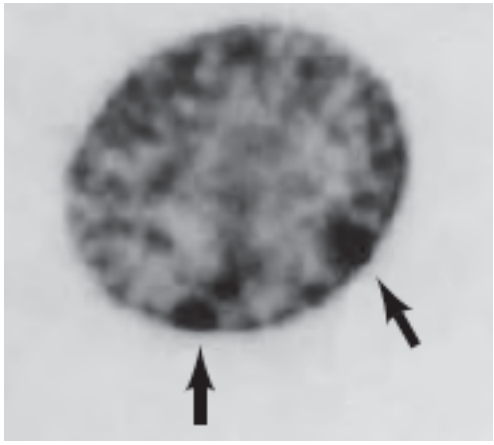
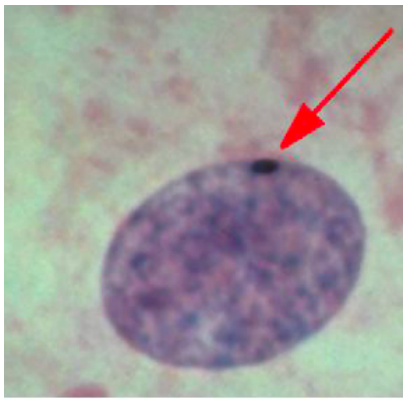
Question 1
Cheek cells are taken from a young person.



Under the microscope one observes two dark spots in the cell nucleus.

1.A What do you conclude when the person is a woman? 2 inacivated X + 1 active X
Karyotype : 47,XXX
1.B What do you conclude when the person is a man? At least 1 Y chromosome
Karyotype 48,XXXY

Question 2
Cheeks cells are taken from a young man.



Under the microscope one observes a dark spot in the cell nucleus.

2.A What do you conclude when the person is a woman? Normal woman
Karyotype 46,XX
2.B What do you conclude when the person is a man?
Karyotype 47,XXY (Klinefelter)

Question 3

Male : dwarf, agouti (wild type)
Autosomal recessive dwarfism
is due to absence of
growth hormone (GH)

Female : normal size, albino

X

X

GH/gh A/a Female F1

male : dwarf (gh/gh), albino (a/a)

A dwarf, agouti male is crossed with a normal size, albino female.
The F1 are 100% agouti, normal size.
F1 females are crossed with dwarf, albino males.

3.A

Describe the phenotypes of the F2 and the relative proportions (%) of these phenotypes assuming that the Tyrosinase locus (causing albinism) and the Growth hormone locus (causing dwarfism) are on the same chromosome (autosome) **28 cM** apart.

In the F1 female the mutations are in ***trans***

3.B

Describe the phenotypes of the F2 and the relative proportions (%) of these phenotypes assuming that the Tyrosinase locus (causing albinism) and the Growth hormone locus (causing dwarfism) are located **on 2 different chromosomes**.

Answer for 3.B
independent assortment
dwarf albino 25%
dwarf agouti 25%
norm. size albino 25%
norm. size agouti 25%

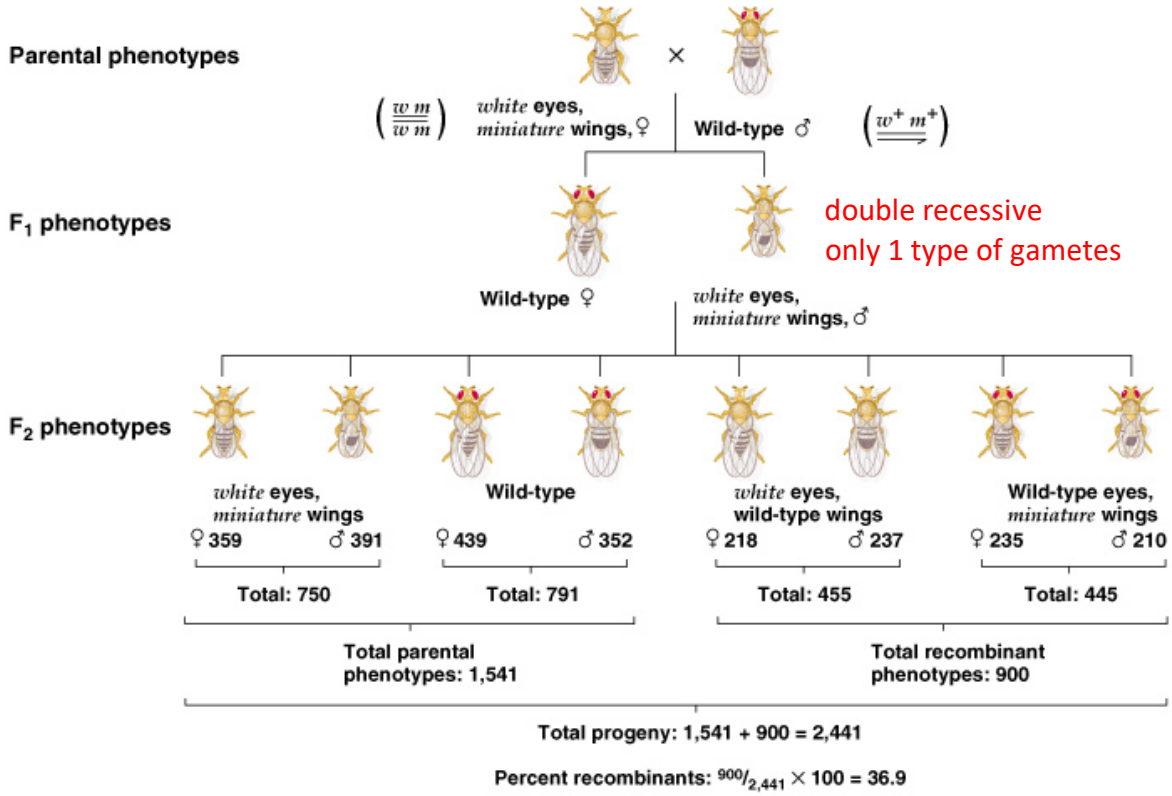
(agouti is the wild-type fur color in mice)

Answer for 3.A
linkage
14% 14 + 14 / 14 + 14 + 36 +36 = 0.28
36%
36%
14%

Question 4

In Drosophila, a locus controlling the color of the eye (red / white) and a locus controlling the size of the wings (long/miniature) are both on the **X chromosome**.

A white eyes, miniature wings female is crossed with a wild-type male.
In the F1, all females are wild-type whereas all male show white eyes and miniature wings.
On crosses F1 female with F1 males. The phenotypes in the F2 are indicated.



double recessive
only 1 type of gametes

Answers between 36 -38 cM are accepted as correct.

From the data calculate the genetic distance between the 2 loci on the X chromosome.

Calculation for the male F2 only :

Total progeny = 391 + 352 + 237 + 210 = 1190
Total recombinant = 237 + 210 = 447
Genetic distance : 447 / 1190 = 0.3756

Calculation for the female F2 only :

Total progeny = 359 + 439 + 218 + 235 = 1251
Total recombinant = 218 + 235 = 453
Genetic distance : 453 / 1251 = 0.3621

Calculation for both males and females:

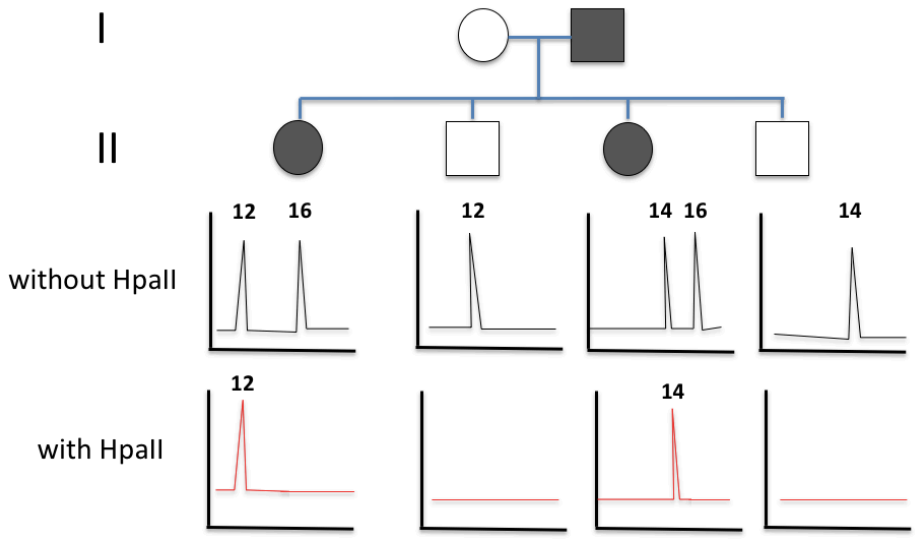
Total progeny = 1190 + 1251 = 2441
Total recombinant = 447 + 453 = 900
Genetic distance : 900/2441 = 36.9

best answer because the sample size is highest !!

The three answers are identical because it always reflects the recombination in the F1 female only. There is no raison not to pool the data for males and females

Question 5

A couple has 2 daughters and 2 sons. The father is colorblind (does not distinguish green from red), the mother distinguishes green from red.
Their 2 sons distinguish red from green but their 2 daughters are colorblind!
According to textbooks, colorblindness is transmitted by the X chromosome:
- males are colorblind when their X chromosome is mutated
- carrier females (1 X chromosome mutated) are not colorblind;
- homozygote females (2 X chromosomes mutated) are colorblind.
The family presented here is exceptional.



The HUMARA assay has been done for all 4 children.
Results without digestion by HpaII are given.

Genotype of the mother: __ 12, 14_____ mother gave 12 to 1rst son and 14 to 2nd son

Genotype of the father: ___ 16 _____ father gave 16 to both daughters

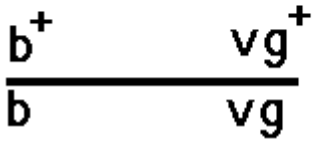
Is the mother carrier for colorblindness? YES NO
The 2 sons have received different X from their mother, but none is affected; the two daughters have inherited different X from their mother but both are affected. This make the mother very unlikely to be carrier.

Draw the most likely results of the HUMARA assay with digestion by HpaII before the PCR. Explain your reasoning.
Both daughters received the mutated X from their father but they would not be affected if X inactivation is random. Let's therefore assume **non random** X inactivation: the paternal X remains always (100%) active and the maternal X is always inactivated. This is explained by a **loss-of-function** mutation of **XIC** on the paternal X chromosome; the paternal X cannot do the inactivation and the maternal X compensates by being always inactivated. The loss-of-function is not detrimental in the father (no X inactivation in men !)

Site web vraiment bon :

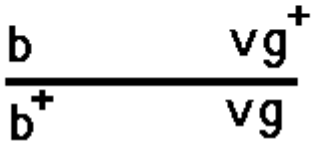
http://web.pdx.edu/~newmanl/ChapterOutlines/Chapter05.html

Cis and Trans double heterozygotes



Cis

both wilds on one chromosome
both mutants on the other chromosome



Trans

one mutant and one wild on each chromosome