BIO 304 Genetics **Exam #3** **Name**_____
SSN_____

RFLP	transposon	inducible	biotechnology	genetic drift
T7 promoter	Ti plasmid	T DNA	*Agrobacterium*	auxotrophic
mutation	knockout	*bicoid (bcd)*	*nanos (nos)*	reverse genetics
gene replacement	dwarfism	gene therapy	PCR	polymorphism
structural genomics	repetitive	dispersed	tandem	telomerase
chromatids	alleles	genes	wild type	polycistronic
satellite	dominant	leaky	gain-of-function	loss-of-function
centromeric	VNTR	fingerprint	SSLP	*in situ* hybridization
Sendai virus	BAC vector	STSs	ESTs	contig
library	ORFs	robotics	two-hybrid analysis	complementary
IS elements	plasmid	R factors	transposase	functional genomics
homeotic	hybridization	nucleosome	telomere	hybrid dysgenesis
retrovirus	P element	activator	retrotransposon	reverse transcriptase
repressor	effector	lactose	allosteric	catabolite repression
cAMP	*lac* operon	TATA box	enhancer	silencer
receptor	oncogene	numerator	*Sxl(Sex lethal)*	autoregulatory
TDF	enumerator	zygotic	*hunchback (hb)*	maternal effect
migration	mutation	homeotic	cardinal genes	recombination
pair-rule	gap	selection	segment polarity	*dorsal (dl)*

Use one of the above terms to best complete each sentence #1-20 below. (2 pts. each)

1. ____ESTs_____ are cDNA sequences used to identify genomic clones and sequences corresponding to transcribed genes.

2. __functional genomics__ is the study of how all genes of an organism are expressed and how their products interact.

3. The ____TATA box_____ is a short DNA sequence found upstream from the transcription start site in many eukaryotic genes at which protein binding is the first step in formation of a transcription initiation complex .

4. ___catabolite repression_____ is a transcriptional regulation system in *E. coli* whereby glucose availability diminishes expression of genes required for utilization of other sugars.

5. A cancer-causing gene is called a(n) ___oncogene_____.

6. ___biotechnology_____ is the use of genetically engineered organisms to produce commercially useful compounds.

7. An array of overlapping genomic clones is called a(n) _____contig_____.

8. ____enhancers_____ are DNA regulatory elements near eukaryotic genes that bind activator proteins and positively regulate gene expression.

9. Most mutations of tumor suppressor genes act as recessive _____loss-of-function_____ mutations.

10. ___polycistronic_____ mRNA encodes multiple, different polypeptide chains.

11. Binding of lactose to the *lac* repressor causes a slight change in the tertiary structure of the DNA-binding domain of the repressor, called a(n) ___allosteric_____ change.

12. ____retrotransposon_____ is the name given to a mobile genetic element that moves via an RNA intermediate.

13. Alternative alleles or chromosome rearrangements that are found in natural populations of organisms are called ____polymorphism_____.

14. ____*TDF*_____ is a Y-linked gene that specifies male differentiation of mammalian embryos.

15. Chromosome ends are maintained in eukaryotic cells by a special reverse transcriptase called ___telomerase_____.

16. Small population size frequently contributes to _____genetic drift_____, which results in genetic variation even among genes that are not subject to natural selection.

17. To analyze a mouse gene for which known mutations are not available one can carry out a deliberate gene disruption experiment to produce a(n) __knockout_____ mutation.

18. Highly repetitive, simple-sequence repeats, often called satellite DNA, are primarily located in _____centromeric_____ chromosome regions.

19. Transcription of the ___*Sxl*_____ gene in early *Drosophila* female embryos is the primary binary switch controlling sex differentiation.

20. Localization of _____*bicoid*_____ RNA at the anterior end of the *Drosophila* egg determines anterior fate to cells originating in that region of the embryo.

21. In mice and flies, ____homeotic_____ mutations result in developmental transformation of the identities of body segments.

22. Mutations of _____pair-rule_____ genes in flies result in embryos with one-half the normal number of segments.

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23. Retinoblastoma, a rare cancer of the eye, is due to the presence of cells that are homozygous for defective alleles (*rb*) of the tumor suppressor gene *RB*. In the most common form of retinoblastoma, called sporadic, this cancer is diagnosed in persons for whom no family history of the disease is known, neither in ancestors nor descendants of the affected person. A rarer form, called hereditary retinoblastoma is transmitted as an autosomal dominant trait. Compare the genetic mechanisms that give rise to sporadic and hereditary retinoblastoma. (10 pts.)

Sporadic: two-step mutation mechanism
zygote is homozygous for normal gene copies (*RB/RB*)
mutation/loss of both normal gene copies required for retinoblastoma

Hereditary: one-step mutation mechanism
zygote is heterozygous for normal and mutant gene copies (*Rb/rb*)
mutation/loss of the normal gene copy results in retinoblastoma

24. In a population of 854 mice, there are two alleles for the *A* locus (*A1* and *A2*). Tests show that in this population there are 384 mice of genotype *A1/A1*, 210 of A1/A2, and 260 of *A2/A2*. What are the frequencies of these two alleles in this population? (5 pts.)

$$p\,(A1) = \frac{384 + [\,\tfrac{1}{2} \times 210]}{854} = 0.57 \qquad p(A2) = \frac{260 + [\,\tfrac{1}{2} \times 210]}{854} = 0.43$$

25. Ten per cent of the males of an infinitely large, randomly mating mouse population are colorblind, due to a sex-linked recessive allele. A representative group of 1000 from this population migrates to an island, where there already 1000 inhabitants (also a randomly mating population) and where 40 per cent of males are colorblind (same allele). Assuming that Hardy-Weinberg equilibrium applies to both populations and to the mixed population formed after the migration, answer the following:

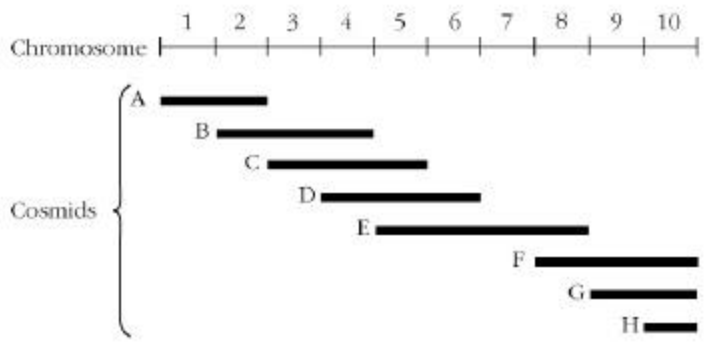
a. What fraction of males can be expected to be colorblind in the generations after the arrival of immigrants? (5 pts.)

Because males are hemizygous for the sex-linked gene, *p* in the infinitely large population is 0.10 and *p* in the island population (pre-migration) is 0.40. The newly constituted population is half immigrant and half former island population. Thus, *p* for the newly constituted population will be [0.10 + 0.40] / 2 or 0.25.

b. What fraction of females can be expected to be colorblind in the generations after the arrival of immigrants? (5 pts.)

$$p^2 = 0.25 \times 0.25 = 0.625 = 6.25\%$$

26. A cloned region (segments 1-10) of *Caenorhabditis* chromosome 2 is represented by the overlapping array of cosmid clones shown below:



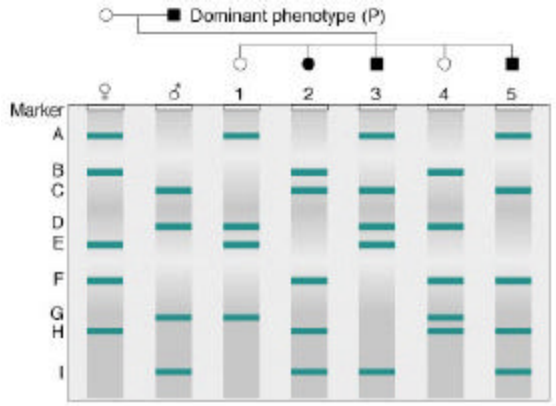
a. A cloned gene *x* hybridizes only with cosmids C, D, and E. Within which chromosome segment does *x* reside? (2 pts.)

Segment 5

b. A cloned gene *y* hybridizes only with cosmids E and F. Within which chromosome segment does *y* reside? (2 pts.)

Segment 8

27. DNA fingerprint analysis using a VNTR probe is carried out on a family in which one parent and three of five offspring exhibit the dominant trait P. The pedigree and Southern blot results are shown below. Answer each of the following questions: (2 pts each)



a. To which VNTR marker(s) is the dominant gene apparently linked?

C and I

b. Name two VNTR marker pairs that behave as if they are linked.

C and I F and H

c. Name at least three VNTR marker pairs that behave as if they were allelic?

A and B E and F E and H G and I G and C

28. The *lac* operon of *E. coli* includes the repressor-encoding *I* gene, the promoter *P*, operator *O*, and enzyme structural genes for â-galactosidase (*Z*) and two other proteins (which we will ignore). For each of the following growth conditions, what level of â-galactosidase do you predict (use "+" for high levels, "-" for low levels)? (15 pts.)

	Genotype	no lactose, no glucose	+lactose, no glucose	+lactose, +glucose
a.	$I^+P^+O^+Z^+$	-	+	-
b.	$I^-P^+O^+Z^+$	+	+	-
c.	$I^+P^+O^+Z^-$	-	-	-
d.	$I^+P^+O^CZ^+$	+	+	-
e.	$I^-P^+O^+Z^+$ / $I^+P^+O^+Z^-$	-	+	-