Exams

Fall 1994

Name: ANSWER KEY

Recitation day_____, time_____, TA_____

7.03 Hour Exam 1

October 5, 1994

Write your name on all **six** pages.

Indicate your recitation section on this page

Write all answers on this handout only

Exam begins at **11:05** and ends at **11:55**

Time will be announced when 5 and 1 minutes remain.

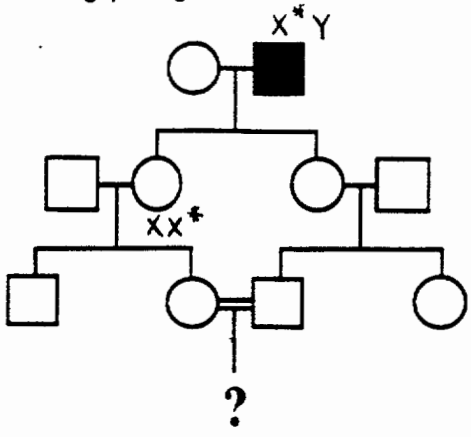
Problem 1	30 points
Problem 2	30 points
Problem 3	40 points
Total	100 points

Name:_____

1 The traits in the pedigrees below are completely penetrant.

□ male ○ female ■ affected male ● affected female

(a 5 pts) The following pedigree shows inheritance of an X-linked recessive trait.



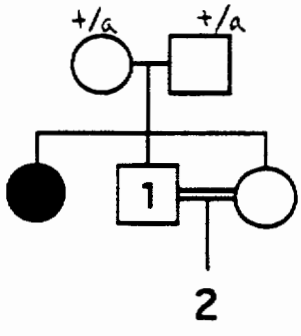
What is the probability that a son from the first-cousin marriage shown will exhibit the trait?

p(son will inherit the recessive allele from his mother) = 1/2

p(mother is a carrier) = 1/2

1/2 × 1/2 = 1/4

(b 10 pts) The following pedigree shows inheritance of the short-eared trait in mice.



autosomal recessive inheritance

i) What is the probability that mouse **1** is a heterozygote?

possible genotypes of mouse 1 : +/a
+/a
+/+

2/3 heterozygotes

ii) What is the probability that mouse **2** (the progeny from the brother-sister mating) will exhibit the short-eared phenotype?

p(mouse 1 is a heterozygote) = 2/3

p(mother of mouse 2 is a heterozygote) = 2/3

p(mouse 2 is homozygous recessive given heterozygous parents) = 1/4

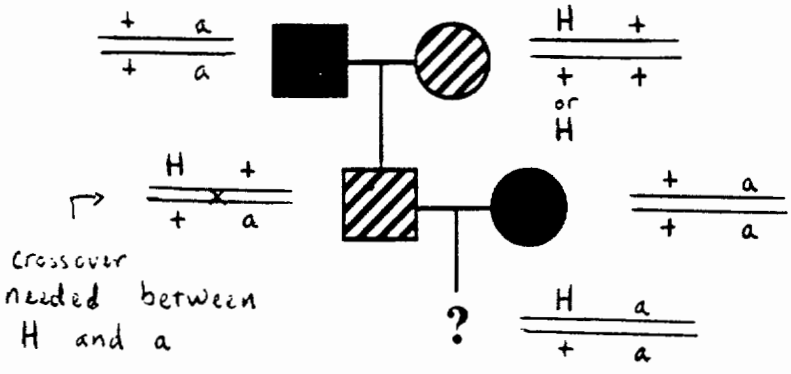
$$2/3 \times 2/3 \times \frac{1}{4} = \frac{1}{9}$$

Name:_____

(c 15 pts) Huntington's disease is caused by an autosomal dominant allele. This pedigree shows inheritance of Huntington's disease in a family. Also present in this family is a particular blood antigen which is a recessive trait. The Huntington's disease locus and the locus for the blood antigen are on the same chromosome and are **5** cM apart.

▨ affected with Huntington's disease ■ exhibits recessive blood antigen

(none of the individuals shown are both affected with Huntington's and exhibit the blood antigen)



What is the probability that the indicated grandchild will both exhibit the recessive blood antigen and develop Huntington's disease? Please show your work.

p(crossover between H and a) = 0.05

p(grandchild to inherit the double mutant chromosome) = 1/2

(0.05)(1/2) = 2.5%

Name:_____

2. Wild-type humbugs have brown bodies and brown eyes. You have isolated mutations in three new humbug genes. The mutation **sp** is dominant and gives humbugs with spotted bodies. The mutation **gr** is recessive and gives humbugs with green bodies. The mutation **bl** is recessive and gives humbugs with black eyes.

You cross two true-breeding mutant strains to produce F1 females heterozygous for **sp**, **gr**, and **bl**. These F1 females are then test-crossed to true-breeding black-eyed, green-bodied males. The phenotypes of 3000 progeny are scored as shown below:

Phenotypes	number
sp bl gr	53
+ + +	61
sp bl +	1390 → parentals
+ + gr	1347 → parentals
+ bl +	4 → double crossovers
sp + gr	2 → double crossovers
sp + +	70
+ bl gr	74

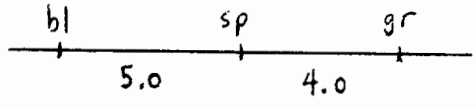
parental: sp bl + / + + gr vs double crossovers: + bl + / sp + gr

order is: bl sp gr

(a 5 pts) What are the genotypes of the two true-breeding parents of the F1 females?

sp bl + / sp bl + & + + gr / + + gr

(b 15 pts) Draw a map showing the order and distances between the **sp**, **gr**, and **bl** genes.



(c 10 pts) A dominant mutation eyeless (**ey**) is identified. You want to map **ey** relative to **bl** but your lab partner claims this can't be done since you obviously can't score the presence of black or brown eyes in an eyeless bug. You don't agree and you cross a true-breeding eyeless bug to a true-breeding black-eyed bug. An F1 female is crossed to a true-breeding black-eyed male. The following phenotypes are observed in 100 progeny:

eyeless	51	~ 10/51 are recombinant
black-eyed	39	parental; ~ 41/51 are parental
brown-eyed	10	recombinants

What is the map distance between **bl** and **ey**?

$$\frac{2(10)}{51 + 39 + 10} \times 100 = 20\ cM$$

Name:_____

3. You have isolated two different yeast strains defective in methionine synthesis (met$^-$). These strains are unable to grow on medium that does not contain methionine. When one strain is mated to the other the diploids can grow on medium that does not contain methionine. From this you conclude that the mutations are in different genes which you call met1 and met2. You sporulate the diploids and score 100 tetrads of three types.

Tetrad type	number of tetrads	
4 met$^-$	75	PD
3 met$^-$:1 met$^+$	20	T
2 met$^-$:2 met$^+$	5	NPD

(a 10 pts) Calculate the distance between met1 and met2.

PD >> NPD, so met 1 and met 2 are linked.

$$M.U. = \frac{T + 6NPD}{2(\text{total})} \times 100 = \frac{20 + 30}{2 \times 100} \times 100 = 25 \text{ cM}$$

(b 5 pts) You are interested in determining the genotypes of the individual spores from one of the tetrads which has four met$^-$ spores. The genotype of each spore clone is tested by mating to each of four test strains that are of either mating type **a** or mating type α and are either met1$^-$ or met2$^-$. The results of these mating tests are shown below where **+** indicates that the mating produced diploids that can grow without methionine in the medium. Based on the behavior of spores 1-3, fill in the expected results for spore 4. Remember that in a tetrad there will be two spores of mating type **a** and two spores of mating type α. Also remember that cells of the same mating type can't mate.

	α met1$^-$	**a** met1$^-$	α met2$^-$	**a** met2$^-$
spore 1	+	−	−	−
spore 2	−	−	+	−
spore 3	−	−	−	+
spore 4	−	+	−	−

(c 5 pts) **A met$^-$ spore** from one of the 2 met$^-$:2 met$^+$ tetrad types is mated to a wild type strain. When the diploid is sporulated, which of the following classes do you expect to occur least frequently and why?

Type I	Type II	Type III
4 met$^-$	3 met$^-$:1 met$^+$	2 met$^-$:2 met$^+$

Type I = NPD

result of 2 crossovers

Parts d,e,f omitted

Name: ANSWER KEY

Recitation day_____, time_____, TA_____

7.03 Exam 2

October 28, 1994

Write your name on all **seven** pages.

Indicate your recitation section on this page

Write all answers on this handout only

Exam begins at **11:05** and ends at **11:55**

Time will be announced when 5 and 1 minutes remain.

Problem 1	30 points
Problem 2	30 points
Problem 3	40 points
Total	100 points

Name:_____

1 Bacteriophage ß makes plaques on bacterial host strains *X14* and *G13*. Five mutant strains **a**, **b**, **c**, and **d** of ß are able to make plaques on host *X14* but not on *G13*. All five mutations are recessive and fail to complement one another and therefore are in the same gene designated H1.

In order to map the mutations with respect to one another, you coinfect *X14* bacteria with pairwise combinations of the mutants and titer the resulting phage on both *X14* and *G13* hosts. The total number of plaques formed on *X14* for each cross is ~1x10^4, and the number of plaques formed on *G13* for each cross is given in the table below:

	a	**b**	**c**	**d**
a	-	50	175	125
b		-	125	75
c			-	50
d				-

a-b : $2 \times 50/10^4 = 1$ cM

a-c : $2 \times 175/10^4 = 3.5$ cM

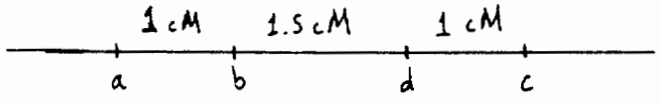
a-d : $2 \times 125/10^4 = 2.5$ cM

b-c : $2 \times 125/10^4 = 2.5$ cM

b-d : $2 \times 75/10^4 = 1.5$ cM

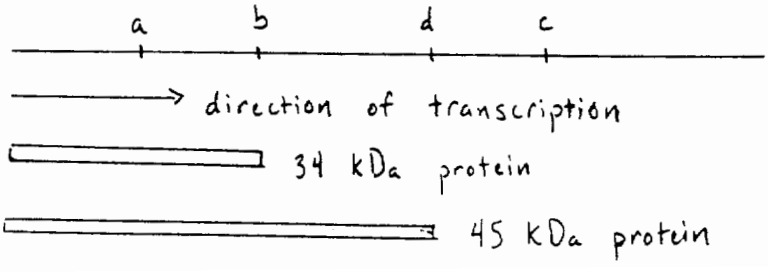
c-d : $2 \times 50/10^4 = 1$ cM

(a 10 pts) Draw a map showing the order of the **a**, **b**, **c**, and **d** mutations and the distances between them in map units.



The protein product of the wild-type H1 gene produced when phage are grown on either *X14* or *G13* has a molecular weight of 55,000 Da. When phage with the **b** mutation are grown on *X14* the H1 protein produced is 34,000 Da. When phage with the **d** mutation are grown on *X14* the H1 protein produced is 45,000 Da. The mutant strains **b** and **d** can make plaques on a derivative of *G13* that contains the amber suppressor supE.

(b 5 pts) Use this new data to determine the direction of transcription and translation of the H1 gene. Redraw the map from part a showing the direction of transcription.



Name:_____

(c 10 pts) The average molecular weight of an amino acid is 110 Da. Given the sizes of H1 protein from the **b** and **d** strains you can determine the physical distance in base pairs **between** the **b** and **d** mutations. When all of the intervals in the genetic map of phage ß are added together the length of the phage genome is 300 map units. What is the size of the phage genome in base pairs?

physical distance between b + d : $\frac{(45-34)\times 10^3 \text{ Da}}{110 \text{ Da/a.a.}} \times 3 \text{ bp/a.a.} = 300 \text{ bp}$

genetic distance between b + d: 1.5 m.u.

∴ recombination freq. in phage β = $\frac{1.5}{300} = \frac{1 \text{ m.u.}}{200 \text{ bp}}$

size of phage genome = 300 × 200 = 60,000 bp

(d 5 pts) When phage with the **c** mutation are grown on *X14* the molecular weight of H1 protein produced is 58,000 Da. Propose a model for the nature of the **c** mutation that explains why the H1 protein from **c** is larger than the wild-type H1 protein.

Model 1 - c mutation alters stop codon, allowing translation to continue until another stop codon is reached

Model 2 - c is an insertion

Model 3 - c is a frameshift and a stop codon is encountered later (- or +) than in w.t.

2 Wild type *E. coli* can grow on medium that contains the sugar maltose as a carbon and energy source. You have isolated two new mutants that can't grow on maltose. You call these mutants **mal1⁻** and **mal2⁻**. Both mutations are linked to the **pyrF** gene by P1 transduction. You are interested in determining the relative order of **pyrF**, **mal1** and **mal2**. To do this you perform two P1 transduction experiments.

Experiment 1: P1 grown on a **pyrF⁺ mal1⁻** strain is used to infect a **pyrF⁻ mal2⁻** strain.

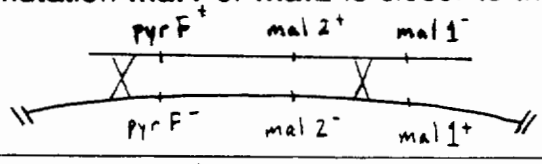
Of the **pyrF⁺** transductants, 5% are **mal⁺**.

Experiment 2: P1 grown on a **pyrF⁺ mal2⁻** strain is used to infect a **pyrF⁻ mal1⁻** strain.

Of the **pyrF⁺** transductants, less than 1% are **mal⁺**.

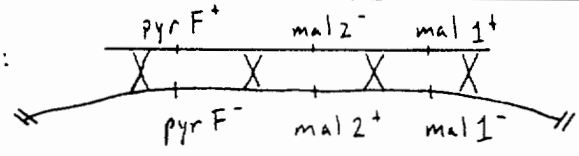
(a 5 pts) Which mutation **mal1** or **mal2** is closer to the **pyrF** gene? mal 2

Experiment 1:



double crossover required

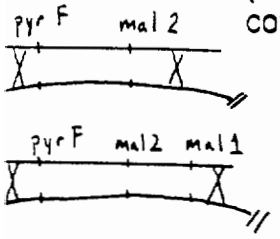
Experiment 2:



quadruple crossover required
∴ rarer

This gene order is consistent with the data:
less than 1% is rarer than 5%.

Name:_____

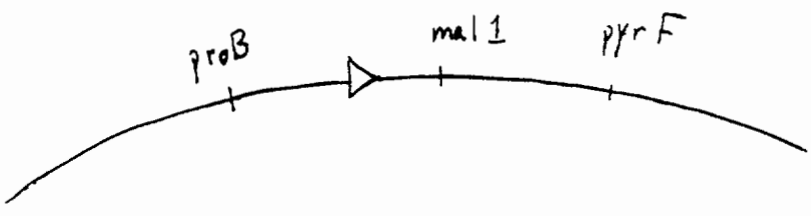


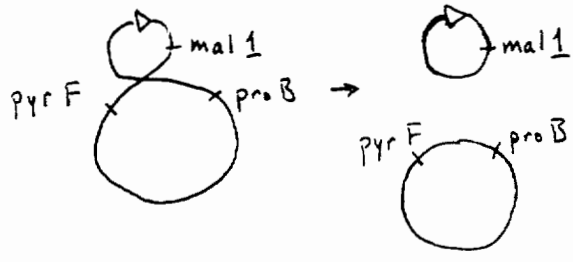
(b 10 pts) Given that **mal2** shows 60% cotransduction with **pyrF** what is the cotransduction distance between **mal1** and **pyrF**?

From experiment 1 one knows that in a cotransduction with pyrF, there is a 5% chance of a crossover between mal 1 + mal 2. Therefore cotransduction distance between mal 1 + pyr F = 60% - 5% = 55%

An Hfr strain is isolated that transfers **proB** early and **pyrF** late. This Hfr is tested for the ability to transfer the **mal+** trait to a **mal1-** mutant . **Mal+** is found to be transfered late. From the Hfr an F' is isolated that transfers **Mal+** early at high efficiency but does not transfer either **proB** or **pyrF**.

(c 15 pts) Draw a map of the relevant portion of the *E. coli* chromosome in the Hfr showing the approximate locations of the **proB**, **pyrF** and **mal** genes. Also show the position and orientation of the origin of transfer for the Hfr. How F' arose:





3 Consider a hypothetical bacterial operon that controls the synthesis of permease for raffinose and the enzyme raffinase both of which are needed for the bacteria to grow on raffinose. When glucose is available as a carbon source synthesis of both the permease and raffinase are shut off as shown below:

	- glucose	**+ glucose**
raffinase	high	low
permease	high	low

There are three <u>linked</u> mutations (A^-, B^-, and C^-) which alter the expression of raffinase activity. You have determined that raffinase and permease are expressed in the same operon and that the A^- mutation is an amber mutation in the gene encoding raffinase enzyme. You construct the strains of bacteria to perform dominance tests and cis-trans tests to determine the nature of the B and C mutations.

Name:_____

Genotype	Raffinase activity - glucose	Raffinase activity + glucose	Permease activity - glucose	Permease activity + glucose
$A^- B^+ C^+$	low	low	high	low
$A^+ B^- C^+$	low	low	low	low
$A^+ B^+ C^-$	low	low	low	low
Dominance tests				
$A^+ B^- C^+ /$ $F' A^+ B^+ C^+$	high	low	high	low
$A^+ B^+ C^- /$ $F' A^+ B^+ C^+$	high	low	high	low
Cis - Trans tests				
$A^+ B^- C^+ /$ $F' A^- B^+ C^+$	low	low	low	low
$A^- B^- C^+ /$ $F' A^+ B^+ C^+$	high	low	high	low
$A^+ B^+ C^- /$ $F' A^- B^+ C^+$	high	low	high	low
$A^- B^+ C^- /$ $F' A^+ B^+ C^+$	high	low	high	low

(a 5 pts) Is the synthesis of raffinase and permease controled by a repressor or by an activator? Why? activator

The B^- and C^- recessive mutations show low activity in the presence and absence of glucose and/or C is recessive, uninducible and trans-acting. Therefore - activator.

(b 5 pts) What is the nature of the B^- mutation?

All acceptable answers

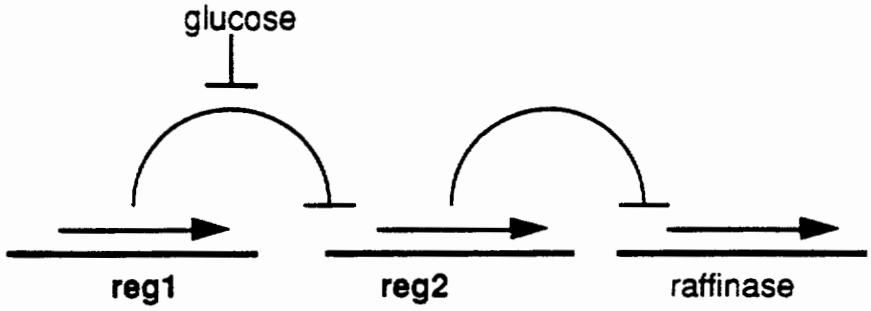
1) cis + recessive
2) mutation in activator binding site
3) mutation in the activator binding site of the operator
4) mutation in promoter

(c 5 pts) Propose a simple molecular model for the role of glucose in raffinase regulation.

Glucose binds to the activator produced by C and prevents it from binding to the activator binding site. Therefore transcription of raffinose and permease only occurs at low levels in the presence of glucose.

Name:_____

In a different bacterial species raffinase is also found to be shut off by glucose but the regulatory circuit is different. Many experiments have produced the following model for raffinase regulation which involves two unlinked regulatory genes **reg1** and **reg2**. **Reg2** is a repressor of the raffinase gene, **reg1** is a repressor of **reg2**, and glucose inhibits the **reg1** repressor. A diagram of this regulatory circuit and phenotypes of reg mutants are shown below.



Raffinase activity

Genotype	**- glucose**	**+ glucose**
wild type	high	low
reg1$^-$	low	low
reg2$^-$	high	high

You have isolated two interesting mutations in the **reg2** gene:

O_2^c : An operator mutation in the **reg2** gene that will not bind the **reg1** repressor.

Reg2^{-d} : A dominant mutation in the **reg2** gene that prevents the **reg2** repressor from binding its operator and will inactivate wild type **reg2** repressor when expressed in the same cell. (This mutation is analogous to the I^{-d} mutation in the *Lac* repressor)

(d 25 pts) Fill in the table below with the raffinase levels expected for strains of the given genotypes.

	Genotype	Raffinase activity - glucose	Raffinase activity + glucose
1)	O_2^c	Low	Low
2)	**Reg2^{-d}**	High	High
3)	**Reg2^{-d}** / F' **Reg2^{+}**	High	High
4)	O_2^c **Reg2^{-d}** / F' **Reg2^{+}**	High	High
5)	O_2^c / F' **Reg2^{-d}**	Low	High

(For these strains assume that **Reg2^{-d}** exerts its dominant negative effect on **Reg2^{+}** regardless of the relative levels of mutant and wild-type proteins)

Name:_____

Name:_____

Recitation day_____, time_____, TA_____

7.03 Exam 3

November 23, 1994

Write your name on all 9 pages.
Indicate your recitation section on this page.
Write all answers on this handout only.
Exam begins at 11:05 and ends at 11:55.
Time will be announced when 5 and 1 minutes remain.

Problem 1	25
Problem 2	40
Problem 3	35
Total	100 points

Name: Answer Key

1. You have decided to study genetics in Truffula trees (see Seuss *et al.*). While studying leaf color, you find two different mutant strains that have pink leaves and are true breeding. Wild-type Truffula trees have red leaves. To analyze the mutants you decide to cross the two pink strains and obtain the following results:

$P_1P_1P_2P_2$ $P_2P_2P_1P_1$

pink1 x pink2

↓

F1 all red $P_1P_1P_2P_2$

↓

F2	**red**		**pink**		**white**
	9	**:**	**6**	**:**	**1**

(a. 10 points) Assign genotypes to each of the phenotypes in the F2 generation. Propose an explanation for the production of pink and red colors.

(9) 3 + 3 = (6)

F_2 genotypes: Red P_1-P_2- Pink $P_1P_1P_2-$ or $P_1-P_2P_2$

White $P_1P_1P_2P_2$ (1)

Must be 2 pathways to make pink pigment $\xrightarrow{P_1}$ pink $\xrightarrow{P_2}$ pink

If Both Don't work get white

If Both Do work enough pink pigment gives a red tree

pink + pink → red

In the forest of your true breeding pink1 trees you find a rare red tree that appears to be a revertant. To analyze this revertant you cross it to a true breeding red strain:

P_1P_1 sup sup P_1P_1 ++

pink1 revertant X red

↓

F1 all red P_1P_1 + sup

↓

F2	**red**		**pink**
	13	**:**	**3**

Name: Answer Key

(b. 15 points) Give the genotypes of the pink[1] revertant and the F2 progeny, being sure to specify what is dominant and recessive. Explain the ratio of red to pink trees in the F2 progeny. Explain how the F2 genotypes result in pink and red phenotypes.

Genotypes: p' revertant must have had unlinked suppressor

pink' revertant p'p' sup sup

F_2 red $P_1\_+\_$ & $P_1\_$ sup sup & p_1p_1 sup sup

9 + 3 + 1 = 13

pink $p_1p_1+\_$ 3

The suppressor is recessive and only suppresses the homozygous P_1 mutant if the suppressor is homozygous

The P_1 mutation is still present and recessive to red P_1 allele

Question #2 omitted

Name:_____

Recitation day_____, time_____, TA_____

7.03 Final Exam

December 16, 1994

Write your name on all 15 pages.
Indicate your recitation section on this page.
Write all answers on this handout only.
Exam begins at 1:30 and ends at 4:30.
Time will be announced when 15, 5, and 1 minutes remain.

Problem 1	10
Problem 2	25
Problem 3	25
Problem 4	15
Problem 5	20
Problem 6	20
Problem 7	40
Problem 8	20
Problem 9	25
Total	200 points

Name:_____

Question #1 omitted

2. In yeast, the biochemical pathway to make adenine involves a number of enzymatic steps and the **ADE2** gene encodes one of these enzymes. Normally yeast makes white colonies but mutants defective in **ADE2** accumulate a chemical intermediate in the adenine pathway that is pigmented and makes the yeast colonies red. A strain that has an amber mutation in the **ADE2** gene is red and is unable to grow without adenine. You isolate three different revertants of this strain that produce white colonies.

(a. 5 pts) Revertant 1 contains an amber suppressor that is linked to **ADE2**. When revertant 1 is crossed to a wild-type strain the following types of tetrads are seen:

Type I	Type II	Type III
4 white	3 white:1red	2 white: 2 red

Of 100 tetrads, there are 75 of type I; 22 of type II; and 3 of type III.

Calculate the distance between the suppressor and ADE2 in cM.

linked b/c PD >> NPD

$$\frac{T+6NPD}{2\Sigma} \times 100 = \frac{22+18}{2\times 100} \times 100 = 20\ cM$$

Part b omitted

Name:_____

(c. 10 pts) At first you guess that revertant 3 also contains an amber suppressor. Then you find that although revertant 3 makes white colonies it requires adenine for growth whereas revertants 1 and 2 can grow on medium without adenine. When revertant 3 is crossed to wild type the same three tetrad types are seen as before:

Type I	Type II	Type III
4 white	3 white: 1 red	2 white: 2 red

When you test the tetrads for growth on adenine you find the following:

Type I	Type II	Type III
2 ade+: 2 ade-	3 ade-: 1 ade+	4 ade-

Thinking about the biochemical pathway for adenine synthesis and the fact that the red pigment is an intermediate in that pathway, propose an explanation for the behavior of revertant 3.

Revertant 3 is a mutation in a gene that is in the pathway before the red pigment is made. It is epistatic to ADE2, so double mutants will be white but will need Adenine for growth

Question 3 omitted

Name:_____

4. (15 pts). Circle whether each of the following statements is true (T) or false (F):

T (F) Homologous chromosomes segregate from each other during mitosis.

(T) F In meiosis recombination occurs after DNA replication.

T (F) In most organisms mitotic and meiotic recombination occur at similar levels.

T (F) It can be determined whether a nondisjunction event occurred in meiosis I or II if the parent is heterozygous for an RFLP unlinked to the centromere.

Parts of this question omitted

Name:_____

5. The pedigree on the following page shows the segregation of a very rare disease that is completely penetrant. The affected individuals are shaded. Also shown are the genotypes for two different RFLP markers. The A locus has alleles A1 and A2, and the B locus has alleles B1 and B2.

(a. 5 pts) What is the pattern of inhertance of the disease trait in this pedigree?

Aut Dom

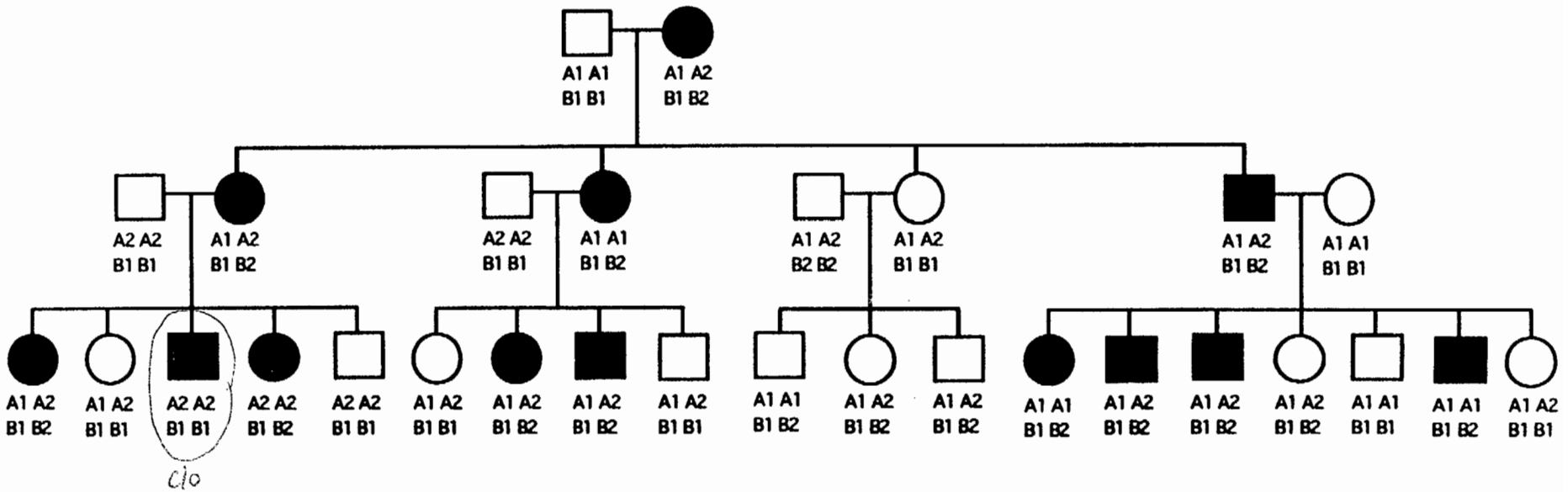
(b. 5 pts) Does the RFLP marker A appear linked or unlinked to the disease locus? If it appears linked, then which allele of this marker segregates with the disease in this pedigree?

No

(c. 5pts) Does the RFLP marker B appear linked or unlinked to the disease locus? If it appears linked, then which allele of this marker segregates with the disease in this pedigree?

Yes - B2 is travelling w/ disease allele

(d. 5pts) Circle any individuals in whom a crossover between the linked marker and the disease locus is revealed by the genotypes of their children.


A1 A1
B1 B1
A1 A2
B1 B2
A2 A2
B1 B1
A1 A2
B1 B2
A2 A2
B1 B1
A1 A1
B1 B2
A1 A2
B2 B2
A1 A2
B1 B1
A1 A2
B1 B2
A1 A1
B1 B1
A1 A2
B1 B2
A1 A2
B1 B1
A2 A2
B1 B1
A2 A2
B1 B2
A2 A2
B1 B1
A1 A2
B1 B1
A1 A2
B1 B2
A1 A2
B1 B2
A1 A2
B1 B1
A1 A1
B1 B2
A1 A2
B1 B2
A1 A2
B1 B2
A1 A2
B1 B2
A1 A1
B1 B1
A1 A1
B1 B2
A1 A2
B1 B1
c/o

Name:_____

6. Imagine an operon in *E. coli* that is required for synthesis of the vitamin thiamine. The genes for thiamine synthesis are controlled by a transcriptional activator that binds to a DNA site in the promoter to turn on transcription. Thiamine supplied in the medium binds to the activator protein and prevents binding to the DNA site and thereby turns off expression of the operon. You have isolated two new mutants in this operon: 1) **p-** is a mutation in the activator binding site that causes expression to be uninducible; and 2) **act^s** is a mutation in the gene for the activator protein that prevents binding to thiamine and causes constitutive expression.

(a. 5 pts) An F' carrying the entire operon with a **p-** mutation is introduced into a wild type strain. **Will expression of the operon be regulated, uninducible, or constitutive?**

reg.

(b. 5 pts) An F' carrying the entire operon with a **act^s** mutation is introduced into a wild type strain. **Will expression of the operon be regulated, uninducible, or constitutive?**

Const.

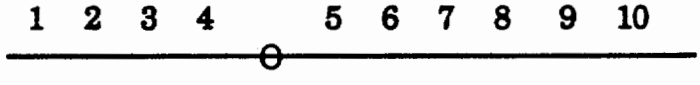
(c. 10 pts) The gene for the activator protein shows 20% linkage by P1 cotransduction to the promoter for the operon. A **p-** mutant is infected with P1 phage grown on an **act^s** host. Transductants that can express the operon are selected. **What fraction of these transductants will express the operon constitutively?**

20%

Name:_____

Question 7 omitted

8. To investigate sex determination in the mouse you collect from your colleagues a set of strains that are genotypically XX but phenotypically male. You also obtain another set that are genotypically XY but phenotypically female. You are pretty certain there must be a mouse Sry gene that specifies male development on the Y chromosome. You have ten molecular markers available to you along the Y chromosome.



You notice that many of the XX male strains contain translocations that attach part of the Y chromosome to the end of an autosome. You prepare DNA from the XX male and XY female strains, make Southern blots, and hybridize them to each of the Y marker probes. In the following table + signifies that the marker is present in the strain and – designates that it is absent.

	Y CHROMOSOME MARKERS									
	1	2	3	4	5	6	7	8	9	10
STRAIN										
XX male										
A	+	+	+	-	-	-	-	-	-	-
B	+	+	-	-	-	-	-	-	-	-
C	+	+	+	+	-	-	-	-	-	-
XY female										
A	-	-	-	-	+	+	+	+	+	+
B	+	-	-	-	+	+	+	+	+	+
C	+	-	+	+	+	+	+	+	+	+
D	+	+	+	+	+	+	+	+	+	+

(a. 10 pts) Are these data consistent with an Sry gene on the mouse Y chromosome? If so, to which genomic region does it map?

2

Part b omitted

Name:_____

9. (a. 5 pts) A recessive allele for microcephaly has a frequency q = 0.001 in a population in Hardy-Weinberg equilibrium. **What is the expected frequency of microcephalic individuals in this population?**

1×10^{-6}

(b. 5 pts) Mutations to produce new alleles for microcephaly occur at frequency $\mu = 10^{-8}$. **If the current frequency of microcephaly was established during human evolution when the fitness of microcephalic individuals was zero but there was a selective advantage for individuals heterozygous for microcephaly, what was the value of the heterozygous advantage h?**

$\mu = 1 \times 10^{-8}$

$q = h/s \qquad s = 1$

$h = 1 \times 10^{-3}$

(c. 5 pts) In modern times say that the fitness of microcephalics has increased to 0.25 and the heterozygous advantage has become zero. **Assuming that these conditions hold for many centuries, what will the steady state allele frequency for microcephaly eventually become?**

$s = 0.75 \qquad q = \sqrt{\mu/s} = 1.15 \times 10^{-4}$

$h = 0$

(d. 5 pts) What is the inbreeding coefficient F for a second-cousin mating?

$1/64$

Name:_____

(e. 5 pts) Given that in the general population the frequency of second-cousin matings is 0.016, compute the fraction of microcephalics whose parents are second cousins.

$$0.016 \cdot \frac{1}{64} \cdot 9 = 2.5 \times 10^{-7}$$

$$\frac{2.5 \times 10^{-7}}{10^{-6}} = \boxed{\frac{1}{4}}$$