

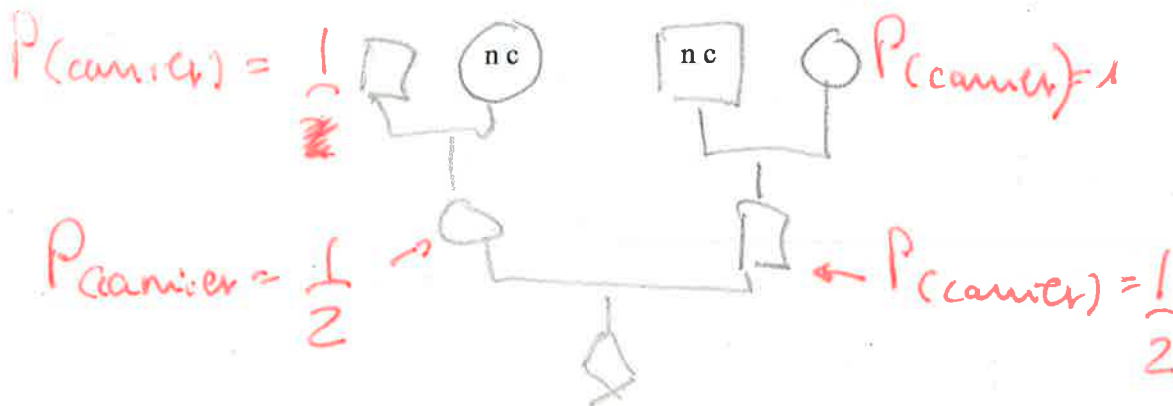
Question 3

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.

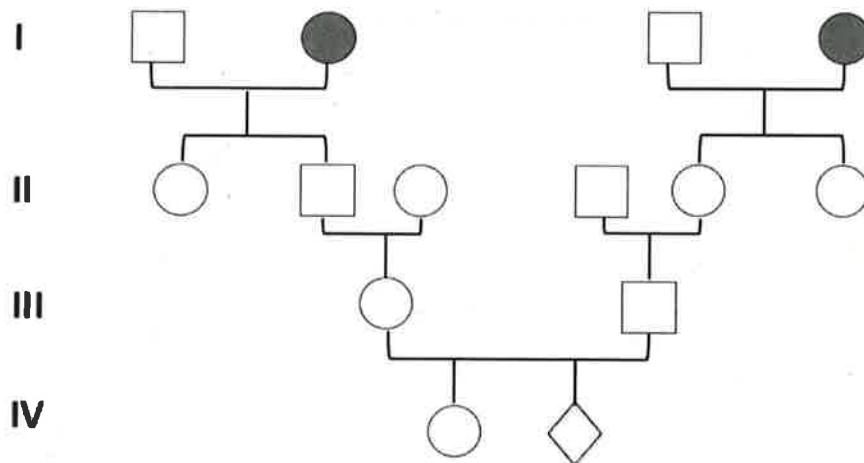
One assumes that the two people entering in the family by marriage are non-carrier for albinism (indicated as **n c** in the figure).

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

$$\text{1st child: } \frac{1}{2} \cdot \frac{1}{2} \cdot \frac{1}{2} \cdot \frac{1}{2} = \frac{1}{16}$$

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)

$$\text{Bayes' : } P(\text{both parents carrier} | 1 \text{ child ok}) = \frac{P(\text{ok}) \cdot P(\text{both carrier})}{P(\text{ok})}$$

$$1 - \frac{1}{16}$$

$$\frac{\frac{3}{4} \cdot \frac{1}{4}}{\frac{15}{16}}$$

$$\frac{\frac{3}{16}}{\frac{15}{16}} = \frac{3}{15} = \frac{1}{5}$$

$$P(2^{\text{nd}} \text{ child} | 1^{\text{st}} \text{ ok}) = \frac{1}{5} \cdot \frac{1}{4} = \frac{1}{20}$$

First name : _____ Last name : _____

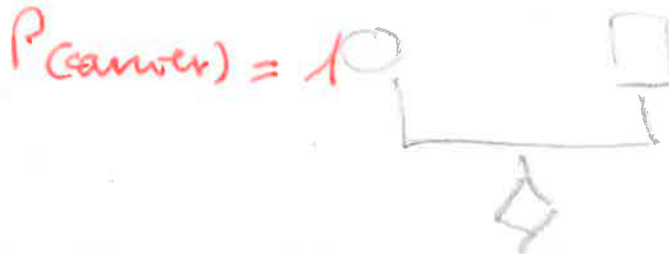
3.4

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.

I

II

III



IV

The couple of generation III is expecting a child.

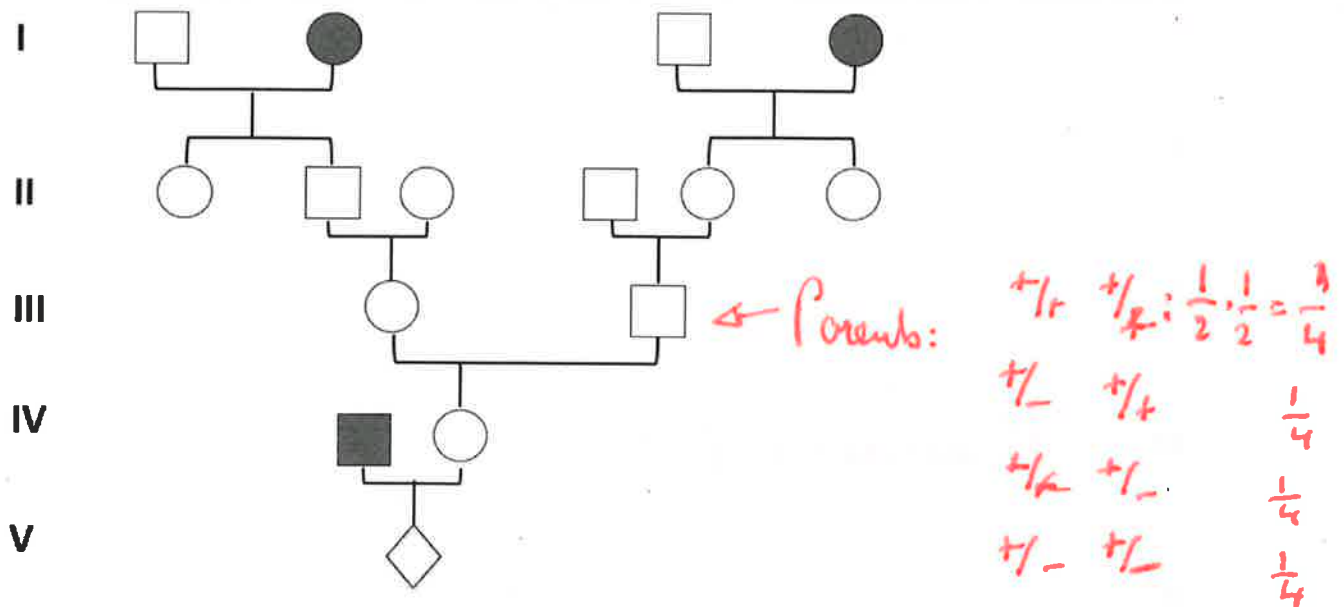
For the first child of this couple what is the probability to be born color blind ?

- if the first child is a girl ? $\frac{0}{1}$ $1 \cdot \frac{1}{2} \cdot 0$
- if the first child is a boy ? $\frac{1}{2}$ $1 \cdot \frac{1}{2}$

(show your calculations)

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)

Parents :	Daughter:		
$+/+$ $+/+$: $\frac{1}{4}$	$+/+$	$+/-$	$-/-$
$+/+$ $+/-$: $\frac{1}{4}$	$\frac{1}{4}$	0	0
$+/+$ $-/-$: $\frac{1}{4}$	$\frac{1}{8}$	$\frac{1}{8}$	0
$+/-$ $+/+$: $\frac{1}{4}$	$\frac{1}{8}$	$\frac{1}{8}$	0
$+/-$ $+/-$: $\frac{1}{4}$	$\frac{1}{8}$	$\frac{2}{16}$	$\frac{1}{16}$
	$\frac{1}{16}$	$\frac{2}{16}$	$\frac{1}{16}$
	$\Sigma \frac{9}{16}$	$\frac{6}{16}$	$\frac{1}{16}$

$$P(\text{daughter } +/-) = \frac{\frac{6}{16}}{\frac{9}{16}} = \frac{6}{9} = \frac{2}{3} \quad P(\text{child } -/-) : \frac{2}{3} \cdot \frac{1}{2} = \frac{1}{3}$$