

Instructions:

Do not open this exam until instructed to do so.

You will have 60 minutes to complete the exam.

You may not leave the examination room during the exam.

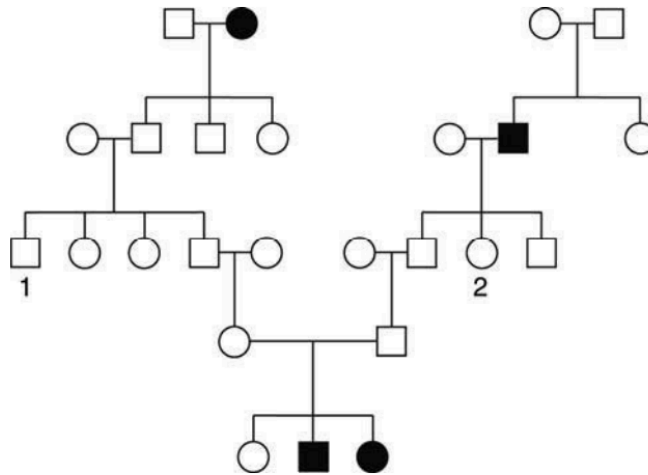
Phones, tablets, computers, and any other electronic device are strictly prohibited. They must be completely out of sight for the entirety of the exam.

You may use a calculator. Phones, tablets, laptops, etc. may not be used as calculators. It must be a separate, regular calculator.

Table 1: Punnett Square Showing Dihybrid Cross

	AB	Ab	aB	ab
AB	AABB	AABb	AaBB	AaBb
Ab	AABb	Aabb	AaBb	Aabb
aB	AaBB	AaBb	aaBB	aaBb
ab	AaBb	Aabb	aaBb	aabb

Question 1 (12 points): Consider the pedigree for a rare genetic disease shown below (assume individuals that marry into this family in the second and third generations are wild type for both alleles):



a) What is the inheritance pattern for this disease? Give one reason to justify your answer.

autosomal recessive

skips generations, males and females equally affected, non-affected parents have affected children

b) What are the possible genotypes for Person 1 (labeled with a “1” under the symbol in the third generation)? Use the allele designations B and b as appropriate.

BB or Bb

c) What is the genotype of Person 2 (labeled with a “2” under the symbol in the third generation)? Use the allele designations B and b as appropriate.

Bb

d) If person 1 and Person 2 had a child, what is the probability that the child would be affected by this disease?

person 1 has a 1/2 chance of being Bb

person 2 is definitely Bb so the probability is 1

if both are carriers, the probability that the child would be affected is 1/4

So $1/2 \times 1 \times 1/4 = 1/8$

Question 2 (12 points): The mutation that causes Duchenne Muscular Dystrophy is an X-linked recessive allele. A female carrier for this allele has three children with an unaffected man.

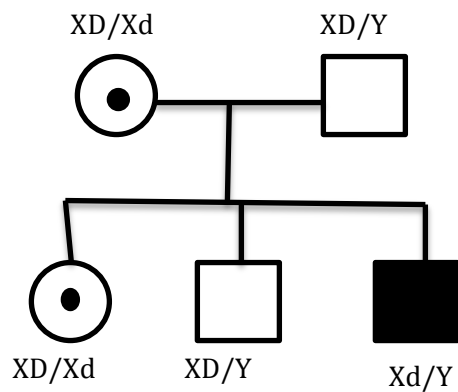
a) What is the probability that the three children are precisely (in this order): one carrier daughter, one affected son, and one unaffected son? Draw a Punnett square to illustrate the possible offspring of this couple. Use the allele designations X^D and X^d .

$$\frac{1}{4} \times \frac{1}{4} \times \frac{1}{4} = 1/64$$

$X^D X^d \times X^D Y$ cross

	X^D	X^d
X^D	X^D/X^D	X^D/X^d
Y	X^D/Y	X^d/Y

b) Draw the pedigree for the family described above and label each individual with the appropriate genotypes.



Question 3 (12 points): Consider the following four gene cross:

(female) $w/w; X/x; Y/Y; Z/z$ x (male) $W/w; X/x; Y/y; z/z$

Assume all four genes assort independently and display complete dominance. Show your work for all questions below.

a) What is the probability of obtaining offspring with the same *phenotype* as the female parent?

$$1/2 \times 3/4 \times 1 \times 1/2 = 3/16$$

b) What is the probability of obtaining offspring with the same *genotype* as the male parent?

$$1/2 \times 1/2 \times 1/2 \times 1/2 = 1/16$$

c) What is the probability of obtaining *either* an offspring with the maternal phenotype OR an offspring with the paternal genotype?

$$(a) 3/16 + (b) 1/16 = 4/16 = 1/4$$

Question 4 (14 points): A pure-breeding female fly with red eyes and a hairy body ($r^+/r^+; h^+/h^+$) is crossed with a pure-breeding male with gold eyes and a smooth body ($r/r; h/h$). The genes are autosomal. Assume complete dominance.

The F1 generation from the parental cross (above) are then **testcrossed**. (this is $r^+r h^+h \times rr hh$)!!!

The ratio of the offspring from the F1 testcross are as follows:

F2 results:

<u>P or R:</u>	<u>phenotype:</u>	<u># of offspring</u>	<u>genotype:</u>
<div style="border: 1px solid black; padding: 2px; display: inline-block;">P</div>	red eyes and hairy body	502	__ $r^+r h^+h$ __
<div style="border: 1px solid black; padding: 2px; display: inline-block;">R</div>	red eyes and smooth body	127	__ $r^+r hh$ __
<div style="border: 1px solid black; padding: 2px; display: inline-block;">R</div>	gold eyes and hairy body	133	__ $rr h^+h$ __
<div style="border: 1px solid black; padding: 2px; display: inline-block;">P</div>	gold eyes and smooth body	498	__ $rr hh$ __

a) Identify the parental type offspring and recombinant offspring by placing a P or an R, respectively, in the boxes to the left of each offspring category.

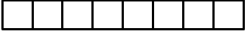
b) On the line to the right of each offspring category, give the genotype of those offspring.

c) Are the r^+ and h^+ alleles linked? If yes, calculate the distance between these genes in map units (be sure to show your work). If no, justify your response using the information from the F2 results above.

$$(133+127)/\text{total}(1260) \times 100 = 20.6\% \text{ recombinants} = 20.6 \text{ map units (or centimorgans.)}$$

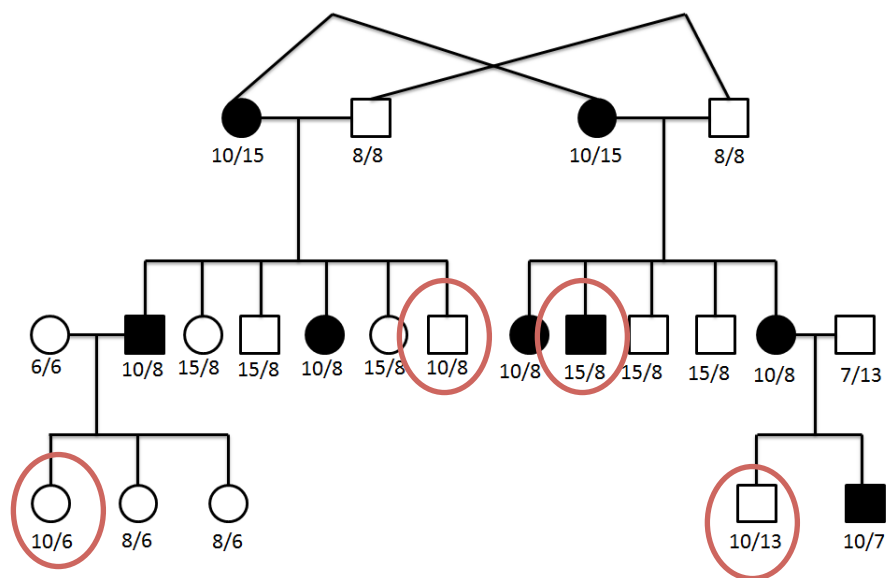
Question 5 (10 points): The following pedigree shows a family where a pair of identical twin sisters marry a pair of identical twin brothers (identical twins have identical DNA). The sisters are both affected with a rare disorder. Assume the twin brothers, and the individuals that marry into the family in the second generation, are homozygous for the wildtype allele. Also shown on the pedigree are designations for minisatellite repeats that are linked to the gene that causes the disorder.

The numbers under each symbol in the pedigree indicate the number of minisatellite repeats identified in that individual using PCR. For example:

 8 repeats

 6 repeats

If a person had the two minisatellite alleles above, they would be designated 8/6.



a) What is the inheritance pattern? Autosomal dominant

b) On the pedigree, circle all recombinant individuals (remember, the minisatellites and the disease-causing gene ARE LINKED).

c) Based on the number of recombinant individuals you identify above, estimate the distance between the minisatellite region and the disease gene in map units. Show your work.

4 recombinants out of 16 offspring = $4/16 \times 100 = 25\% = 25$ map units

Question 6 (9 points) (to assist you, a generic dihybrid punnett square is on the front page of the exam)

In a flowering plant the $a/a; B/B$ genotype results in yellow flowers, and the $A/A; b/b$ genotype gives red flowers. Wild-type flowers are orange and are observed in plants with genotype $A/A; B/B$. Two possibilities have been proposed for the biosynthetic pathways of flower color:

1] yellow pigment \rightarrow red pigment \rightarrow orange pigment

2] red pigment \rightarrow yellow pigment \rightarrow orange pigment

In order to distinguish between these possibilities, you cross a pure breeding yellow plant with a pure breeding red plant.

a) What is the genotype and phenotype of the F1 offspring?

$A/a; B/b$ Orange

b) You self-cross the F1 offspring. If Pathway 1 is correct, what ratio of offspring would you expect in the F2 generation? If Pathway 2 is correct, what ratio of offspring would you expect in the F2 generation?

Cross a red pure line to a yellow pure line to obtain a dihybrid $A/a; B/b$, then self this dihybrid and inspect the F₂. If possibility 1 is correct, then we expect a ratio of 9 orange : 4 yellow : 3 red. If possibility 2] is correct, we expect a ratio of 9 orange : 4 red : 3 yellow.

Note: Since we are trying to differentiate between 9:3:4 and 9:4:3, it's essential that we have a large enough sample size.

Question 7 (10 points) Gaucher's Disease is an autosomal recessive disorder that prevents the proper lysosomal breakdown of cell membrane components. Symptoms affect many organ systems and include hepatomegaly (enlargement of the liver), bone and joint pain, yellow-brown pigmentation of the skin, and blood clotting deficiencies.

a) **Briefly** describe variable expressivity, using Gaucher's Disease as a specific example.

some patients might have hepatomegaly but not joint pain, and others may have both symptoms (for instance, there are many possibilities here)

b) **Briefly** describe incomplete penetrance, using Gaucher's Disease as a specific example.

A few individuals within a pedigree are homozygous recessive for the Gaucher's Disease allele but do not express any symptoms

c) a man and a woman that both have Gaucher's disease marry and have 7 children. None of the children are affected by the disorder. What is the best explanation as to why the children are not affected?

These two people likely have mutations in different genes that lead to Gaucher's disease, and there for the mutations complement each other (complementation)

Question 7 (10 points): Indicate whether the following events apply to mitosis, meiosis, both, or neither (circle only one per statement):

a) Alignment of chromosomes along the metaphase plate

Mitosis only Meiosis only both Mitosis and Meiosis neither

b) Separation of sister chromatids

Mitosis only Meiosis only both Mitosis and Meiosis neither

c) Resulting daughter cells are $4n$

Mitosis only Meiosis only both Mitosis and Meiosis neither

d) Separation of homologous chromosomes

Mitosis only Meiosis only both Mitosis and Meiosis neither

e) Resulting daughter cells are genetically identical

Mitosis only Meiosis only both Mitosis and Meiosis neither