Class: 3rd Secondary/Life Science **Duration: 3 hours**
Subject: Biology **Mark: 20 points**

Mid-Year Exam

I-A : The Conditions of spermatozoïdes action: (5 pts)

The spermatozoon is a very differentiated cell able to fertilize an ovule under precise conditions. With an aim of finding these conditions, one carried out the following experiments:
E_1: Spermatozoa are removed from the somniferous tubules are introduced into the uterus. No significant migration towards the fallopian tube is observed.

E_2: The sperm taken starting from the vas deferens canal is introduced into the uterus; fertilization is normal.

E_3: The sperm taken from the vas deferens canal is put in a test tube in the presence of ova.. No fertilization was possible.

E_4: The injection of a spermatozoon in an oocyte leads to a fertilization. The acrosome remains intact, whereas it normally opens in contact with the oocyte.

$E_{5:}$ We separate active spermatozoa from the remaining sperm, before placing them in a physiological solution deprived of fructose. The spermatozoa become immobile and incapable of fertilization.

1- Interpret these experiments and deduce the conditions of spermatozoids action.

-B: Genetic and phonotypic sex

The following table shows different chromosomic charts as well as the corresponding phenotypes which they are female or male. (A= autosomal chromosomes)

	Female phenotype	Male phenotype
chromosomic charts	Document 1 44A+XX	Document 3 44A+XY
	Document 2 44A+X	Document 4 44A+XXY
	Document 5 44A+XY (without gene SRY)	Document 6 44A+XX (presence of gene SRY)

1- Compare documents 1 and 2, as well as documents 1 and 4. What can deduce with regard to the differentiation towards the male or female phenotype.

2- Interpret the results of documents 5 and 6.What can we deduce about the gene SRY being normally on chromosome Y.

3-Formulate a hypothesis explaining the presence of chromosomic charts of documents 5 and 6 of certain individuals**.**

II: Identification of the disease of albinism (5 pts)

Albinism is a hereditary deficiency characterized by the absence of pigmentation in the skin, eyes, and hair due to the absence of a black pigment called melanin. Tyrosinase is an enzyme involved in many of the chain reactions responsible for the biosynthesis of this pigment.

This disease is rare, recessive, autosomal and affects $^{1}/_{20.000}$ of new born world wide. A study performed on 800 children selected from an isolated community, showed that 20 children were heterozygous for albinism.

1. Calculate the proportion of heterozygous children in this community; and then determine the genetic risk for a child to be affected with albinism.

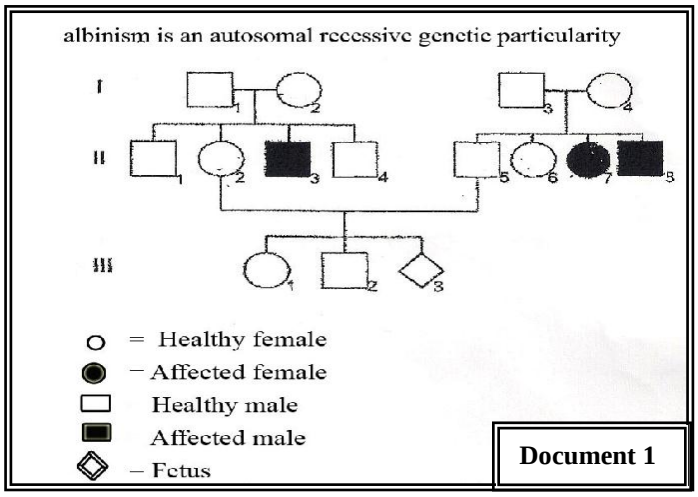
2. Compare the genetic risk obtained to the world wide risk. Formulate a hypothesis that explains the difference between these two risks.

Document 1 reveals the pedigree of family presenting albinism.

3. By referring to the pedigree, justify that the disease is recessive and autosomal.

4. Indicate the genotype of the individuals II_2, II_3 and II_5. Justify your reasoning.

5. Determine the genetic risk for the couple II_2 and II_5 to have and affected fetus.



Document 1

The gene that codes for the synthesis of tyrosinase enzyme exits in many alleles. Three alleles are considered: Two alleles **TYRCOD1** and **TYRCOD2** that code for the synthesis of functional tyrosinase that leads to the synthesis of melanin. One recessive allele **TYRCOD3** that codes for the synthesis of an inactive tyrosinase that does not lead to the synthesis of melanin.

Document 2 shows the identification of the alleles of the gene tyrosinase by two restriction enzymes Xho II and Xba I.

	Enzyme Xho II		**Enzyme Xba I**	
	Number of side	**Fragment length (pb)**	**Number of side**	**Fragment length (pb)**
TYRCOD1 (T_1)	2	172,283,1135	0	1590
TYRCOD2 (T_2)	3	172,283,564,571	0	1590
TYRCOD3 (T_3)	3	172,283,564,571	1	530,1060
Pb = base pair				

Document 2

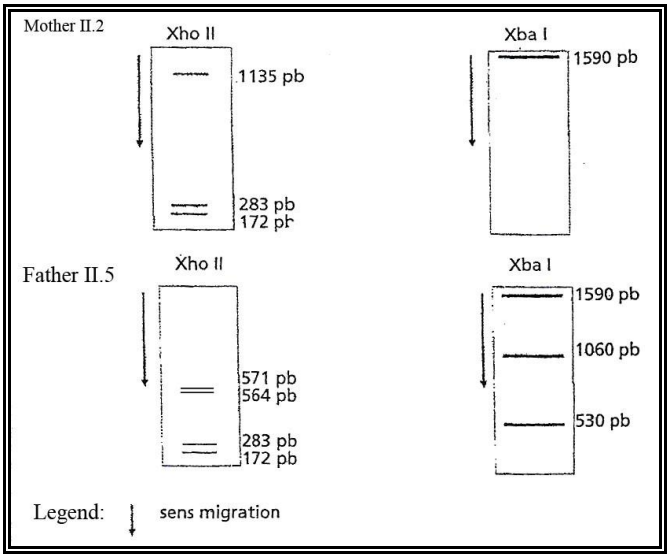
6. Pose the problem that arises from the use of the enzyme Xho II.

7. Interpret document 2 verifying that the posed problem is solved.

Document 3 reveals the results of get electrophoreses, done by Southern Blot, of the parents II_2 and II_5.

8. Explain the result of document 3 specify the real genotype of each parent II_2 and II_5.

9. Identity the phenotype of the fetus III_3.



Document 3

III: Selection of Pigs (5 pts)

In the breeding of pig, a gene responsible for the increased sensitivity to the stress and which also influences the quality of the meat was identified. This gene exists in two forms: the allele N and the allele n.

The table of document 1, shows the genotypes and the phenotypes of three kinds of pigs:

genotype	Sensitivity to the stress	Quality of the meat
NN	Weak	Good
Nn	Weak	Very good
nn	Very strong (important mortality)	Bad

Document1

1- Referring to the table of document 1, answer the following questions:

a. Show that the allele N is dominant.

b. Which genotype of pigs, is recommended ? Justify.

To obtain pigs of recommended genotype, one carries out the two following crossings;

Crossing 1: Pigs of genotype NN are crossed with pigs of genotype nn (NNxnn).

Crossing 2: Pigs of genotypes Nn are crossed with pigs of genotype Nn (NnxNn).

2- Make the factorial analysis and give the results of each crossing.

3- Identify the crossing which makes it possible to obtain pigs of quality breeding.

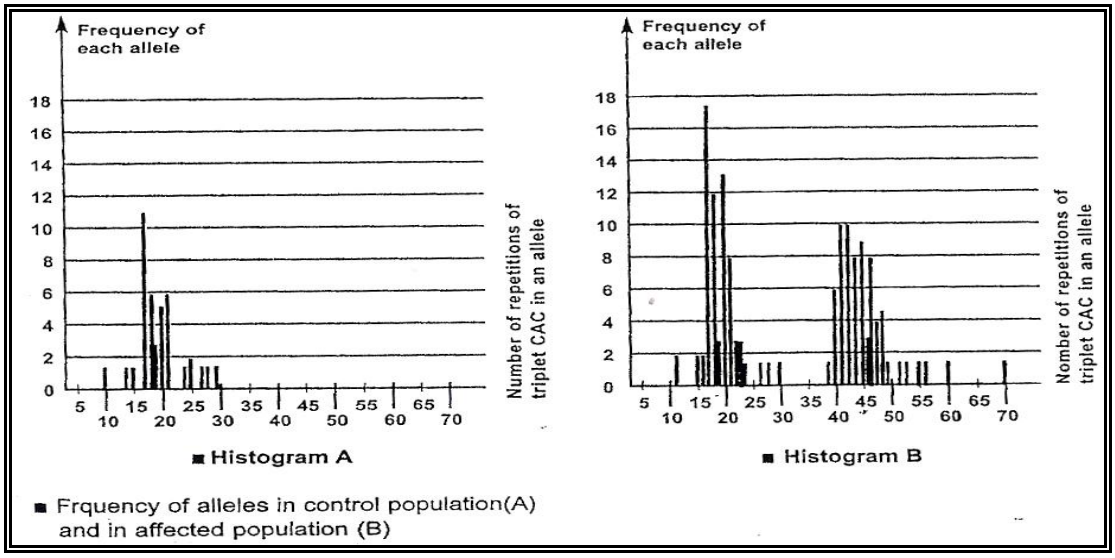
IV : Huntington chorea and its origin (5pts)

Huntington chorea is a dominant genetic disease which appears by disorders of nervous activity.

The gene (LT15) whose certain alleles are the origin of huntigon chorea posseses a region formed by the repetition of triplet CAG(non transcribed strand)CAG-CAG-CAG-CAG…..CAG(n).The gene alleles differ by the number of triplet repetitions..

Document 1:

show the frequency of the alleles in a controlled population (A) and in an affected population (B)

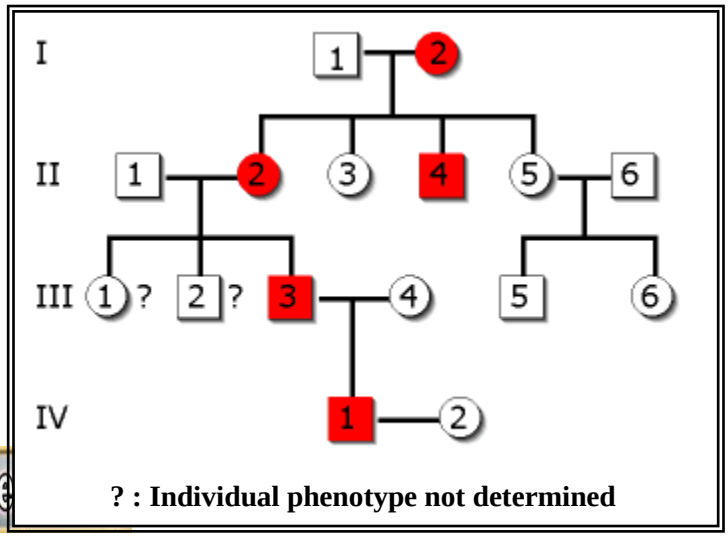


Document1

1- Compare the number of repetition of triplet CAG in the alleles of population A and B. What can you deduce about the origin of the disease of the population B.?

2- Formulate a hypothesis concerning the cause of the variation of the number of repetition in the two populations.

Document 2: This document represents the family tree of a family affected by Huntington chorea.



Document 2

3- Formulate a hypothesis concerning the gene localization responsible for the disease.

Document 3 : Show the alleles of gene (LT_{15}) of some individuals of the preceding family tree

	A number of repetitions of triplet CAG in the allele
Allele 1 of individual I_1	10
Allele 2 of individual I_1	15
Allele 1 of individual I_2	50
Allele 2 of individual I_2	20
Allele 1 of individual III_3	45
Allele 2 of individual III_3	25
Allele 1 of individual III_4	20
Allele 2 of individual III_4	20

Document 3

4- Construct a histogram showing the variation of the number of repetition of triplet CAG according to the alleles of the various individuals in document 3.

Document 4: Recent techniques make it possible to determine the nature and the number of alleles starting from the analysis of chromosome to a chromatid in the diploïd cells.

The following table shows the obtained results concerning the individuals of document 3. .

Individuals	I_1	I_2	III_3	III_4
A number of normal alleles	2	1	1	2
A number of affected alleles	0	1	1	0

Document 4

5 -Use the results of this table to test the hypothesis of question 2.

6- Explain how this table also allows to determine that the allele of this disease is dominant.

7- Write the genotypes of individuals I_1, I_2, III_3, III_4.

8- Write the genotypes of couple III_5 and III_6 in document 2.Calculate the probability for this couple to have a sick child.