

First name : _____ Last name : _____

Question 1

10 pt

The 200-base long sequence given below, from the genome of a Bacterium, is the coding strand immediately downstream from a promoter.

```
5' taccagtaga cgcctacgaa taaccaacca accaaccggt
   tatgcaccag gctaaacgaa tgccacatgg caaaacgtaa
   tggatccga cggctctgga gtaagcagac acacgtcact
   atggattggc tggcctgcgt ccgaaaattc ctgaaaaatt
   gccccacaca agctccttag gcgggactcc ctcaaattag 3'
```

Indicate the length in amino acids of the protein encoded by the sequence as given

_____ amino acids

From now on we consider 1 and **only 1 mutation at the time** : mutations do not accumulate.

Indicate the length in amino acids of the protein when at the position 44 G → C

_____ amino acids

Indicate the length in amino acids of the protein when at the position 72 A → T

_____ amino acids

Indicate the length in amino acids of the protein when stuttering between positions 72 to 75 adds one

A : caaaacg → caaaaacg

_____ amino acids

Indicate the length in amino acids of the protein when at the position 78 T → A

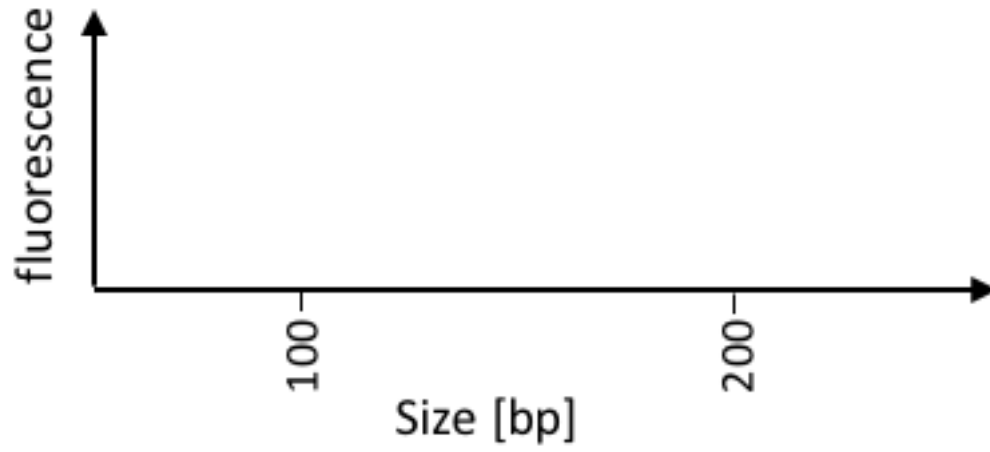
_____ amino acids

Chose 15-nucleotid long primers to PCR amplify the whole sequence given above :

5' _____ 3'

5' _____ 3'

After the PCR has been completed with fluorescent primers, the PCR product is analyzed by capillary electrophoresis. Draw the expected result :



Comment :

First name : _____ Last name : _____

Question 2

6 pt

Seq 1 5'cagagactgg atttacagca taagtttgcg3'

Seq 2 5'cacagactgg attaacagca taagtttgcg3'

Seq 3 5'cacagagtgg attaagagct ttagttaccg3'

Seq 4 5'cacagagtgc attaacagca ttagttagcg3'

Seq 5 5'cacagactgc attaagagct taagttaccg3'

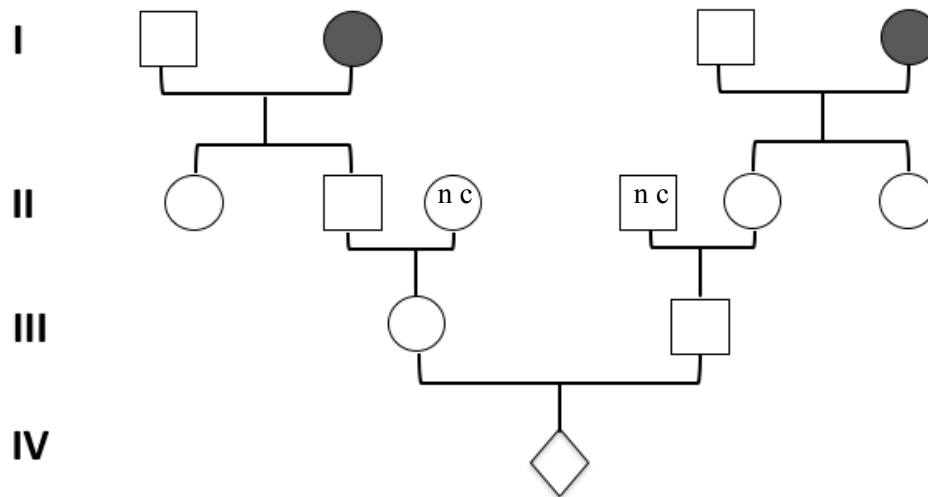
Use the **UPGMA** method (algorithm) to draw a phylogenic tree reflecting the relation between the 5 sequences given above. Indicate distances on each branch of the tree. Show your calculations.

Question 3

20 pt

3.1

The pedigree shows a family in which the 2 women in the first generation (I) are albino. Albinism is a phenotype transmitted on the autosomal recessive mode.



The couple of generation III is expecting a child.

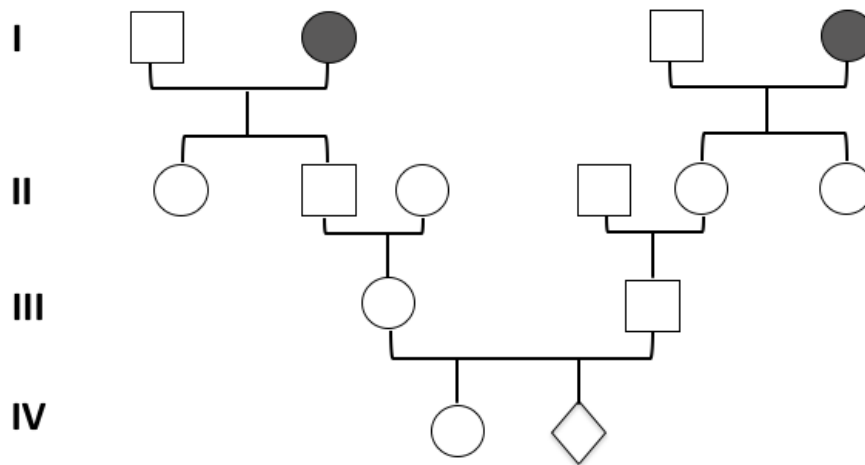
- The pregnant woman has a paternal grand-mother with albinism.
- The father of the child to be born has a maternal grand-mother with albinism.

One assumes that the two people entering in the family by marriage are non-carrier for albinism (indicated as **n c** in the figure).

For the first child in the generation IV what is the probability to be born with albinism? (show your calculations)

First name : _____ Last name : _____

3.2



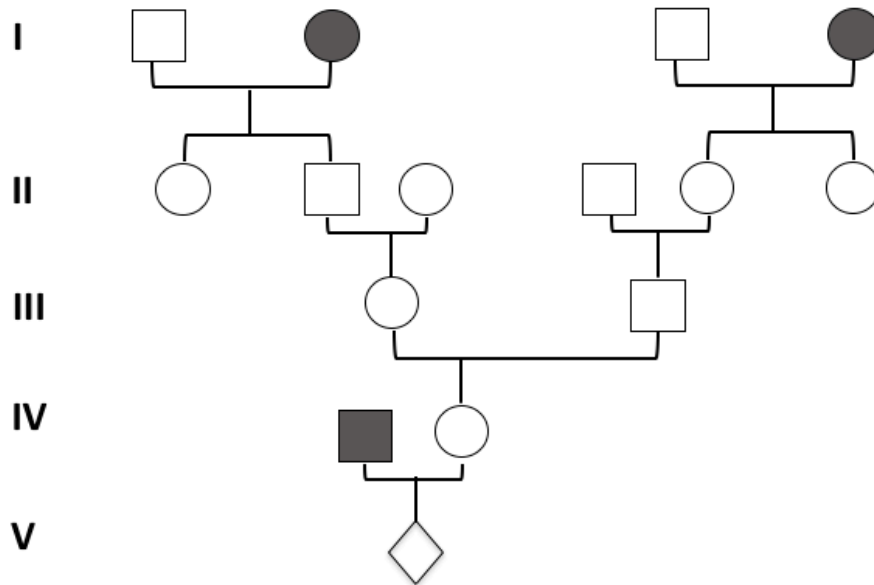
The same family 4 years later: the couple is expecting a second child

Knowing that the first child is a girl non affected with albinism, what is the probability for the second child to be born with albinism ?

(show your calculation)

3.3

Same family 25 years later. The non-albino woman in generation IV is now expecting a child whose father is albino.

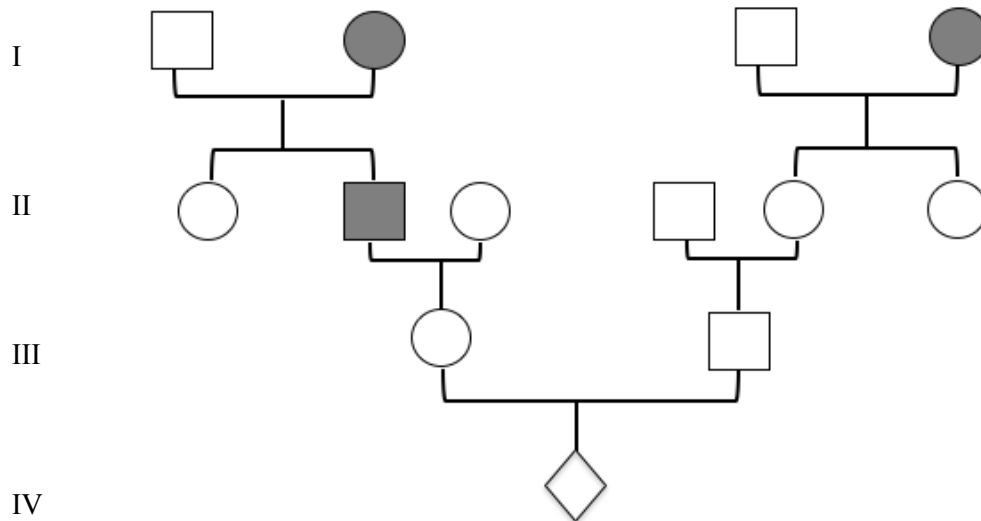


For the first child of this couple (generation V) what is the probability to be borne with albinism ?
(show your calculations)

First name : _____ Last name : _____

3.4

Red/green color blindness (daltonism) is affecting 3 members of the family whose pedigree is shown below. Red/green color blindness is a phenotype transmitted on the X-linked recessive mode.



The couple of generation III is expecting a child.

For the first child of this couple what is the probability to be born color blind ?

• if the first child is a girl ? _____

• if the first child is a boy ? _____

(show your calculations)

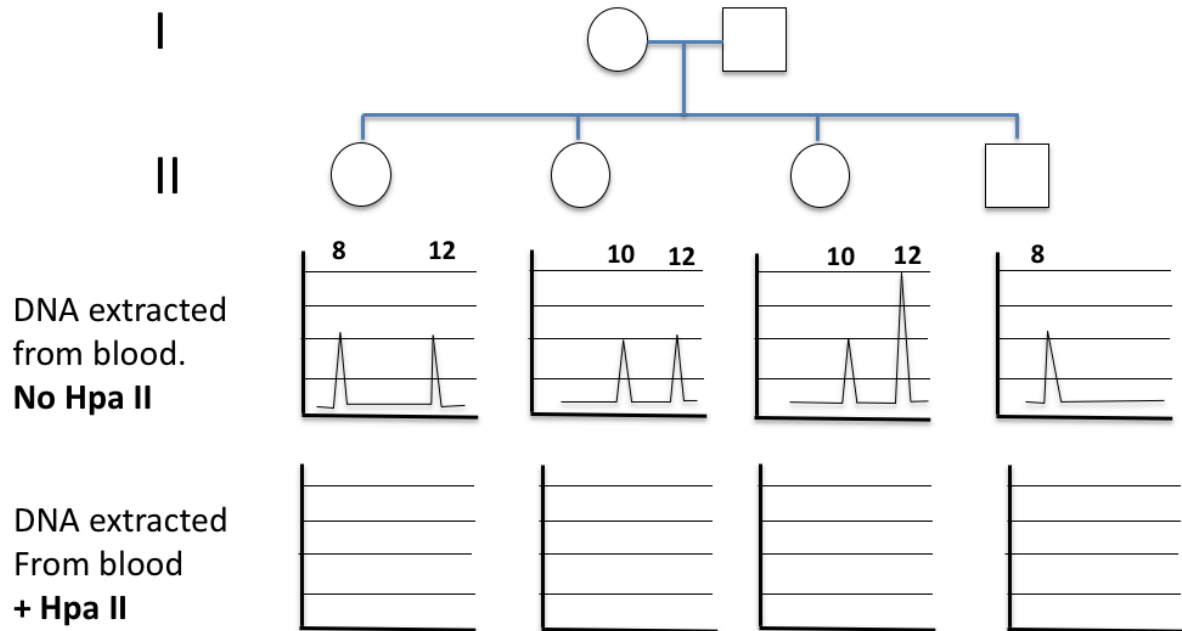
First name : _____ Last name : _____

Question 5

7 pt

A couple has 4 children, 3 girls and a boy.

A **HUMARA test** is done for the 4 children. The result of the test when the restriction enzyme Hpa II is not used is shown on the figure.



5.1 Indicate
 the genotype of the father : _____
 the genotype of the mother : _____

5.2 Is anything noticeable with the results when **Hpa II** is not used ? YES NO
 If YES, describe what is noticeable.

5.3 On the figure draw the expected results of the test when **Hpa II** is used. Assume the X Inactivation Center (XIC) is not mutated on any X chromosome in this family.

Question 6

7 pt

6.1 Uniparental disomy.

- Explain what is uniparental disomy :

- Explain what succession of events lead to uniparental disomy in most cases.

6.2 Pseudo-autosomal regions

- Which human chromosomes have pseudo-autosomal regions ?

- On chromosomes having pseudo-autosomal regions, where are these regions located ?

- Describe what makes the pseudo-autosomal regions of a chromosome different from the rest of the chromosome.
