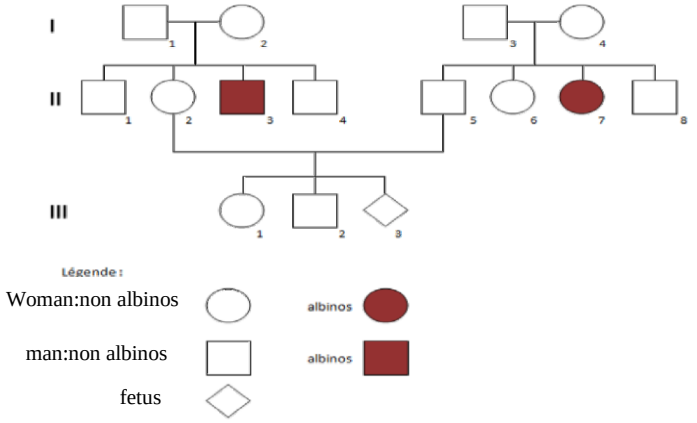
Class: 3 rd. Secondary (L. S.) **Mid-Year Exam** **Points:** 20 pts.
Subject: Biology **Duration:** 180 min.

Exercice I : Transmission of a hereditary character, the albinisme (6.5 pts)

In human, the albinism is a rare genetic disease (1 case on 20000 births). The albinism is due to the absence of melanin, a brown pigment responsible for the coloration of skin and hair.

Document 1 : Perigree of a family affected by albinism



1- Determine the location and the mode of transmission of the studied gene.
2- Can we talk about a family at risk concerning the couple II-2 and II-5? Justify your answer.
3- Indicate the genotypes of the members of the family in generations I and II.
4- Evaluate te risk for the fetus III-3 to be affected by this disease.
5- Compare this risk to the general risk of this disease. To what do you attribute this difference?

Document 2 :

	Enzyme Xho II		Enzyme Xba I	
	Number of sites	Length of fragments (pb)	Number of sites	Length of fragments (pb)
TYRCOD 1	2	172, 283, 1135	0	1590
TYRCOD 2	3	172, 283, 564, 571	0	1590
TYRALBA 3	3	172, 283, 564, 571	1	530, 1060

Document 2a: Identification of the alleles of tyrosinase gene by two restriction enzymes, Xho II and XbaI.
In this family, the albinism is due to a gene found on an autosome ,it has 3 alleles:
- 2 alleles TYRCOD 1 and TYRCOD 2 that code for functional tyrosinase,
- 1 recessive allele TYRALBA 3 that codes for a non functional tyrosinase.

6- Compare the effects of the enzymes Xho II and Xba I on the 3 alleles in documents 3a and 3b and deduce the cause of the use of enzyme Xba I.

Document 2b: schematic representation of the alleles and the restriction sites of enzymes Xho II and XbaI

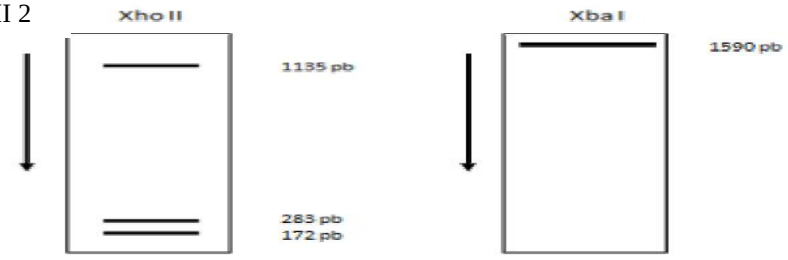
DNA of allele TYRCOD 1

DNA of allele TYRCOD 2

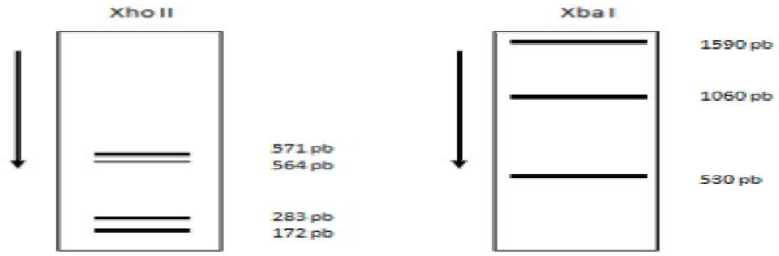
DNA of allele TYRALBA3

Document 3 : Separation by electrophoresis of the fragments obtained after the action of restriction enzymes on the gene of tyrosinase in parents II.2 and II.5

Mother II 2



Father II 5



Légende :

Migration sens

7- Interpret the document 3 and deduce the real genotypes of parents II2 and II5.

8- Does the risk of the fetus III-3 change after this genetic diagnosis? Justify your answer.

9- What are the advantages of the new techniques of biotechnology used in this diagnosis?

Exercice II – The effect of irradiation on genetic information. (3.5 pts)

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

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

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

1st Cross	Individuals Female [+] × male [z] homozygote	Results of cross 50 % males [+] 50 % females [+]
2nd Cross	Femelle [z] × male [+] homozygote	50% males [z] 50 % females [+]

3rd Cross	Femelle [z] × male [+] Homozygote, irradiated Non- irradiated with X rays	50 % males [z] 50% : - females [+] -rare females [z]

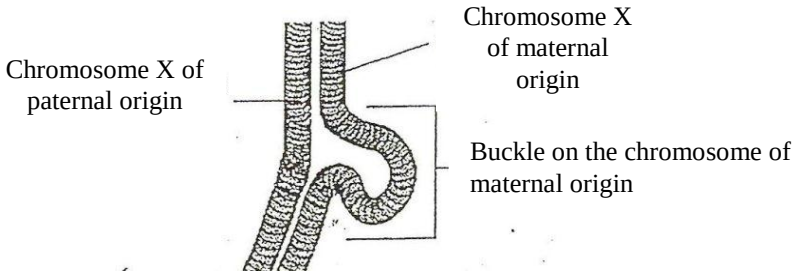
1- Based on document 1, determine the mode of transmission and the localization of the gene of the character color of eyes in drosophilae.

2- Formulate a hypothesis which explains the appearance of the phenotype « zest » in certain females of the 3rd cross.

Document 2: Consequences of the irradiation

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

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



3- Use the document 2 to confirm or infirm the hypothesis proposed in question 2.

4- Draw the pair of chromosomes implicated in the appearance of the phenotype « color of eyes » in drosophilae of each of the 3 descendants of the 3rd cross.

Exercice III: Chromosomal abnormalities (6.5 pts)

A- The following documents show different chromosomal abnormalities and experiments on the mice development.

Document 1: chromosomal abnormalities and sex determining

Sexual Chromosomes	XX	XY
Phenotypes	Boy	Girl
Gonads	Normal Testicles producing male hormones	Ovaries
Abnormality	A chromosome X carrying a fragment of short arm of chromosome Y	Chromosome Ywith absent short arm

1- Analyze the document 1 and deduce the role of the short arm of chromosome Y.

2- Formulate a hypothesis that explains how the short arm of chromosome Y performs its function.

In order to determine the genetic cause of these abnormalities, the following experiments of document 2 are done.

Document 2: Experiments on the development of mice

Experiments	**Results**
Deletion of short arm of chromosome Y in a mouse egg and injection of gene Sry	Development of a male mouse
Injection of gene Sry, in an egg cell of a mouse having the sexual chromosomes XX	Development of a male mouse

3- Do thes experiments validate your hypothesis ? Justify the answer.

We find certain individuals suffering from a disease called gonadic dysgenesis. They present a female phenotype and their karyotype reveals 46 chromosomes in which XY are the sex ones; their gonads are undifferentiated. The document 3 shows the peptide sequences of the protein coded by the gene Sry:

- In a normal individual (sequence A)
- In an individual affected by gonadic dysgenesis (sequence B).

Document 3 : The peptide sequences of the protein coded By the gene Sry in the two individuals

1 5 10 15 20 25

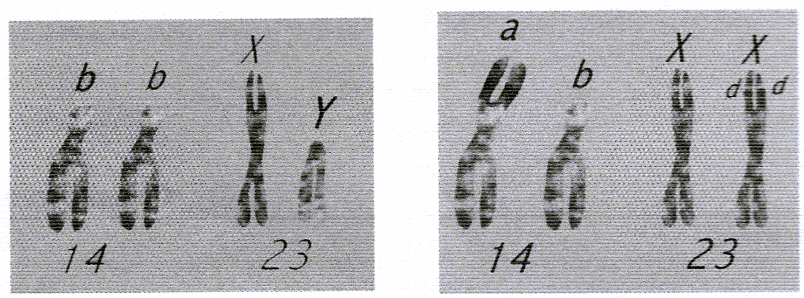
A MetGlnAspArgValLysArgProMetAsnAlaPheIleValTrpSerArgAspGlnArgArgLysMetAlaLeuGluAsn

B MetGlnAspArgValLysArgProIleAsnAlaPheIleValTrpSerArgAspGlnArgArgLysMetAlaLeuGluAsn

4- Compare the two peptide sequences.
5- Indicate the genetic cause of this difference in the protein of the affected individual.
6- Explain the origin of this disease in the affected individual in spite of the presence of a complete chromosome Y.

B- Chromosomal Translocation

The documents 1 and 2 are taken from the karyotypes of two parents having normal phenotype. One of them has a chromosomal aberration and he is carrier of a gonosomal disease, the daltonism.

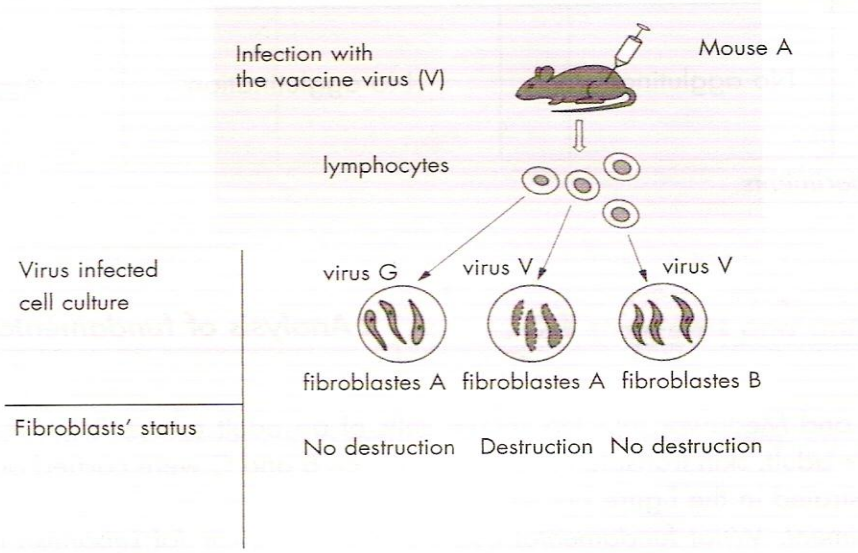


document(1) document(2)

1- Identify the individual having these two abnormalities.
2- The chromosomal aberration found in this case is autosomal or gonosomal ? Justify your answer.
3- Determine the parental gametes and show by a punnet square the chromosomic recombination during fertilization.

Exercice IV : The double recognition by lymphocytes (3.5 pts)

A mouse (A) is injected by a vaccine virus (V). Ten days later, lymph nodes are removed and lymphocytes are extracted. These lymphocytes are put in vitro with fibroblast cells infected by the le virus V or by another virus G, these cells are from mouse A or from another mouse B. The experimental procedure and the results are shown in the following figure (from an experiment of Zinkernagel).



1- Describe the above experiments.
2- Interpret thes experiments, what can you deduce concerning the conditions of lymphocytes activity?
3- What is the type of the lymphocytes involved in this reaction ? Justify the answer.

Good Work