Specify the band which corresponds to the mutated allele. Individual 5 is normal and normal allele is recessive so her genotype is $X^A X^A$ (written only in capitals) referring to Southern blot individual 8 has band A so band A corresponds to normal allele, thus band B corresponds to mutated allele.

Exercise 3

- 1) Chromosomal formula: (47; XX; +13) (%)
- 2) Free trisomy, since according to document 1 (karyotype) the extra chromosome 13 is free and not attached to another chromosome. (1)
- 3) Hypothesis : (translocated) trisomy 13 is inherited (genetically transmitted) in this family. (%)
- 4) Both karyotypes show 46 chromosomes, 44 autosomes and 2 gonosomes: XX in the woman's karyotype while XY in the husband's karyotype.
The husband's karyotype shows normal chromosomes, while woman's karyotype shows a missing 13 chromosome and a chromosome 14 longer than its similar (or a translocation of chromosome 13 on chromosome 14) (1%)
- 5) This woman has a normal phenotype since the genetic material is conserved although a translocation took place. (1)
- 6) Since the husband is normal and the woman shows a translocation in her karyotype, then any abnormality will be linked to the woman and not the husband; by focusing only on involved chromosomes 13 and 14; The possible gametes of this woman are :



From the above possible gametes 50% will lead to normal children (%)

Exercise 4: (5 pts)

A benefit of a chromosomal abnormality

Scientists have noticed that individuals with certain chromosomal abnormalities have a lower risk of developing certain types of cancer. In order to verify this idea, studies and experiments were carried out.

Study 1: A study of the partial karyotype (the chromosomes concerned by the studied chromosomal anomaly are only represented) is carried out in a normal individual and in two affected individuals A and B presenting the same phenotype, (Dawn syndrome). The results of this study are shown in document 1.

Karyotype	Chromosome 14	Chromosome 21
Normal		
Affected A		
Affected B		

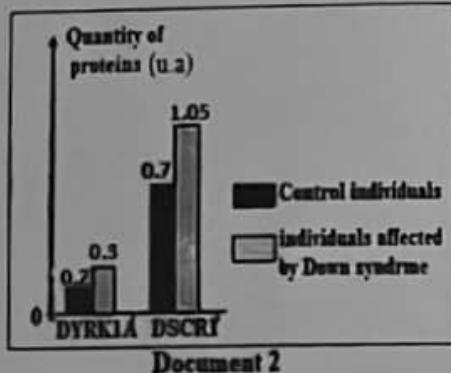
Document 1

- Justify why individuals A and B have the same phenotype.

Study 2: In order to study the effect of producing high quantities of two proteins coded by the two genes DYRK1A and DSCR1 located on chromosome 21 on the growth of tumor, 3 experiments are performed.

Experiment 1: the scientists measure the quantity of these two proteins. Document 2 shows the results in individuals with Dawn syndrome and in control individuals.

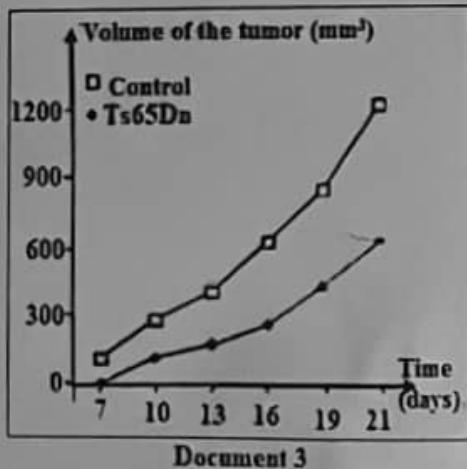
- Compare the results obtained in document 2.
- What can you conclude ?



Document 2

Experiment 2: Ts65Dn mice are animal models of Down syndrome and in particular possess 3 copies of each of the DYRK1A and DSCR1 genes. Cancer was artificially induced in these mice and control mice. The volume (growth) of the cancerous tumors is then measured for 3 weeks. Document 3 represents the obtained results.

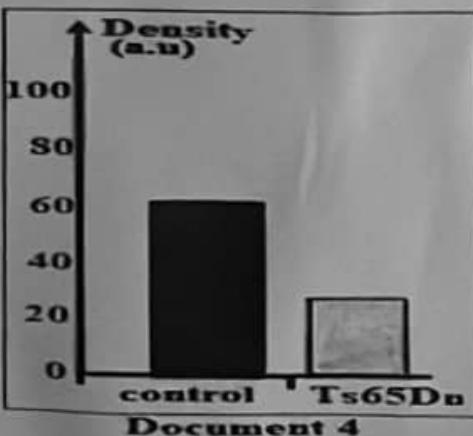
- Show, from document 3, that Dawn syndrome can delay (slow down) the growth of cancerous tumors.



Document 3

Experiment 3: In Ts65Dn mice and control mice, the density (growth) of blood vessels in artificially induced cancerous tumors is measured. It should be noted that blood vessels provide the oxygen and nutrients necessary for the multiplication and survival of cancerous tumor cells. Document 4 illustrates the obtained results.

- What can you draw out from results of document 4?
- Explain, based on all of the above, why individuals with Down syndrome have a lower risk of developing cancer.



Document 4

6. Determine the genotypes and phenotype of the fetus.
The fetus has 2 bands A corresponding to N allele and B for H1 allele so his genotype is Hn. H1 dominates n so his phenotype is EH

7. calculate the risk of the fetus to be affected. $P(\text{Affected}) = \frac{1}{5}$

$$P(\text{Aff}) = P(\text{mother is given}) \times P(\text{father is Hn}) \times P(\text{Pathogen})$$
$$= 1 \times \frac{1}{5} \times \frac{1}{2} = \frac{1}{10}$$

$$\text{P(Affected)} = 1 - P(\text{normal})$$
$$= 1 - \frac{9}{10} = \frac{1}{10}$$

Exercise 1: (10 pts)**Reproduction**

In order to study some aspects of the reproductive function in man, we propose the use of data represented in documents 1 and 2.

Document 1 represents schematically a section of a testis:

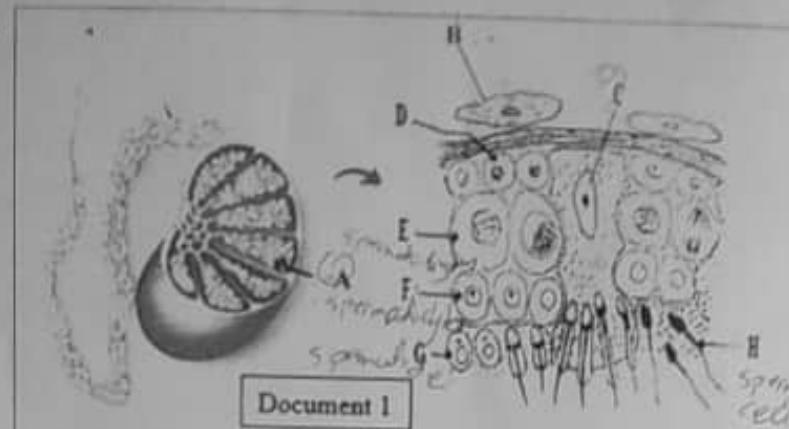
Q1 Name the process shown in document 1.

2) Label document 1 from **A** to **H**.

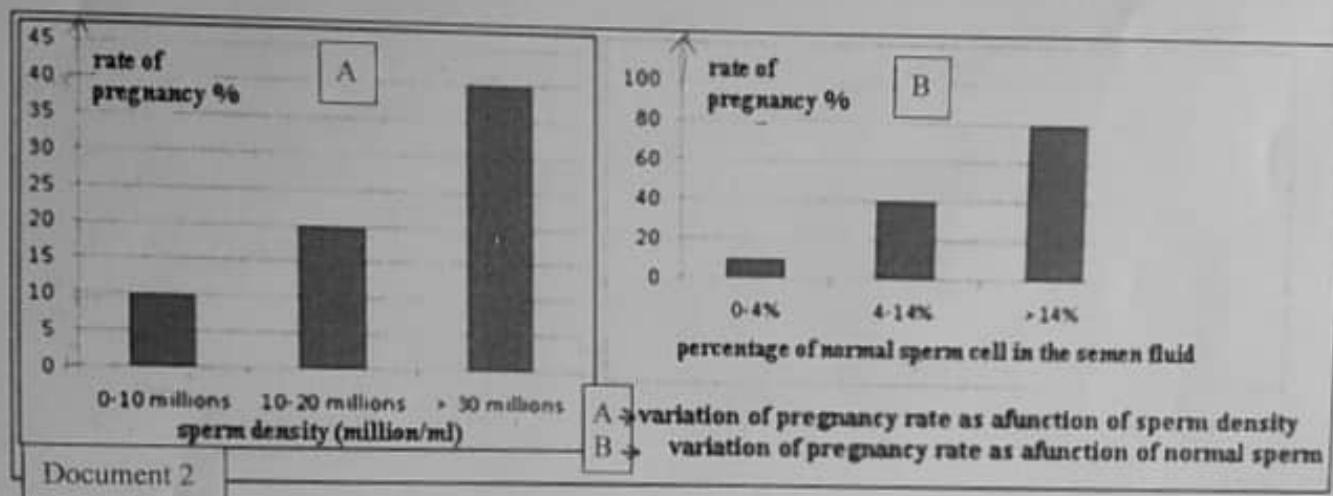
3) State the function of **B** and **C**.

- The structure **H** results from an important physiological phenomenon.

4) Name this phenomenon.



In document 2, two studies were done in the framework of studying the factors that affect male infertility. The male is considered fertile when the rate of pregnancy upon sexual contact is above 25%.



Q2 Transform document 2-A into a table.

Q3 What can you deduce from document 2-A?

7) Draw out from B a cause of infertility in men.

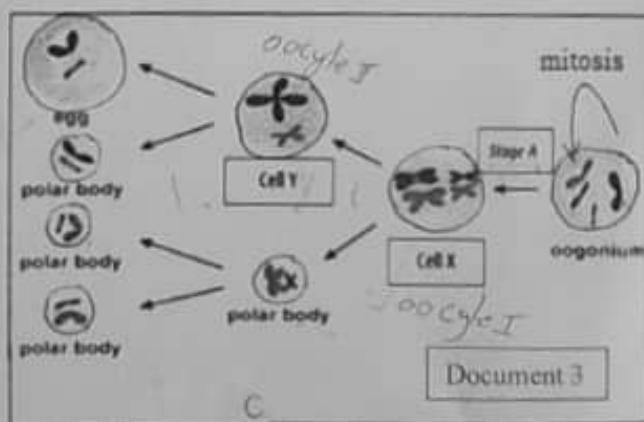
- The study of woman's gametogenesis resulted in establishing the following scheme in document 3 showing partial karyotypes.

8) Name the process shown in document 3

9) Name stage A, cell X, and Cell Y

10) Write the chromosomal formulas for a human cell shown at X and Y

11) Based on acquired knowledge and documents 1 and 3, differentiate between gametogenesis in males and females.



4. Determine the risk for to have affected children

To determine the risk to have an affected child of genotype d/d by a recessive disease:

Rule= risk of mother to be heterozygous \times risk of father to be heterozygous \times risk of the child to receive the allele of the disease from both heterozygous parents.

Risk of the parent to be heterozygous=

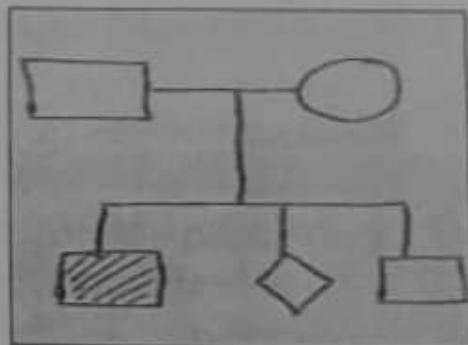
.2/3 if the parent has family history (affected brother or sister)

- Given in the population, if the parent has no family history (no affected siblings)

Risk of the child to receive the allele of the disease from both parents= $\frac{1}{2}$ from the mother and $\frac{1}{2}$ from the father.

Case 1: (couple I1-I2)

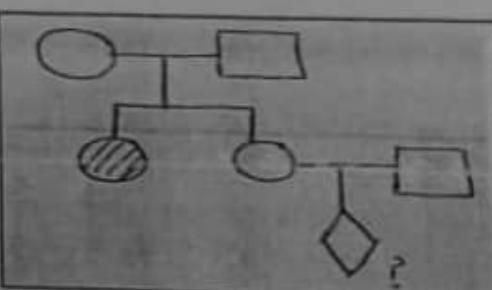
- Risk mother to be hybrid= Risk father to be hybrid =1 since they have affected child d/d that must inherit the diseased allele from each parent.
- Risk for child to inherit the diseased allele from each hybrid parent= $\frac{1}{2} \times \frac{1}{2} = \frac{1}{4}$
($\frac{1}{2}$ to take diseased allele from mother since $\frac{1}{2}$ to inherit the normal allele and $\frac{1}{2}$ to inherit the diseased allele)
($\frac{1}{2}$ to take diseased allele from father since $\frac{1}{2}$ to inherit normal allele and $\frac{1}{2}$ to inherit diseased allele)
- So, Risk= $1 \times 1 \times \frac{1}{4} = \frac{1}{4}$



Case 3: one of the parents has affected sister or brother (history) and the other parent has no history; couple II1-II2

- Risk for father to be hybrid = risk to be hybrid in a population = $1/20$ (since he has no family history)
- Risk for mother to be hybrid = $2/3$ (since parents of the mother are normal having the normal allele and they have affected daughter thus they are carriers for diseased allele as well).

Thus the mother of fetus may be N/N, N/d or d/d. So, risk to be hybrid is $2/3$.



- Risk for child to take diseased allele from each hybrid parent= $\frac{1}{2} \times \frac{1}{2} = \frac{1}{4}$

Amal Educational Institutes
School: Mostafa Shomran.

Name : *Mazen Ismail*
Date : Monday 24/10/2022.
Section: Life science "English section".

Time : 60 minutes.
Subject: Biology.
Class : Third secondary.

Teacher: Mazen Ismail

*Don't watch the clock.
Do what it does.
Keep going.*

Exercise I: (2½pts)

For each of the following items, there may be one or more correct answers. On your copy state the number of each item and the letter (s) corresponding to the correct answer (s).

- 1) In men, each cell that undergoes meiosis produces generally:
 - a. Four sperm cells.
 - b. Three sperm cells.
 - c. Two oocytes and two polar bodies.
 - d. An oocyte and two polar bodies.

- 2) The following document represents a calendar of the sexual cycle of a woman marking on it the date and the duration of menses.
 - a. The duration of the cycle is 29 days.
 - b. The duration of the cycle is 28 days.
 - c. The time of ovulation is November 12.
 - d. The time of ovulation is November 7.

October 2022							November 2022						
M	T	W	T	F	S	S	M	T	W	T	F	S	S
					1	2							
3	4	5	6	7	8	9	7	8	9	10	11	12	13
10	11	12	13	14	15	16	14	15	16	17	18	19	20
17	18	19	20	21	22	23	21	22	23	24	25	26	27
24	25	26	27	28	29	30	28	29	30	31			
31							29	30	31				

- 3) Oocyte II are:
 - a. Haploid cells.
 - b. They do not complete their division except after fertilization.
 - c. They contain double chromosomes.
 - d. Diploid cells.

- 4) Maturation is a step:
 - a. Of spermatogenesis that starts before birth.
 - b. Of spermatogenesis that starts at puberty.
 - c. That starts before birth in oogenesis.

- 5) The entry of sperm cells into oocyte II blocked at metaphase 2:
 - a. Triggers fertilization.
 - b. Results in the completion of meiosis of the oocyte that gives an ootid and a 2nd polar body.
 - c. Results in the completion of meiosis of the oocyte that gives an ootid and the 1st polar body.
 - d. Results in the completion of meiosis of the oocyte that gives a zygote and the 2nd polar body.

The sperm produced in the testes lack motility and the ability to fertilize. In order to determine the factors necessary to obtain a motile spermatozoon capable of fertilizing the oocyte, the following experiments are carried out:

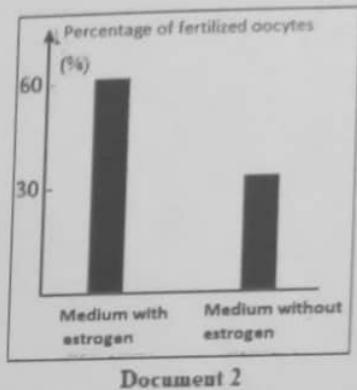
Experiment 1: Sperm isolated from the lumen of the seminiferous tubule are deposited at the cervix of a female rat at ovulation. The spermatozoa fail to move and do not reach oocyte II. ✗

Experiment 2: Sperm isolated from the epididymis are deposited at the cervix of a female rat at ovulation. The spermatozoa reach oocyte II. ✓

- Draw out a conclusion from experiments 1 and 2.

Experiment 3: spermatozoa isolated from the epididymis are cultured in vitro with oocytes in the presence and absence of estrogen (female hormones secreted by the ovary). The percentage of fertilized oocytes is measured and represented in document 2.

- Deduce the site of sperm capacitation.



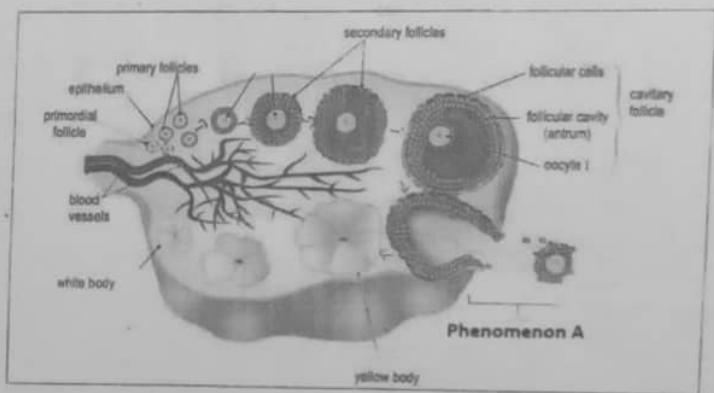
Document 2

Exercise 3: (6.5 pts)

Female sexual cycle

The ovary is the female gonad, it releases each cycle a cell, this cell is the result of significant cell division that occurs partially in the ovary. The figure in document 1 represents a schematic representation of the ovary at a point in the female sexual cycle.

- Name the phenomenon A.
Define it.
- Name the two cells in the figure that belong to oogenesis. Justify
- Schematize, at $2n = 6$, the cell division that occurs during the transformations observed in document 1.
- Explain the expression «occurs partially» mentioned in the text.
- Represent the karyotype of the laid cell at $2n=8$.
- What is the fate of the cell released by the ovary in the absence of spermatozoa?



Document 1

EXERCISE Trisomy 13

Trisomy 13 is a chromosomal abnormality due to the presence of an additional chromosome 13. It is characterized by the association of cerebral malformations (holoprosencephaly), facial dysmorphism, ocular abnormalities, postaxial polydactyly, visceral malformations (cardiopathy) and very severe psychomotor retardation. All of these symptoms are known as Patau's syndrome. This syndrome has a risk of 1/12 000 in the case of free trisomy 13 which is due to an error in the disjunction of chromosomes 13 during one of the meiotic divisions. About half of pregnancies with trisomy 13 result in abortion, 90% of children born with trisomy 13 die in their first year of life. Document 1 shows a karyotype of an individual with Patau's syndrome.

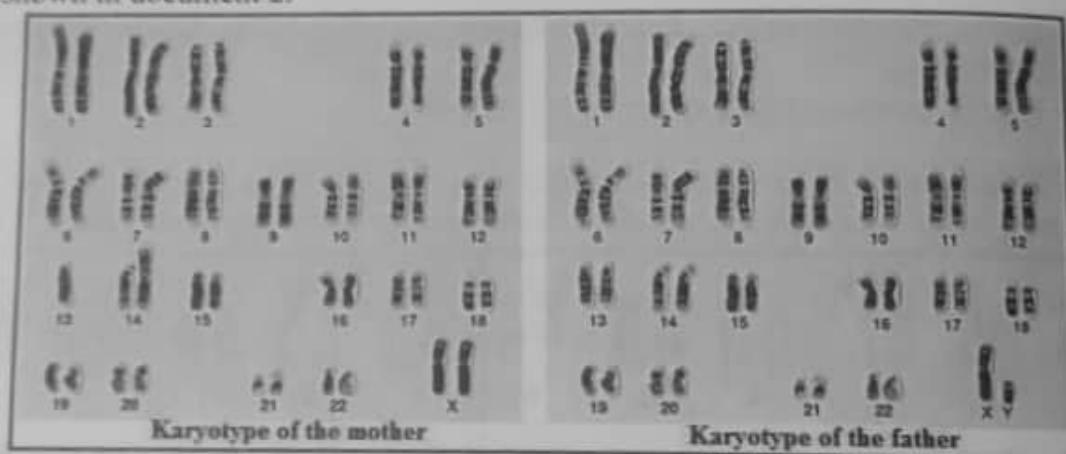
1. Write the chromosomal formula of the karyotype in document 1.
2. Specify whether this trisomy 13 is linked or free.

**Document 1**

One family suffers from multiple abortions and two cases of trisomy 13. A normal woman in this family has had an abortion of a fetus with trisomy 13.

3. Formulate a hypothesis about the origin of trisomy 13 in this family.

In order to determine the cause of the repeated abortions of this woman's family and to clarify the origin of trisomy 13, a doctor decides to realize the karyotype of this woman and her husband. These karyotypes are shown in document 2.

**Document 2**

1. Compare the karyotype of the mother to that of her husband.
2. Explain the normal phenotype of this woman.
3. Schematize, by limiting to chromosomes 13 and 14 in the woman, the anaphase that is at the origin of the gamete leading to abortion.

This woman decides to have another child and she wonders about the chance she has to have a normal child and the risk for this normal child to face the same difficulties of having normal children. Considering that, in the woman, normal chromosome 14 separates from abnormal chromosome 14:

4. Schematize, considering the chromosomes 13 and 14 only, the different gametes produced by this woman while indicating their proportions.
5. Determine:
 - 3.1. The chance for this woman to have a normal child.
 - 3.2. The risk for this woman's normal child to have a problem similar to hers.

e.g. Huntington disease

given : uncoordinated involuntary movements

- physiological problems

origin of disease is mutation.

1. Pickout from the text

- a. origin of disease : origin of disease is mutation
- b. characteristics : uncoordinated involuntary movements
- physiological problems

given Age (years) 10 30 40 60 70

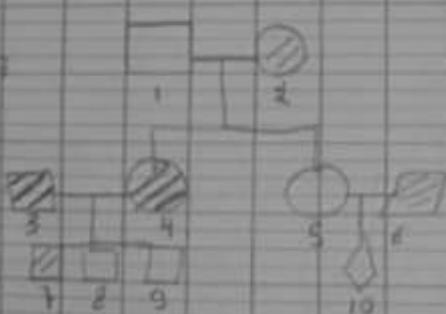
% of individuals 0 30 60 90 100

Showing symptoms
%

2. Interpret the obtained Results (Analysis + Significance)

The % of individuals showing symptoms increases, this means symptoms increase from 0% to 100% as age reaches 70 years. This shows that age favours symptoms of Huntington.

Given:



3. Indicate the dominant and recessive alleles Justify your answer.

Huntington's allele is dominant symbolized by "H" and normal is recessive symbolized by "n"

since 3+4 are diseased couples they gave 8 and 9 normal children
normal allele is masked by diseased allele in parents.

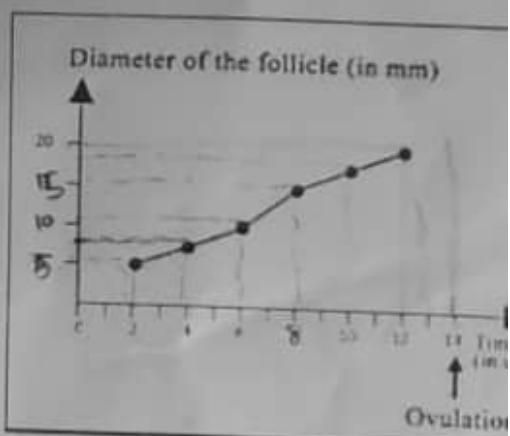
Exercise 1

Female sterility

Mrs. X is a female who has been married for 10 years and suffers from the absence of pregnancy. The doctor suggested that there is a problem at the level of her follicles.

Document 1 shows the development of the diameter of the follicle in a control female body.

1. Name the follicles starting from the smallest to the most mature.
2. Pick out from document 1 the diameter of the follicle necessary for ovulation.
3. Transform the graph of document 1 into a table that shows the development of the diameter of the follicle in mm as a function of time in days.
4. Explain what happens to the oocyte and the ruptured follicle after ovulation if fertilization takes place



Document 2 shows the diameter of the oocyte in the ovary of Mrs. X (A) and in a control human female (B) at day 14.

5. Compare the diameters of the 2 follicles shown in document 2.
6. Explain from what precedes the sterility of Mrs. X.





Grade:12

Basic mechanisms of sexual reproduction

Name: _____

Biology

Duration : 90 min

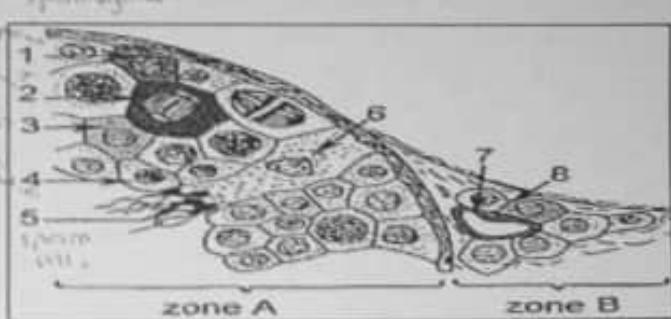
Exercise 1:

Reproductive functions in human species (8 points)

A study is done on certain aspects of reproductive functions in human species.

Document 1 represents a simplified cross section in a testis of a human male.

- ✓ Label the two represented zones. (1pt)
- ✓ Annotate 1 to 5 then 7. (0.5pt)
- ✓ 1. Name the process that allows the transformation from structure 1 to 5. (0.5pt)
- ✓ 2. Indicate the number of chromosome and chromatids for structures 2,3 and 4. (1.5pt)



Document 1

Two experiments were done to determine the role of certain cells within the testicle (document 2).

- ✓ Specify each of the cells 6 and 8. (1pt)

Exp.1	Destruction of cell 6	Cell 1 couldn't change into cell 5
Exp.2	Destruction of cell 8	Lack of testosterone secretion

Document 2

To confirm the role of the testicles in reproductive function, experiments were realized on two lots of mice: lot-1 and lot-2. The experiments and their results are summarized in Document 3:

Lot	Experiments	Results
1	Zone- A was destroyed by X-rays	Sterility + Maintenance of the secondary sexual characteristics.
2	Zone- B was destroyed by X-rays	Sterility + Regression of the secondary sexual characteristics

Document 3

- 5- Interpret the obtained results. (1.5pt)
- 6- Using the preceded given and your acquired knowledge, give two possible causes of sterility in men. (1pt)

Exercise 2:

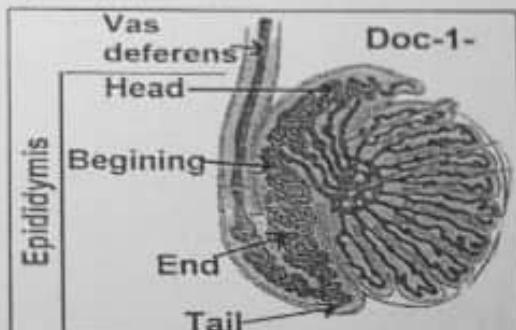
Function of the epididymis (6 points)

In the seminiferous tubules of the testicles of a sheep, sperm cells are immobile.

During their transmission along the regions of the epididymis, three types of sperm cells are observed with different aspects of movement, as shown in document 1.

Type a : The flagellum of the sperm cells oscillates without any displacement (change of place).

Type b : The movement of the sperm cells is disordered (sperm cell turns around itself).



Document 1

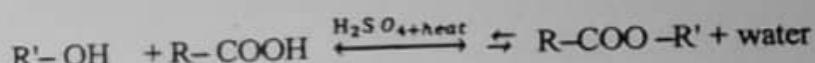
Exercise 9
Equilibrium in Esterification reaction

Given

Organic compound	Molar mass in g/mol ⁻¹	Density in g/ml ⁻¹
Alcohol (A) is 2° : secondary	74	0.81
Carboxylic acid (B)	88	1.05
Ester (E)	144	-

1- The Esterification reaction:

The manufacture of ester (E) is done by heating 50 ml of alcohol (A) with a volume V of carboxylic acid (B) in presence of sulfuric acid solution (H_2SO_4). At the end of the reaction, it is found that the yield of reaction is 60 %. the following equation of the esterification reaction is given:



- 1.1- Specify the role of sulfuric acid solution
- 1.2- Determine the volume V of carboxylic acid (B).
- 1.3- Calculate the mass of ester (E) formed (actually).
- 1.4- Calculate the value of K_c .

2- Another experiment:

The experiment is repeated with 40 ml of alcohol (A) and V ml of carboxylic acid at the same temperature. The yield of the esterification reaction is now 0.8

- 2.1- specify whether the mixture of reactants is equimolar or not.
- 2.2-Determine the number of moles of carboxylic acid. Knowing that alcohol A is the limiting reactant
- 2.3- Deduce the volume V of carboxylic acid taken initially.

Answers :

arts	Expected answers	Pts.
1.1-	H_2SO_4 :Sulfuric acid is used as a catalyst (when it is used in small quantity) that speeds up the reaction without being a reactant or that appears at the end of the reaction chemically unchanged.	
1.2-	<p>Imp : → Since the alcohol used is secondary and the yield is 60 %, hence the mixture of reactants must be equimolar (same number of moles).</p> $n_o(\text{Alcohol}) = n_o(\text{Acid}) = n$ $n_o(\text{alcohol}) = \frac{m(\text{alcohol})}{M(\text{alcohol})} = \frac{\rho \times V(\text{alco})}{M} = \frac{0.81\text{g}}{\text{ml}} \times \frac{50\text{ml}}{74\text{g/mol}} = 0.55 \text{ mol}$ <p>the mixture of reactants is equimolar \Rightarrow</p> <p>thus $n_o(\text{acid}) = 0.55 \text{ mol} \Rightarrow m(\text{acid}) = n \times M(\text{acid}) = 0.55 \times 88 = 48.4 \text{ grams}$</p> $V(\text{acid}) = \frac{m(\text{acid})}{\rho(\text{acid})} = \frac{48.4}{1.05} = 46 \text{ ml} .$	
1.3-	<p>$m(\text{ester})$ actually obtained =?</p> $\% \text{ yield} = \frac{m(\text{ester}) \text{ actually}}{m(\text{ester}) \text{ theoretically}} \times 100$ <p>yield = 60% is given .</p> <p>Mass of ester theoretical is calculated as such:</p> <p>Assume that the reaction is complete \Rightarrow</p> $R'-OH + R-COOH \longrightarrow R-COO-R' + \text{water}$	