BIO304 S03 Exam III Name KEY
(Note that you must show your work to receive partial credit) ID # _____

1. Fill in the blanks with the **best** answer from the list provided. (1 pt each)

euploid	polyploid	aneuploid	tetraploid
haploid	monoploid	diploid	triploid
monosomy	disomy	trisomy	amphidiploid
allopolyploid	autopolyploid	homeologous	Down syndrome
Turner syndrome	Klinefelter syndrome	Patau syndrome	duplication
deletion	translocation	pericentric inversion	paracentric inversion
acentric	dicentric	pseudodominance	pseudolinkage
synteny	reverse genetics	knockout	indel
frameshift	transposable elements	antisense RNA	dsRNAi
somatic mutation	germline mutation	clone	genetic screen
genetic selection	auxotrophy	prototrophy	conditional
modifier	null	hypermorph	neomorph
amorph	leaky	gain-of-function	promoter
TATA-binding protein	promoter-proximal element	transcriptional activation factor (TAF)	maternal effect mutations
enhancer	silencer	operon	inducer
operator	catabolite repression	helix-loop-helix	helix-turn-helix
zinc finger	leucine zipper	homeodomain	HMG box
imprinting	complementation	codominance	incomplete dominance
epistasis	suppression	enhancement	penetrance
expressivity	master switch	pattern formation	morphogen
gap genes	allosteric	segment polarity genes	homeotic selectors
pair-rule genes	autoregulation	population	fitness

Downs Syndrome — Human condition arising from having three chromosome 21 copies
trisomy — Chromosomal condition for the above
conditional — Mutation that shows a phenotype only under certain circumstances
genetic selection — Type of mutational scheme in which only organisms of the desired genotype are recovered
acentric — Chromosome with no centromere
fitness — Ability of a given genotype to survive
leaky — Mutation that causes reduced function
master switch — Mechanism in which a single gene controls a developmental cascade
pattern formation — Process in which cell fates are determined based upon position within the organism
allosteric — Protein for which activity is regulated by changes in structural conformation
homeotic selectors — Genes that code for factors that determine the specific identity of individual body segments
epistasis OR suppression — Relationship in which mutations in one gene override the phenotype caused by mutation of a second gene
deletion OR duplication — One type of unbalanced rearrangement
penetrance — Measure of the proportion of individuals with given genotype that show the expected phenotype
somatic mutation — Type of genetic defect that cannot be inherited by progeny

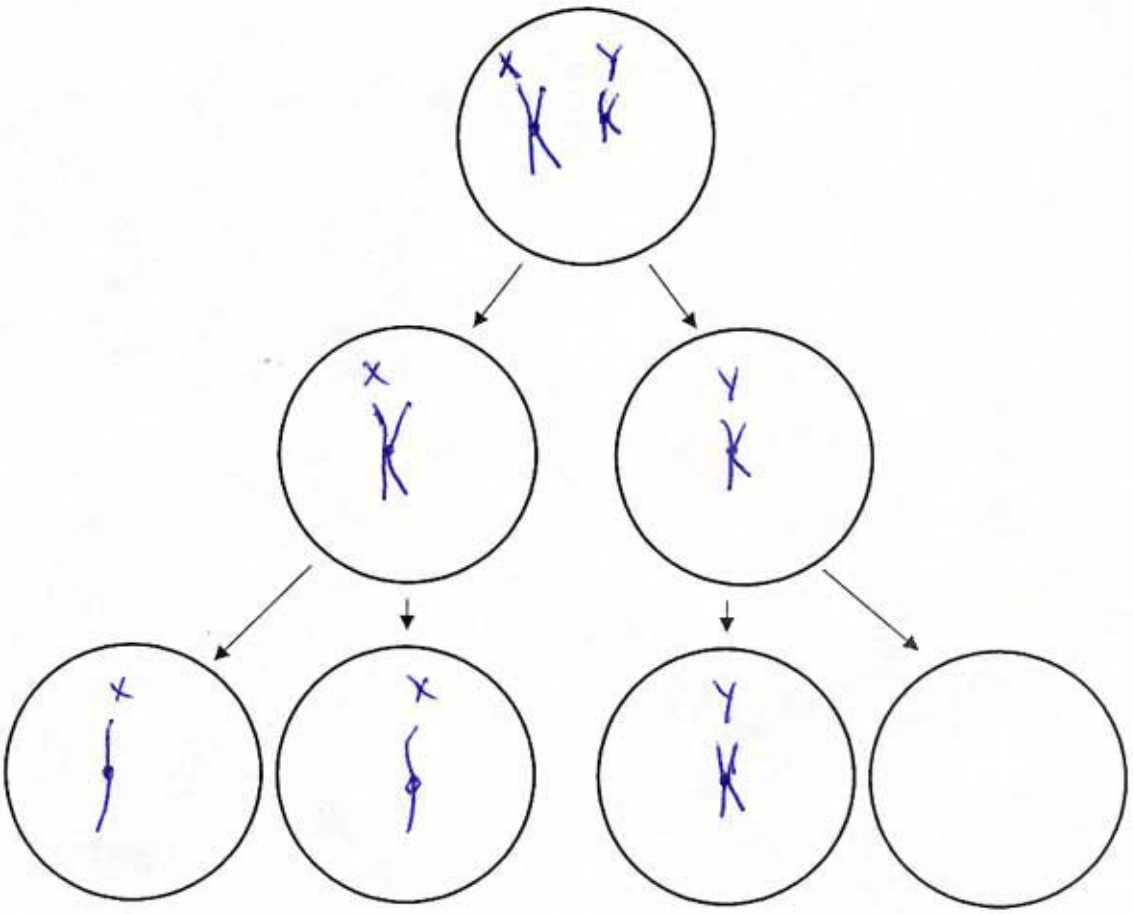
2. A couple has a son who, upon inspection of his sex chromosomes, is found to be XYY.
   a) What is the expected phenotype of this chromosomal aberration? (2 pts)

Normal (Some studies have suggested an increased tendency for violent behavior, but this is questionable)

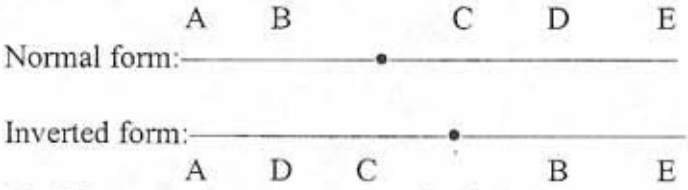
   b) In what parent could the nondisjunction causing this aberration have happened? (2 pts)

father

   c) Diagram the segregation of the X and Y chromosomes during the two meiotic divisions that gave rise to the aberrant gamete. (8 pts)



3. A rearrangement is present in chromosome 2 of mice. The normal form and rearranged forms are illustrated below:

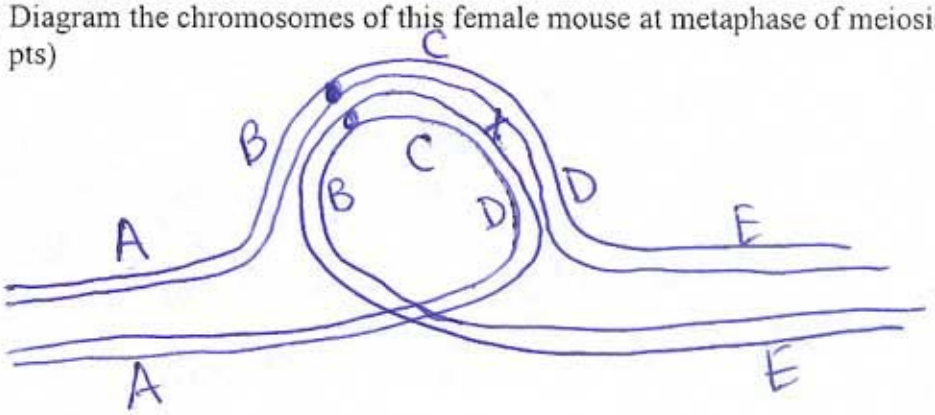


(a) What type of rearrangement is this? Be as specific as possible. (2 pts)

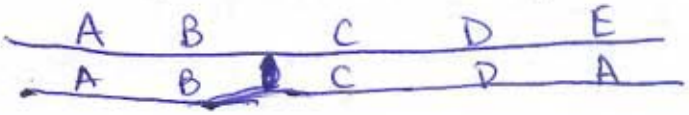
Pericentric inversion

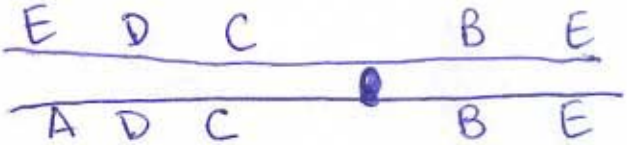
(b) A female mouse is heterozygous, carrying one each of the two forms of the chromosome shown above.

(1) Diagram the chromosomes of this female mouse at metaphase of meiosis I. (4 pts)



(2) Assume that a crossover occurred during meiosis in the female mouse between *C* and *D*. Diagram the chromosomes at anaphase of meiosis I (4 pts)





(3)How many of the gametes produced by the meiosis described above will generate viable offspring? (2 pts)

Two out of four

4. A researcher wishes to learn about the process by which *Drosophila* eye pigmentation occurs. To learn more about the genetics, she performs a mutagenesis screen and recovers four recessive mutations (which she names *a-d*) that cause the flies to have white eyes instead of the wild-type red eye color. After establishing lines of each of these new mutations, she crosses each of the homozygous mutants to each of the others and recovers the F1 progeny. She then testcrosses the F1 progeny to a line that is homozygous mutant for all of the new alleles. Below is a table of her results.

Cross	F1 Phenotypes	Testcross Phenotypes
a x b	all red eyes	75% red, 25% white
a x c	all red eyes	75% red, 25% white
a x d	all white eyes	all white eyes
b x c	all red eyes	90% red, 10% white
b x d	all red eyes	75% red, 25% white
c x d	all red eyes	75% red, 25% white

a) What is the common name we use to describe the type of cross used to generate the F1 progeny? (2 pts)

complementation test

b) How many genes are represented by these four mutants? (4 pts)

3 (a and d are allelic)

c) What can you say about the chromosomal locations of the genes involved? (4 pts)

b + c are genetically linked (ie on the same chromosome)

5. The table below represents data obtained from mutations of different components of the lac operon. The three mutations, a⁻, b⁻ and c⁻, are mutations in lac I, O, and Z, though not necessarily in that order. The effects of each of mutant on lacZ activity were determined by including X-gal in the media. Remember that β-galactosidase can cleave X-gal into a blue product. The color of colonies from each genotype in the presence or absence of lactose is indicated in the table. Assume there is no source of glucose in the medium.

	Genotype of bacteria	With Lactose	Without Lactose
1	$a^+ b^+ c^+$	blue	white
2	$a^+ b^+ c^-$	blue	blue
3	$a^- b^- c^+$	white	white
4	$a^- b^+ c^-$	white	white
5	$a^+ b^+ c^+ / a^- b^- c^-$	blue	white
6	$a^+ b^+ c^- / a^- b^- c^+$	blue	white
7	$a^+ b^- c^+ / a^+ b^+ c^-$	blue	white

Based on your knowledge of the lac operon, indicate which gene (I, O or Z) each of the mutants (a, b, or c) corresponds to. Support your answer by explaining your logic. (10 pts)

To get no activity, βgal must be mutant (white colonies)
3 tells us that either a or b must be lacZ⁻
4 tells us that either a or c must be lacZ, ∴

LacZ = a

Knowing this we can look for the trans-regulatory activity of lac I + the cis activity of lac O!
In 6, activity is normally regulated by using the wild-type activity of c from the F episome to control activity of the chromosomal copy of lacZ (a) this indicates trans-activity ∴ LacI = c

So lac O = b

6. A particular species of wild flower normally has purple petals. However, there are two true-breeding mutant lines of this species, one with red flowers and one with blue flowers. To understand the genetics of flower coloration in this species, crosses were made between each of the mutant and wild-type lines, with the results shown in the table below.

Parents	F1 phenotypes	F2 phenotypes
purple x red	all purple	101 purple, 33 red
purple x blue	all purple	192 purple, 63 blue
red x blue	all purple	272 purple, 121 red, 89 blue

a) How many different genes are represented by these mutant alleles? (2 pts)

2

b) Define symbols to represent each allele of each gene involved here. Use those symbols to indicate the gene and allele relationships that are uncovered by these crosses. In other words, what are the genotypes of each of the phenotypic classes observed in the F1 and F2 generations? You may find it helpful to start by drawing a Punnett square. (9 pts)

R = wild-type red allele r = recessive mutant red
B = wild-type blue allele b = recessive mutant blue

Punnett Square of F1:

	RB	Rb	rB	rb
RB	RR; BB 1	RR; Bb 2	Rr; BB 3	Rr, Bb 4
Rb	RR; Bb 5	RR; bb 6	Rr; Bb 7	Rr, bb 8
rB	Rr, BB 9	Rr Bb 10	rr BB 11	rr Bb 12
rb	Rr Bb 13	Rr bb 14	rr, Bb 15	rr, bb 16

Ratio of F1 progeny in the dihybrid cross is 9:4:3 (purple, red, blue) ∴ red is epistatic ove
blue in the double homozygote (16)

R_, B_ = purple R_, bb = blue rr, B_ = re
rr, bb = red

7. When homozygous, a small deletion in *Drosophila* causes embryos to be missing all of the even numbered segments.

a). Is this likely to be due to the loss of a gap gene, maternal gene, segment polarity gene, pair-rule gene, or homeotic selector gene? (3 pts)

pair-rule gene

b). Upon molecular mapping of the deletion, it is found that four genes are missing in this mutation. The normal embryonic expression in wild-type for each of these genes was determined using in situ hybridization to RNA. The results are shown below. Which of these genes is likely to be responsible for the patterning phenotype described above? (3pts)

1  2  3  4 

2 - it is expressed in every other segment (7 stripes out of 14 segments)

c) The above in situ hybridization is repeated, but this time in embryos that are mutant for the gap gene hunchback. Would you expect to see the same expression pattern for the gene you selected in part b? Why or why not? (3 pts)

No, because portions of the embryo would be lost due to mutation of hunchback. This would alter pair-rule gene expression because the gap genes control pair-rule expression.

d) The reciprocal experiment is also performed, that is, in situ hybridization to hunchback RNA is performed in embryos that are homozygous mutant for the small deletion. Would you expect to see the same expression pattern for hunchback in wild-type and the deletion homozygote? Why or why not? (3 pts)

Yes, because gap genes are expressed before pair-rule genes.

8. In a population of rats, the fitnesses and frequencies of genotypes at present are:

	Frequency	Fitness
HH	0.4	1.0
Hh	0.5	0.8
hh	0.1	0.6

What will be the frequency of the *h* allele in the next generation? (8 pts)

In ~~this~~ generation:

freq. H = p = 0.4 + ½(0.5) = 0.65

freq. h = q = 0.1 + ½(0.5) = 0.35

In next generation:

freq. HH = $p^2 W_{HH}$ = $(0.65)^2(1)$ = 0.4225

" Hh = $2pq\, W_{Hh}$ = 2(0.65)(0.35)(0.8) = 0.364

" hh = $q^2 W_{hh}$ = $(0.35)^2(0.6)$ = 0.0735

Total = 0.86

Adjust to total of 1

HH = $\frac{0.4225}{0.86}$ = 0.491

Hh = $\frac{0.364}{0.86}$ = 0.423

hh = $\frac{0.0735}{0.86}$ = 0.0854

h = (0.0854) + ½(0.423) = **0.30**

OR the short way:

$q' = \text{freq } a/a + \frac{1}{2}\text{freq } A/a = q^2 \frac{W_{aa}}{\bar{W}} + \frac{1}{2}\left(2pq \frac{W_{Ao}}{\bar{W}}\right.$

$\bar{W} = (0.4)(1) + (0.5)(0.8) + (0.1)(0.6) = 0.86$

$p = 0.4 + \frac{1}{2}(0.5) = 0.65$ $\quad q = 0.1 + \frac{1}{2}(0.5) = 0.35$

$\therefore\ q' = (0.35)^2\left(\frac{0.6}{0.86}\right) + \frac{1}{2}\left(2 \times 0.65 \times 0.35 \times \left(\frac{0.8}{0.86}\right)\right) =$ **0.30**

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9. The table below gives the percentage of homozygous individuals for alleles *A* and *a* in several different populations.

Population	A/A	a/a	A/a
1	80 %	1 %	19%
2	48 %	24 %	28%
3	60 %	5 %	35%
4	36 %	16 %	48%

For each population, indicate whether it is in Hardy-Weinberg equilibrium. You **must** show your work to receive credit. (10 pts)

Population 1 → freq. A = $p = 0.8 + \frac{1}{2}(0.19) = 0.895$

" $a = q = 0.01 + \frac{1}{2}(0.19) = 0.105$

In next generation, $A/A = p^2 = (0.895)^2 = 0.801$

$A/a = 2pq = 2(0.895)(0.105) = 0.188$

$a/a = q^2 = (0.105)^2 = 0.011$

These are all very close to original generation → YES

Population 2 → freq $A = p = 0.48 + \frac{1}{2}(0.28) = 0.62$

$a = q = 0.24 + \frac{1}{2}(0.28) = 0.38$

In next generation, $A/A = p^2 = (0.62)^2 = 0.384$

$A/a = 2pq = 2(0.62)(0.38) = 0.471$

$a/a = q^2 = (0.38)^2 = 0.144$

All are substantially different from original generation → NO

Population 3 → $p = 0.6 + \frac{1}{2}(0.35) = 0.775$

$q = 0.05 + \frac{1}{2}(0.35) = 0.225$

In next generation, $A/A = p^2 = (0.775)^2 = 0.601$

$A/a = 2pq = 2(0.775)(0.225) = 0.349$

$a/a = q^2 = (0.225)^2 = 0.051$

Same as original → YES

Population 4 → $p = 0.36 + \frac{1}{2}(0.48) = 0.6$

$q = 0.16 + \frac{1}{2}(0.48) = 0.4$

Next generation, $A/A = p^2 = (0.6)^2 = 0.36$

$A/a = 2pq = 2(0.6)(0.4) = 0.48$

$a/a = q^2 = (0.4)^2 = 0.16$

Identical to original → YES