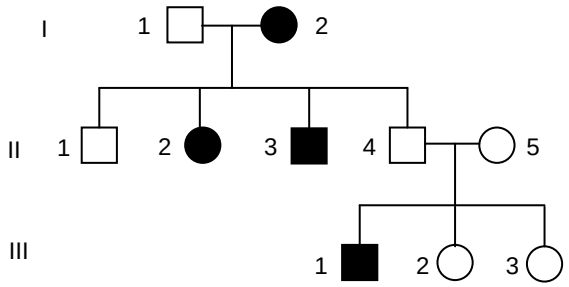
Class: 3 rd. Secondary (L. S.) | **Mid-Year Exam** | **Points:** 20 pts.
Subject: Biology | **Duration:** 180 min.

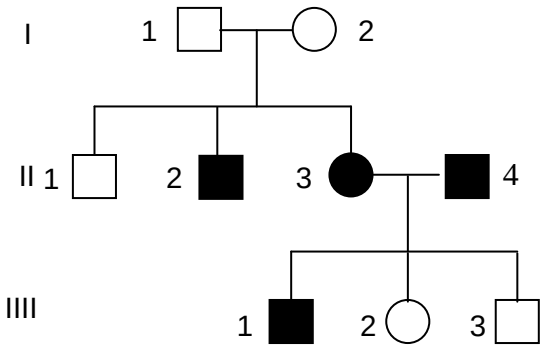
Exercise I: The albinism and its causes. **(4.5 pts.)**

The albinism results from a general lack of pigmentation: white hair, clear skin, and non-pigmented iris. The following pedigree represents two families A and B whose certain members are albinos.

Family A



Family B



1- From the **family A**, find the mode of transmission of albinism and the localization of the gene responsible of this abnormality.

We estimate that in human population, 1% of the individuals are heterozygote for the character of albinism found in the family A.

2- What is the risk for two normal individuals to have an albino child?

3- What is the risk for the couple II-4 and II-5 in the family A to have another affected child?

4- The man II-1 from the family A married a normal woman, what is the risk to have an albino child?

5- In the family B, all children are legal, what is the proposed problem upon analyzing the pedigree of the family B?

In order to solve the proposed problem, we study the synthesis of melanin pigment. It results from two successive reactions that occur in the cytoplasm of the cell:

Precursor P $\xrightarrow[E_1]{}$ tyrosine $\xrightarrow[E2]{}$ melanin (dark brown)

E_1: enzyme 1
E_2: enzyme 2

The analysis of hair roots of individuals II-3 and II-4 of **family B** shows the normal presence of precursor P .Immerse their hair roots on a tyrosine solution. The hair roots of II-3 will be dark and the analysis proves the presence of melanin. On the contrary, the hair roots of II-4 remain colorless.

6- Analyze this information and deduce the cause of albinism of individuals II-3 and II-4 of the family B.

The synthesis of enzyme E_1 is controlled by 2 alleles: allele (A) that triggers the synthesis of this enzyme. A is dominant over the recessive allele (a) that prevents this synthesis, while the synthesis of enzyme E_2 is controlled by 2 alleles: allele (B) that allows the synthesis of this enzyme and which is dominant over the recessive allele (b) that prevents this synthesis. The two couples of alleles: Aa and Bb are carried by two different chromosomes.

7- From the above information, and by taking into consideration the two genes: Aa of enzyme E1 and Bb of enzyme E2, find the possible genotypes of individuals II-3 and II-4 of family B.

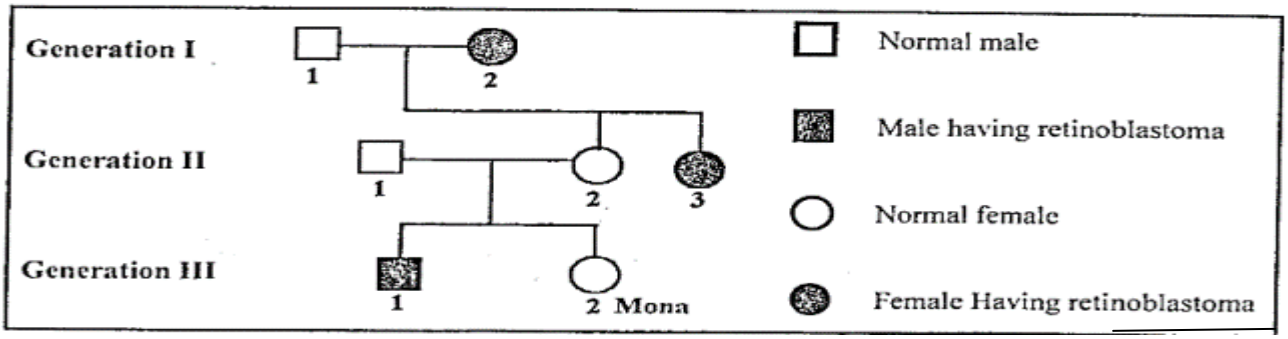
Exercise II:Retinoblastoma and its genetic origin. **(6.5 pts.)**

A- Retinoblastoma is a kind of cancer characterized by the appearance of a tumor in the retina of the eye. Normal individuals have a gene on chromosome 13 that suppresses (stops) cancer by inhibiting mitosis and stopping the rapid increase of cells and thus preventing the appearance of the tumor. This gene, in babies and infants, may be subject to two events that result in this type of cancer. These two events are:

- A point mutation of substitution hitting the allele of the suppressor gene.
- The deletion of a part of chromosome 13 at the locus of the suppressor gene.

- Concerning the first event of the mutant allele, two mutant alleles of the suppressor gene must be present in an individual to have a cancer. Moreover, this mutant allele is transmitted by germ cells to zygotes, thus, the presence of a mutant allele in a family indicates a risk to develop a cancer.

- Concerning the deleted allele, it occurs in somatic cells. The deletion of the allele in an individual results in developing cancer.Document 1 shows the pedigree of the family of Mona (III-2) whose some members are affected by retinoblastoma.



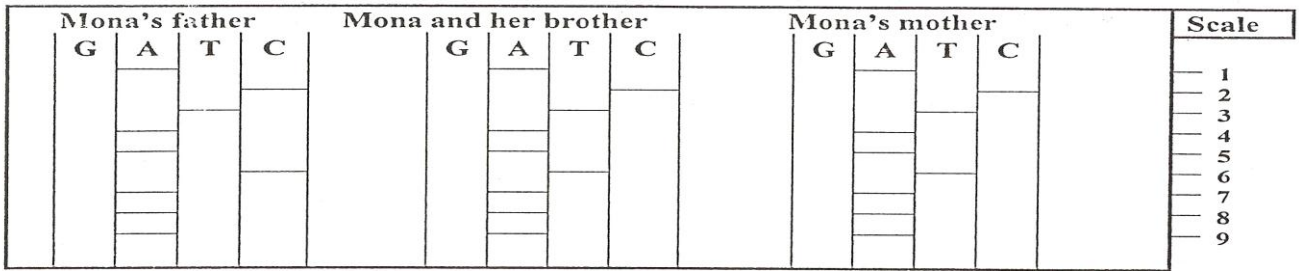
Document 1: The pedigree of Mona'family.

The father of Mona (II-1) is normal for both alleles of cancer suppressor gene and do not show any of the two mutational events and he is from outside of the Mona's mother family.

In the frameworks of knowing whether Mona has a risk to develop cancer, the following genetic methods are applied.

First method:

In order to determine the presence of the substitution in the mutant allele of the suppressor gene in Mona, her brother and her parents, a special technique is realized (Sanger technique) by which we can determine the nitrogenous base sequence of DNA of the normal and the mutant allele allowing to specify the substitution:



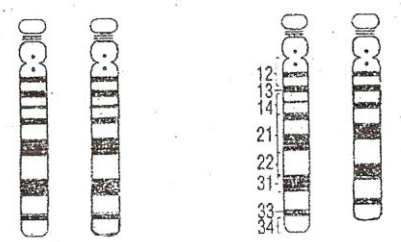
Document 2

Note: Sanger method can indicate the arrangement of nitrogenous bases depending on special reading method in which the N-bases are sequenced from 1 to 9.

1- Write the sequence of nitrogenous base for the allele of each of Mona, her brother and her parents.
2- Interpret the results of Sanger technique.

Second method:

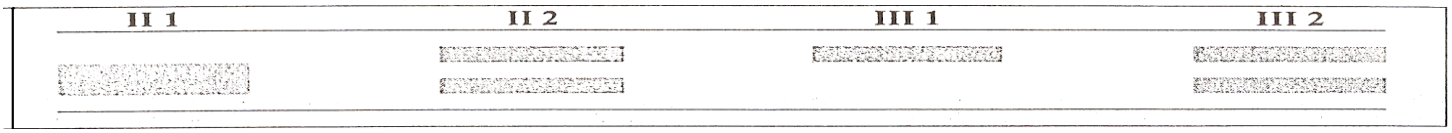
It involves the analysis of chromosome 13 (karyotype) with the different loci (dark bands) Mona's father and brother.

Chromosomes 13 of the father  Chromosomes 13 of the brother

Document 3

3- Interpret the karyotype of chromosome 13 of Mona's father and brother revealed in document 3.
4- Formulate a hypothesis that explains the cause of cancer in Mona's brother, based on documents 2 and 3.

Third method: Southern blot technique is applied after gel electrophoresis to compare the restriction fragments of the sequences of nitrogenous bases of the cancer suppressor alleles on the chromosome 13.The result is shown in document 4.



Document 4

5- Interpret the results of document 4.Do the result confirm your hypothesis? Justify the answer.
6- Does Mona have a risk of cancer? Justify your answer by using document 4.
7- Indicate the advantage of this technique.

B- Other applications of The DNA analysis: the paternity test.

This method is used in paternity test, it needs many days but it allows sharp answers: it eliminates the relationship or it leads to a strong probability of parental relation (bigger than 99%).

In fact, each individual has in his chromosomes DNA portions that code for protein synthesis and others that do not have a discovered role. Between these non-coding DNA, certain sequences of nucleotides link up repeated to the identical. These repetitive sequences are called micro satellites. The size of the micro satellite corresponds to the number of repetition in the DNA sequences; this number is variable from one individual to another. This size is a characteristic of the chromosome. It is inherited by the same way as the alleles of the chromosome. The paternity test consists on determining the size of certain micro satellites. By comparing the characteristics of each chromosome of the child and the potential parents; it is possible to determine the real parents.
We take the genetic cards of two couples: Sarah- Adam and Leila- Samer, and the child of one couple between them: Sonia.

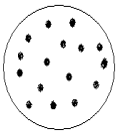
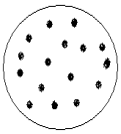
The following table shows the size of the micro satellite ACGCC in each chromosome. For example, Sarah has on her first chromosome 14 the sequence ACGCC**ACGCC**ACGCC (number of repetition= 3), and on the other chromosome 14 the sequence ACGCC**ACGCC**ACGCC**ACGCC** (repetition =4).

	Size of micro satellite (kb) on the homologous chromosomes				
Analyzed chromosome	Sarah	Adam	Leila	Samer	Sonia
Chromosome 1	8 and 15	8 and 18	2 and 8	5 and 13	8 and 13
Chromosome 8	13 and 14	5 and 7	17 and 20	5 and 5	5 and 17
Chromosome 13	4 and 15	3 and 9	4 and 4	4 and 18	4 and 4
Chromosome 14	3 and 4	2 and 8	2 and 14	3 and 8	2 and 3

1- Pick up from the text:
a- The definition of the micro satellite.
b- The characteristics of the micro satellite.
2- Analyze the table and deduce the real parents of Sonia.

Exercise III: Conditions of an immune response. **(3 pts.)**

A-The following experiments are realized in order to determine the conditions of the immune response.

<table>
<tr><td>group of mice</td><td>Group A</td><td>Group B</td><td>Group C</td><td>Group D</td></tr>
<tr><td rowspan="3">treatments</td><td rowspan="2">none</td><td colspan="3">Irradiation (destruction of the stem cells of bone marrow) and ablation of the thymus.</td></tr>
<tr><td>Injection of thymus cells</td><td>Injection of thymus and bone marrow cells</td><td>Injection of bone marrow cells</td></tr>
<tr><td colspan="4">Injection of sheep red blood cells (SRBC)</td></tr>
<tr><td colspan="5">Prelevement of the serum and mix it with SRBC</td></tr>
<tr><td>Results</td><td>  </td><td>  </td><td>  </td><td>  </td></tr>
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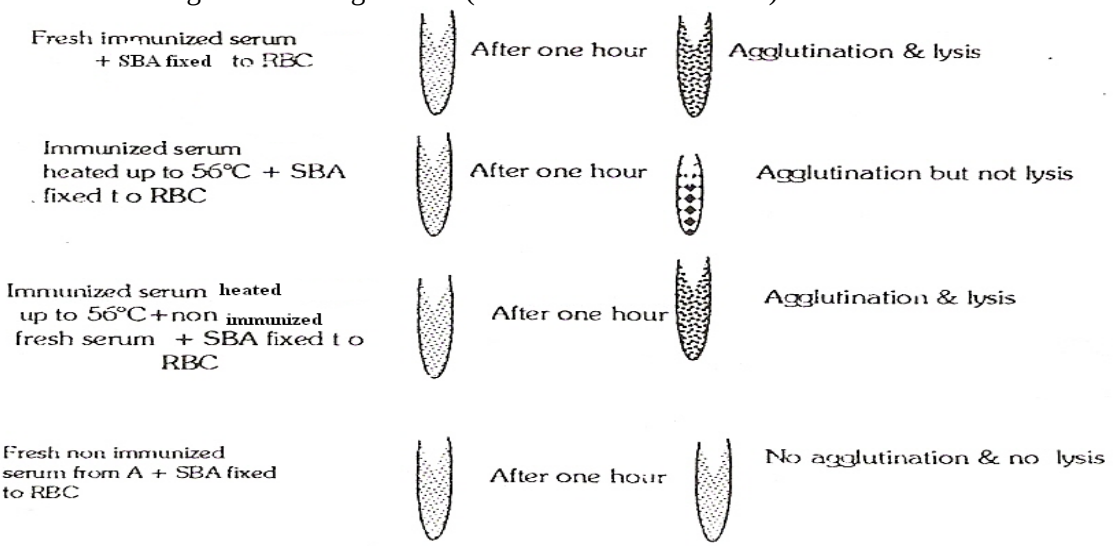
	Agglutinated SRBc	Free SRBC	Agglutinated SRBc	Free SRBC

Document 1: experiments realized on different group of mice.

1- Interpret the experiments of document 1.

B- In the framework of studying the property of antibodies and the complement proteins, studies are done on the serum of immunized animals against the antigen SBA (serum of bovine albumin) as shown in document 2.



Document 2

2- Explain the results of document 2 and find out the properties of antibodies and complements.

Exercise IV: The role of LT_4. **(6 pts.)**

A-Many experiments are done on nude mice, which are deprived of the thymus since birth, to specify the importance of some molecules in the immune response.

		Box 1	Box 2
Content of the box	Spleen cells of a nude mouse	+	+
	Sheep red blood cells (SRBC)	+	+
	Supernatants (do not contain antibodies) coming from a culture of: spleen cells of a normal mouse with SRBC.	_	+
	results	Absence of agglutination of SRBC	agglutination of SRBC

+: presence; _: absence

Document 3

1- Describe the experiments of document 3.

2- Formulate a hypothesis that indicates the content of the supernatant.

3- Do the results of the experiments of document 3 validate your hypothesis? Justify your answer.

B- Other experiments are done to clarify the importance of LT4 in the immune system.

Lymphocytes are taken from monkey. They are then classified into different populations: LB, LT_4, and LT_8.

- LB are placed in a cultural chamber having antigen X. Only 0.01% of LB is precipitated in the bottom of the chamber even after washing(chamber 1a). Then different types of cells are added (1b and 1c).
- LT_8 are placed in a cultural chamber having cancerous fibroblasts of a monkey. Only 0.01% of LT_8 is precipitated in the bottom of the chamber even after washing(chamber 2a). Then activated LT_4 is added (chamber 2b).

Chamber	1a	1b	1c	2a	2b

Lymphocytes placed in the chamber	LB fixing antigen X	LB fixing antigen X + activated LT_4	LB fixing antigen X + activated LT_8	LT_8 fixing cancerous fibroblast cells of monkey	LT_8 fixing cancerous fibroblast cells of monkey + activated LT_4
Results	No antibody is found in the chamber	Presence of antibodies in the chamber	No antibody is found in the chamber	No lysis of fibroblasts	lysis of fibroblasts

Document 4

4- Interpret the above table, what can you deduce?

5- Make a functional diagram that shows the different steps of the immune responses revealed in document 4 and the cells involved in these responses.

Good Work