BIO 304 Genetics (Fall 2003) **Exam #2** **Name_____KEY_____**
SSN_____

transformation	conditional mutation	penetrance	expressivity
Southern blotting	hybridization	epistasis	co-dominance
nonsense mutation	translocation	amplification	transposase
missense mutation	alkyltransferase	vector	long terminal repeats
electrophoresis	nitrocellulose	VNTR	ampicillin
transduction	silent mutation	inverted repeats	polymerase
reverse transcriptase	heterokaryon	bacteriophage	duplication
transposable element	plasmid	probe	restriction enzyme
dicentric	transversion	episome	suppression
lysogeny	complementation	histone	dideoxy sequencing
Northern blotting	tautomerization	inversion	F factor
nondisjunction	autopolyploid	ligase	polymorphism
frameshift	Prozac	expression vector	independent assortment
conjugation	reversion	cDNA	two-hybrid
sister chromatids	RFLP	biotechnology	heteroduplex DNA
complementation	nonsense suppressor	transition	tautomeric shift
thymine dimerization	intercalating agent	polyploid	aneuploid
temperate phage	prophage	loss-of-function	gain-of-function
allopolyploid	clone	synteny	replicative transposition
glycosylase	catalase	lytic	conservative transposition
monoploid	PCR	pleiotropy	homeologous
auxotroph	library	stem cells	crossover

Use one of the above terms to <u>best</u> complete each sentence #1-15 below. (2 pts. each)

1. _____cDNA_____ is a DNA copy of an RNA molecule.

2. ___reverse transcriptase__ is an RNA-dependent DNA polymerase.

3. Knockout mice are created by replacing a normal gene segment with a modified segment within embryonic _stem cells_____, then using the latter to create a chimeric embryo.

4. __transformation____ is a method of DNA transfer in bacteria in which environmental DNA is taken up through the cell wall and membrane of the cell.

5. A ___library_____ is a collection of DNA clones that represent the genome or RNA population of an organism or tissue.

6. A restriction site difference that is polymorphic within a population can be used to carry out _____RFLP_____ mapping of neighboring genes.

7. ___histone_____ genes commonly exist in tandem repetitive arrays in eukayote genomes.

8. ____transposase_____ is a transposon-encoded protein that mediates movement of the transposon.

9. ____two-hybrid_____ analysis detects physical interactions between two proteins in yeast cells, by fusing each protein to a separate domain of the yeast GAL4 regulatory protein and studying the ability of the combination to activate a UAS-*lacZ* gene.

10. _____VNTR_____ sequences are one- to five-kilobase sequences consisting of variable numbers of repeat units, each unit being 15 to 100 nucleotides in length.

11. _____catalase_____ is an enzyme that prevents DNA damage by reducing reactive oxygen species.

12. A type of DNA damage caused typically by UV light is ___thymine dimerization_____.

13. A karyotype that has an extra set of chromosomes is a type of ____polyploid____ genome.

14. Retrotransposons have sequences at the ends that are best described as ___long terminal repeats_____.

15. ___translocation_____ is a rearrangement that moves a segment of DNA to a new chromosome.

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16. Wild-type tigers are orange and black with white stripes, have eyes that glow in the dark and have bad breath. A mutant strain of tigers is developed in which animals are albino (*a*), have glowless eyes (*g*) and sweet breath (*b*), each determined by a single gene. True-breeding wild-type females were crossed to males having all three mutant traits, producing an F1 of entirely wild-type animals. F1 females were testcrossed to *a g b* males, producing an astonishing number of progeny:

Phenotype	Number	
$a^+ g^+ b^+$	429	parental types
$a\ g\ b$	433	parental types
$a^+ g\ b$	55	SCO I (*a-b*) - 108
$a\ g^+ b^+$	53	SCO I (*a-b*) - 108
$a^+ g\ b^+$	13	SCO II (*g-b*) - 28
$a\ g^+ b$	15	SCO II (*g-b*) - 28
$a\ g\ b^+$	2	DCO - 2
	1000	

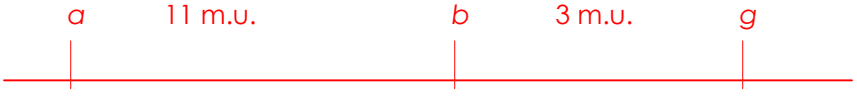
a. Draw a map of these three genes, indicating their order and the map distances between them (10 pts.)

108 + 2 = 110

110 ÷ 1000 = .11

28 + 2 = 30

30 ÷ 1000 = .03



b. Calculate the coefficient of coincidence and the interference value for these results. (3 pts.)

COC = observed DCO ÷ expected DCO = 2 ÷ [.03 X .11] = 2 / 3.3 = 0.61

interference = 1- COC = 1 – 0.61 = 0.39

17. A cross is made between an *E. coli* Hfr strain that is $a^+ b^+ d^+$ and an F^- strain that is $a^- b^- d^-$. Interrupted mating studies showed that b^+ enters the recipient strain last. The b^+ recombinants were then tested for the presence of the *a* and *d* alleles, producing the following results:

$a^+ b^+ d^+$	326	parental type
$a^- b^+ d^+$	2	DCO
$a^+ b^+ d^-$	14	SCO I (*a-d*)
$a^- b^+ d^-$	58	SCO II (a-b)

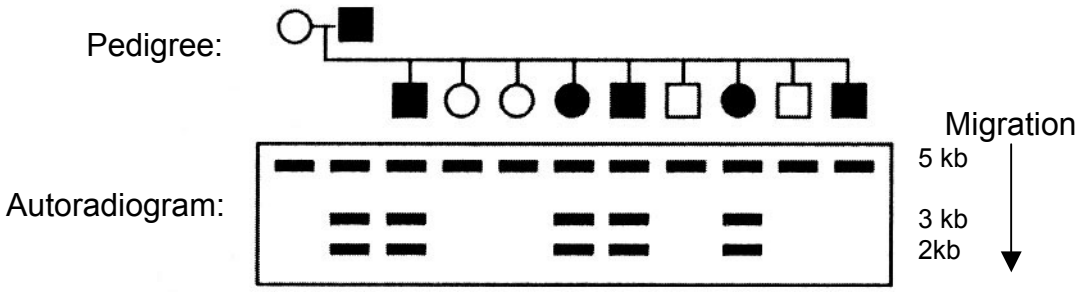
	400	

Draw a map of these genes, showing the gene order and frequencies of recombination between each pair. (6 pts.)

14 + 2 = 16
16 ÷ 400 = .04

58 + 2 = 60
60 ÷ 400 = 0.15

18. DNA studies were performed on a large family that shows an autosomal dominant disease of late onset. A DNA sample from each member is digested with *Taq*I and subjected to Southern blot analysis, using a particular unique sequence probe:



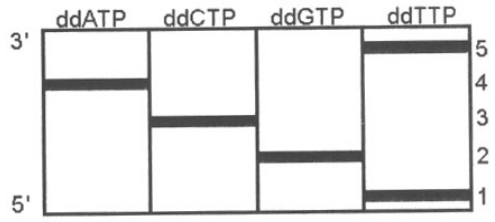
a. What is the relationship between the restriction pattern observed with this probe and the gene for the disease? (7 pts.)

The 3+2 kb morph is linked to the disease gene; the 5 kb morph is linked to its wild-type allele. This correlation applies in 8 of 9 progeny.

b. How do you explain the pattern seen in the rightmost son? (3 pts.)

A recombination event occurred between the two sites in the father, producing the disease allele in *cis* with the 5 kb morph.

19. Using dideoxy nucleotide DNA sequencing, you observe the following autoradiogram:



What is the 5'-3' nucleotide sequence of the <u>template</u> strand that you have sequenced? (6 pts.)

5' A T G C A 3'

20. Geneticists working on the Human Genome Project, focusing on one band of chromosome 11, had a collection of eight cloned fragments (clones 1-8). They also determined that seven sequence-tagged sites (STSa-g) were distributed among these clones. The STS distribution on the various clones was as follows:

Clone	STS content
1	a, b, e, f
2	a, b
3	f
4	a, c, f
5	c, d
6	e
7	g
8	e, g

Arrange these clones into a contig, showing the regions of overlap and positions of STSs. (8 pts.)

STS: d c f a b e g

1 f a b e

2 a b

3 f

8 e g

4 c f a

5 d c

6 e

7 g

21. For each of the following aberrant sex chromosome configurations, a nondisjunction event is responsible. Indicate during which meiotic division the nondisjunction could have occurred assuming first that it occurred in the mother, then assuming it occurred in the father. Your possible answers for each are: meiosis I, meiosis II, either, or not possible. (12 pts.)

Offspring	Meiotic division if nondisjunction occurred in mother	Meiotic division if nondisjunction occurred in father
XXY	either	Meiosis I
XYY	not possible	Meiosis II
XXX	either	Meiosis II

22. What is meant by missense, nonsense, and synonymous mutations? Which of the three is most likely to disrupt gene function and why? (6 pts.)

Missense mutations are changes that cause the substitution of one amino acid for another in the encoded protein.
Nonsense mutations are changes that cause the substitution of a stop codon for an amino acid in the encoded protein.
Synonymous mutations are changes in the nucleic acid sequence in the coding region of a gene that do not cause a change in the encoded protein.

Nonsense mutations are most likely to disrupt gene function because they cause the truncation of the encoded protein, deleting all amino acids that follow the mutation in the coding sequence.

23. Indicate with an "X" which of the following enzymes act in each of the listed *E. coli* mutation repair systems. Enzymes may be active in none or more than one of the listed systems. (6 pts)

	Excision repair	Recombinational repair	Mismatch repair	UV photodimer splitting
DNA polymerase I	X	(either OK)	X	
Photolyase				X
DNA ligase	X	(either OK)	X	
Glycoslylase	X			
RecA		X		

(One point for each correct X, minus one point for each incorrect X, sum not < 0 or >7)