# Final Exam Practice

Final Exam is on Monday, DECEMBER 13 9:00 AM - 12 NOON

# BRING PICTURE I.D.

Exam Review on Thursday, Dec. 9 (new material only) 7-9 PM

Exam Tutorial Friday, Dec 10th 1-3 PM

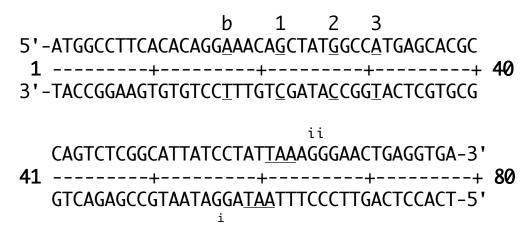
In the space provided next to each definition or description, **clearly** write the letter of the appropriate term from the list of terms given on the last page.

 A short, single-stranded DNA that serves as the necessary starting material for the synthesis of the new DNA strand in PCR
 The synthesis of DNA using DNA as a template
 The building blocks of DNA and RNA
 The synthesis of protein using information encoded in mRNA
 The location in a eukaryotic cell where the electron transport chain occurs
 The major component of cell membranes
 The genetic composition of an organism
 A gene that lies on one of the sex chromosomes
 An organism without membrane-bound organelles
 A cell with 1n chromosomes
 The building blocks of proteins
 A cell with 2n chromosomes
 A major source of energy that has the general formula $(CH_2O)_n$
 An enzyme needed for completion of lagging strand synthesis, but not leading strand synthesis
 The synthesis of RNA using one strand of DNA as a template
 An observed characteristic of an organism

#### Question 1, continued

- \_\_\_\_\_ A DNA molecule that is distinct from the chromosome; this molecule can be used to move foreign DNA in or out of a cell
- \_\_\_\_ The DNA from a eukaryote formed by the enzyme reverse transcriptase; this DNA lacks introns
- \_\_\_\_\_ An organism with 2 identical alleles for the same gene
- \_\_\_\_ A membrane protein involved in signal transduction; activation involves binding a GTP molecule
- \_\_\_\_\_ An organism with genetic material inside a nucleus
- \_\_\_\_\_ An organism with 2 different alleles for the same gene
- \_\_\_\_\_ A measure of the affinity of an enzyme for its substrate
- \_\_\_\_\_ A gene that lies on any chromosome except the sex chromosomes
- \_\_\_\_\_ The membrane that surrounds the cell
- \_\_\_\_ One of the alternate forms of a gene found at a given locus on a chromosome
- \_\_\_\_ A technique for the rapid production of millions of copies of a particular region of DNA
- \_\_\_\_\_ Proteins with a signal sequence are directed to this cellular organelle

The following double-stranded DNA contains sequence of a eukaryotic gene:



a) Transcription begins at the underlined A/T at base pair 17 (**b**) and proceeds to the right. What are the first 12 nucleotides of the resulting mRNA? Indicate the 5' and 3' ends of the mRNA.

b) The first 7 amino acids of the protein encoded by this gene are:

### NH3+ -met-ala-met-ser-thr-pro-his-tyr....COO-

i) underline the nucleotides which correspond to the 5' untranslated region of the primary RNA transcript made from this gene.

ii) draw a box around the intron region in this gene.

c) Consider each of the following three mutations independently.

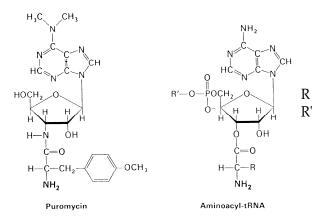
i) How would the resulting protein change if the underlined G/C base pair at position 22 (1) was deleted from the DNA sequence? Briefly explain.

ii) How would the resulting protein change if the underlined G/C base pair at position 27 (2) was changed to a C/G base pair? Briefly explain.

iii) How would the resulting protein change if the underlined A/T base pair at position 31 (3) was deleted from the DNA sequence? Briefly explain.

### **Question 2, continued**

d) Puromycin is an antibiotic that has an effect on both prokaryotes and eukaryotes. Puromycin, which is structurally similar to the aminoacyl terminus of an aminoacyl-tRNA (see diagram), inhibits protein synthesis by releasing nascent polypeptide chains before their synthesis is completed.



R represents the side group of the amino acid R' is the remainder of the tRNA

Explain how puromycin can affect this result on growing polypeptide chains and why the peptide chain is released.

### **Question 3**

a) Many patients are coming into the emergency room with a disease caused by an unknown pathogen! A doctor studies this pathogen in order to create a vaccine against it. She discovers that the infectious agent is an intracellular bacterium and its cell surface is coated with human-like proteins. Considering the mechanism of the pathogen, the doctor decides to generate a live-attenuated vaccine instead of a heat-killed vaccine.

i) What are the two advantages of using a live-attenuated vaccine vs. a heat killed vaccine in this case?

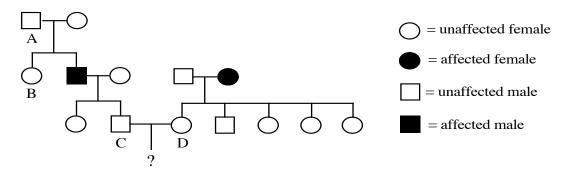
ii) What is a disadvantage of using a live-attenuated vaccine?

b) When a rabbit protein is injected into rabbits, no antibodies against this protein are generated. If, however, the same rabbit protein is injected into guinea pigs, the guinea pigs generate antibodies against the rabbit protein. Briefly (in one or two sentences) explain this observation.

c) The genomes contained in almost all of the somatic cells in an adult human are identical. Name one (diploid) cell type that is an exception to this and specify the process by which the genetic variation occurred.

d) Will siblings have the exact same antibody repertoire? What about identical twins? Briefly explain your reasoning.

a) Below is the pedigree for a family with an autosomal recessive disease, disease X.



i) What is the genotype of individual A at the disease **X** locus? Use "+" to indicate the wildtype allele and "-" to indicate the mutant allele.

ii) What is the probability that individual B is a carrier of disease X?

iii) Individuals C and D decide to have a child. What is the probability that the child will have disease **X**?

iv) What is the probability that the child of individuals C and D will be a carrier of disease X?

b) The most common mutant allele of the disease X gene is a deletion of three nucleotides which eliminates a phenylalanine at amino acid residue 508. Although the mutant X protein is made, it is not localized to the plasma membrane.

i) Assuming the altered **X** protein is stable, where might it be found?

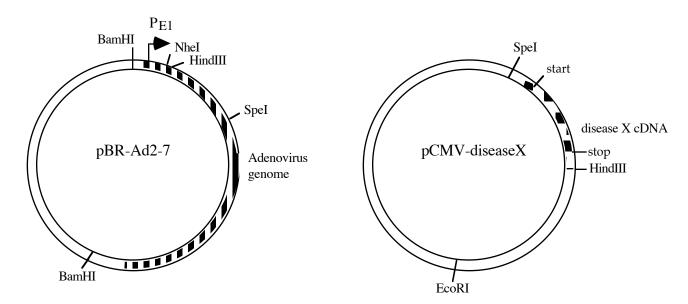
ii) Describe another mutation in this gene that could prevent the disease **X** protein from localizing to the plasma membrane.

### **Question 4, continued**

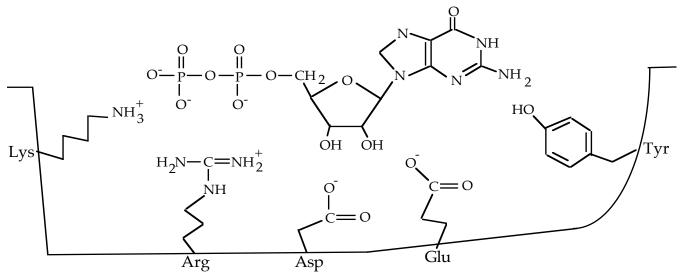
c) Researchers are currently working on gene therapy for disease **X** patients. The most promising therapy has involved incorporating the disease **X** gene into an adenovirus. Because adenovirus is a double-stranded DNA virus that targets lung epithelial cells, it can be used to deliver the disease **X** gene to the lung cells of the affected individual.

i) The adenovirus used in these studies is able to produce gp19, a protein that inhibits the display of MHC I molecules on the surface of cells. Why is this a desirable property of the virus used to deliver the disease **X** gene?

ii) Using the plasmids and restriction enzymes provided, design a procedure to create a, double-stranded DNA to incorporate into the adenovirus particle. The final product should be <u>linear</u>, contain the majority of the virus genome and have the disease **X** gene under control of the E1 promoter ( $P_{E1}$ ). *NheI* and *SpeI* create the same sticky ends. All the other restriction enzymes create unique cuts.



The figure below shows GDP in the binding pocket of a G protein.



a) Circle the strongest interaction that exists between:

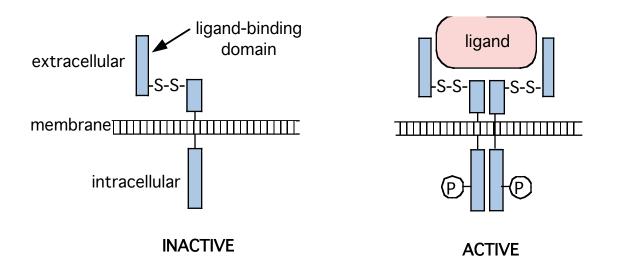
van der Waals	covalent	hydrogen bond	ionic
iii) the side chain of Tyr a	nd the guanine base	of GDP	
van der Waals	covalent	hydrogen bond	ionic
ii) the side chain of Glu a	nd the ribose group o	of GDP	
van der Waals	covalent	hydrogen bond	ionic
i) the side chain of Lys an	d the phosphate gro	up of GDP	

b) You make mutations in the GDP-binding pocket of the G protein and examine their effects on the binding of GDP. Consider the size and the nature (e.g. charge, polarity, hydrophilicity, hydrophobicity) of the amino acid side chains and and give the <u>most likely</u> reason why each mutation has the stated effect. Consider each mutation independently.

i) Arg is mutated to a Lys, resulting in a G protein that still binds GDP.

ii) Asp is mutated to a Tyr, resulting in a G protein that cannot bind GDP.

The bos/seven receptor is required for differentiation of a particular cell, called R7. It is a receptor tyrosine kinase with the structure below. As a monomer, the protein is inactive. Binding of ligand causes the receptor to dimerize, causing phosphorylation of the intracellular domain, activating the protein. During processing of the protein, the extracellular domain is cleaved and a disulfide bridge forms between two cysteines, tethering the ligand-binding domain to the rest of the protein.



a)

i) How would receptor activity be affected by changing one of the two cysteines shown above to an alanine? Explain.

ii) What effect would this mutation have on the differentiation of R7?

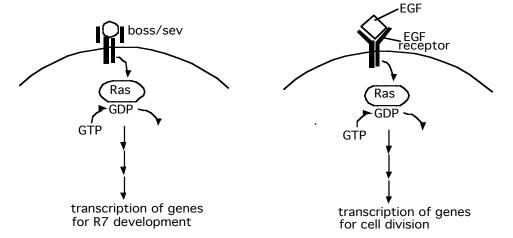
b) Name three amino acids that would be likely to be found in the transmembrane domain. What property do those amino acids have in common, and why do they cause the transmembrane domain to stay in the membrane?

d) Draw a schematic of the receptor tyrosine kinase (discussed above) prior to any cleavage or modification using the template below. Include the domains of this protein that are required for targeting to and insertion in the plasma membrane. Also label the intracellular and extracellular domains.

N \_\_\_\_\_ C

e) Activation of the above receptor causes Ras to exchange GDP for GTP, thereby activating it. This activated Ras can activate a signal transduction cascade, which ultimately results in the

transcription of genes required for R7 differentiation. In different cells in the same animal, Ras can be activated by an activated growth factor receptor. This leads to transcription of genes required for cell division.



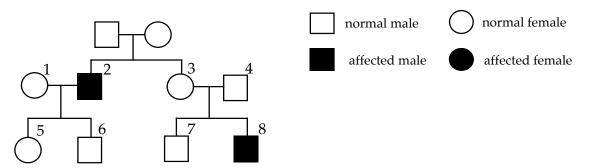
i) How is it possible for the activation of Ras to lead to transcription of different sets of genes?

ii) Given that these cells exist in the same animal, name one component in the pathway that could be mutated to give each of the following results (consider each situation independently). Describe how the mutant component differs from the wild-type component, and whether it is a loss-of-function or gain-of-function mutation.

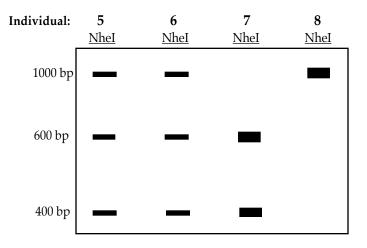
- You never see differentiation of R7 cells.
- You see uncontrolled cell proliferation.

### Question 7

You are studying a common genetic condition. The mutant allele differs from the wild-type allele by a single base-pair (bp) substitution. This substitution eliminates a *NheI* restriction site that is present in the wild-type allele. (The mutant allele is not cut by *NheI*.) A pedigree of a family exhibiting this condition is shown below:



You isolate DNA from four individuals in the pedigree. Using PCR techniques, you amplify a 1000 bp portion of their DNA that includes the site affected by the mutation. You digest the PCR products with *NheI* and analyze the resulting DNA fragments on a gel:



a) Based on these data, is this gene located on an autosome or the X-chromosome? Briefly justify your reasoning.

b) Based on these data, is the mutant phenotype dominant or recessive to wild-type and why?

c) If individuals 3 and 4 have a daughter, what is the probability that she will be affected? Justify your reasoning.

You sequence the region around the *Nhe*I site in the wild-type PCR product. You then sequence the corresponding region in the mutant PCR product and discover that not only did the mutation eliminate the *Nhe*I site in the mutant allele but it has created a new *Pvu*II restriction site. The recognition sites for the two enzymes are indicated below.

 NheI cuts at:
 5' GCTAGC 3'
 PvuII cuts at:
 5' CAGCTG 3'

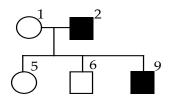
 3' CGATCG 5'
 3' GTCGAC 5'

A portion of one strand of the wild-type DNA sequence is shown below:

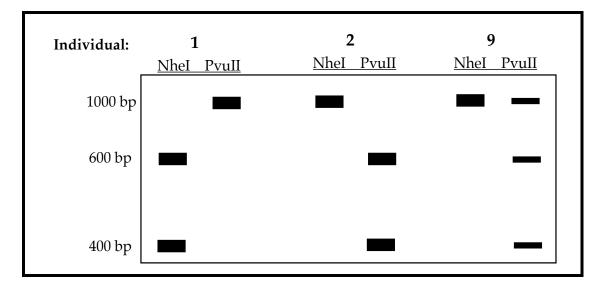
### 5'....GCTAGCTG...3'

d) What is the sequence of this same region in the mutant allele? Indicate the 5' and the 3' ends of the DNA sequence.

e) Individuals 1 and 2 have another child, 9, who is affected by the genetic condition.



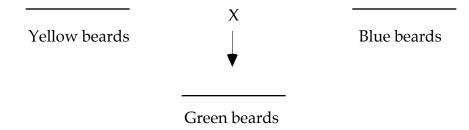
You PCR amplify the 1000 bp region affected by the mutation from individuals 1, 2, and 9, digest the PCR products with *NheI* or *PvuII*, and analyze the restriction fragments on a gel:



What event occurred and how does this explain the data shown above?

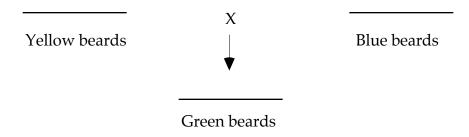
While walking through the sub-basement of the Infinite Corridor late one night you come upon an enclave of gnomes. You are struck by the color of their beards, which are all blue. (Gnomes are diploid organisms, both male and female gnomes have beards, and you can assume that the gnomes are true-breeding for this trait.) The following week you are busy pulling a hack at Harvard when you spy another enclave of gnomes. All of these gnomes have yellow beards. (Again assume that the gnomes are true-breeding for this trait.) Curious, you collect a few yellow-bearded gnomes from Harvard and bring them back to MIT. Later you discover that several of the yellow-bearded gnomes and blue-bearded gnomes have mated. The offspring of these matings are all green-bearded. Below are two possible explanations for these results.

a) Possibility 1: Beard color is controlled by a single locus. Give the genotypes in the blanks below.



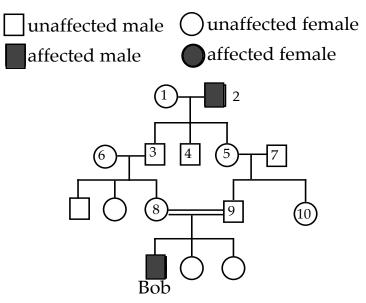
b) Possibility 2: Beard color is controlled by a pathway of two distinct enzymes encoded by the A and Q genes.

i) Give one genotype in each of the blanks below. Use A and Q to designate the wild-type alleles. Use a and q to designate the <u>loss-of-function</u> alleles.



ii) When two F1 green-bearded gnomes mate, they produce 64 green-bearded gnomes, 27 blue-bearded gnomes, and 22 yellow-bearded gnomes. Given your answer to i) above, draw the pathway for beard color. Be sure to include at which step each of the genes functions.

Bob, a sophomore at MIT, failed 8.01 his freshman year. His parents are both physicists, but he remembers that his greatgrandfather also failed physics. Bob constructs the following family pedigree and is convinced that his poor performance in physics is an inherited genetic trait.

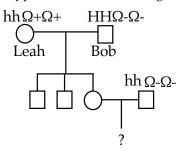


a) If Bob's hypothesis is true, what is the most likely mode of inheritance?\_\_\_\_\_

b) Individuals marrying into the family are homozygous for the wild-type allele. Complete the table below. Use G or g to denote the alleles of this gene. Be sure to note any ambiguities.

individual	genotype
2	
3	
4	
9	
10	

c) Bob meets Leah in his remedial physics class. Bob is a hard worker (homozygous for the H allele). Leah is lazy (homozygous for the h allele). The H locus is linked to a chromosomal marker, which exists in two forms  $\Omega^+$  or  $\Omega^-$ . Circle the <u>non-recombinant</u> genotypes of Bob and Leah's grandchildren.

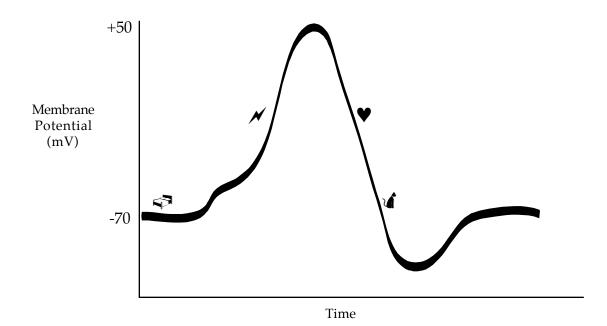


 $\begin{array}{cccc} hh\Omega^+\Omega^+ & hh\Omega^-\Omega^- & hh\Omega^+\Omega^- & HH\Omega^+\Omega^+ & HH\Omega^-\Omega^- \\ \\ HH\Omega^+\Omega^- & Hh\Omega^+\Omega^+ & Hh\Omega^-\Omega^- Hh\Omega^+\Omega^- \end{array}$ 

genotypes

Sprina 2004 Final Fxam Practice

The following is a plot of an action potential measured at a single spot along an axon. Four points are highlighted along the curve,  $\triangleleft$ ,  $\checkmark$ ,  $\checkmark$ ,  $\checkmark$ ,  $\checkmark$ ,  $\checkmark$ .



a) On the table below, identify which ion (Na<sup>+</sup>, K<sup>+</sup>, Ca<sup>++</sup>, Cl<sup>-</sup>) is undergoing the greatest net flow across the membrane at the points indicated and state the direction that the ion is moving (into the cell or out of the cell).

 Point	Ion	Direction (in/out)
×		
•		

b) The membrane potential is -70 mV at points  $\triangleleft$  and  $\checkmark$ , on the plot above. Which of the voltage-gated ion channels is closed at point  $\triangleleft$ , but open at point  $\checkmark$ ?

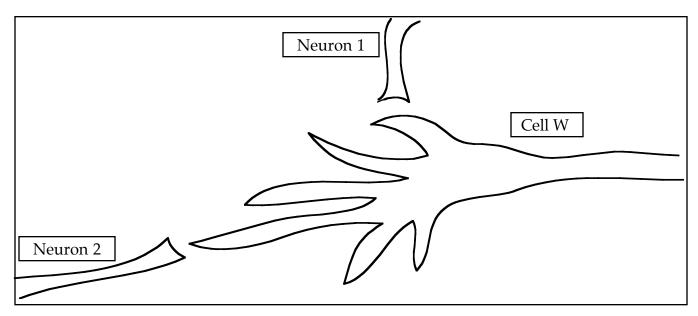
c) What dictates the closing of the voltage-gated channel that is open at point  $\varkappa$ ?

d) There are at least three states in which the voltage-dependent Na<sup>+</sup> channel exists. At ♥ on the above plot, the majority of voltage-dependent Na<sup>+</sup> channels would be in which state? Circle the best answer.

Open	Closed	Inactivated
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### Question 11 continued

Two different pre-synaptic neurons, neuron 1 and neuron 2, synapse onto cell W as shown below. When neuron 1 is stimulated, the membrane of cell W is locally depolarized. When neuron 2 is stimulated, the membrane of cell W is locally depolarized to exactly the same extent as seen with neuron 1.



e) Circle the one correct statement below.

- If stimulated equally, neuron 1 is more likely to result in an action potential in cell W than neuron 2.
- If stimulated equally, neuron 2 is more likely to result in an action potential in cell W than neuron 1.
- If stimulated equally, neuron 1 and neuron 2 are equally as likely to result in an action potential in cell W.

f) If you were exposed to a toxin that irreversibly blocked voltage-gated Ca<sup>++</sup> channels, indicate whether the following statements would be TRUE or FALSE.

Т	F	Secretory vesicles filled with neurotransmitters would stay in the nerve.
Т	F	Your muscles would end up in a rigid contraction.
Т	F	Secretory vesicles filled with neurotransmitters would fuse with the plasma membrane.

To investigate the yeast metabolic pathway for serine biosynthesis, you screen for serine auxotrophs (mutants which are unable to grow without serine supplied in their growth medium).

You isolate four such mutants, which are recessive to the wild-type strain, and you test them for growth on medium supplemented with several intermediates (A, B and C) known to be part of the pathway. The results are shown below ("+" represents growth, "-" represents no growth).

Strain	minimal medium	minimal + A	minimal + B	minimal + C	minimal + serine
wild-type	+	+	+	+	+
m1	-	+	+	-	+
m2	-	-	-	-	+
m3	-	+	-	-	+
m4	_	+	+	_	+

You then mate the haploid m1 strain with the haploid m4 strain to create a diploid yeast strain carrying both the m1 and the m4 mutations. You test the diploid for growth on the same conditons as above and observe that the diploid exhibits the same growth requirements as the m1 or the m4 haploid.

a) Are the m1 and m4 mutations in the same gene or in different genes? Briefly explain your reasoning.

b) Draw the metabolic pathway for the synthesis of serine, consistent with the data given above. Include the intermediates (A, B, and C) and serine, and indicate which mutants (m1, m2, m3, m4) are defective at each step in the pathway.

c) You create a haploid strain that has both the m1 and m3 mutations.

i) This haploid mutant will grow on media supplemented with which of the following intermediate(s): **A**, **B** and/or **C**?

ii) When grown on minimal medium this haploid will accumulate which of the following intermediate(s): A, B and/or C?

# Solutions Question 1

- AA A short, single-stranded DNA that serves as the necessary starting material for the synthesis of the new DNA strand in PCR
- DD The synthesis of DNA using DNA as a template
- S The building blocks of DNA and RNA
- HH The synthesis of protein using information encoded in mRNA
- Q The location in a eukaryotic cell where the electron transport chain occurs
- W The major component of cell membranes
- L The genetic composition of an organism
- FF A gene that lies on one of the sex chromosomes
- BB An organism without membrane-bound organelles
- M A cell with 1n chromosomes
- B The building blocks of proteins
- G A cell with 2n chromosomes
- D A major source of energy that has the general formula  $(CH_2O)_n$
- T An enzyme needed for completion of lagging strand synthesis, but not leading strand synthesis.
- GG The synthesis of RNA using one strand of DNA as a template
- U An observed characteristic of an organism
- Y A DNA molecule that is distinct from the chromosome; this molecule can be used to move foreign DNA in or out of a cell
- E The DNA from a eukaryote formed by the enzyme reverse transcriptase; this DNA lacks introns
- O An organism with 2 identical alleles for the same gene
- K A membrane protein involved in signal transduction; activation involves binding a GTP molecule
- J An organism with genetic material inside a nucleus
- N An organism with 2 different alleles for the same gene
- P A measure of the affinity of an enzyme for its substrate
- C A gene that lies on any chromosome except the sex chromosomes
- X The membrane that surrounds the cell
- A One of the alternate forms of a gene found at a given locus on a chromosome
- Z A technique for the rapid production of millions of copies of a particular region of DNA
- H Proteins with a leader peptide are directed to this cellular organelle

a) 5' AAACAGCUAUGG 3'

5'-	ATGGCCTTCACACAGGAAACAGCTATGGCCATGAGCACGC
	+
3'-	TACCGGAAGTGTGTCCTTTGTCGATACCGGTACTCGTGCG

CAGTCTCGGCATTATCCTATTAAAGGGAACTGAGGTGA -GTCAGAGCCGTAATAGGATAATTTCCCTTGACTCCACT -!

b)

c) Consider each of the following three mutations independently.

i) The mutation is before the start codon so does not change the protein sequence.

*ii)* The start codon (at nucleotides 25 - 27) would be changed and protein synthesis would now start at the next start codon (position 31 - 33). The protein would be shorter by two amino acids.

iii) This frameshift mutation will result in the protein being terminated prematurely because a new stop codon was created. As a result of this deletion, the new sequence of the peptide would be :  $H_3N^+$ -methionine-alanine-COO<sup>-</sup>

**d**) *Puromycin functions by entering the A site of the ribosome. Here, because puromycin is structurally similar to the aminoacyl-tRNA, it can participate in formation of a peptide bond with the nascent polypeptide chain. Puromycin causes peptide release from the ribosome because there is no t-RNA anticodon to link the mRNA to the peptide chain.* 

### **Question 3**

i) What are the two advantages of using a live-attenuated vaccine vs. a heat killed vaccine in this case?

It'll mimic the disease by invading cells, thus it will illicit both a humoral and cellular response.

Surface proteins will not be denatured by heat.

ii) What is a disadvantage of using a live-attenuated vaccine?

*Could acquire virulence factors, Need a "cold chain" (expensive refrigeration), it may make people sick.* 

b) The rabbit protein is recognized as foreign (non-self) by the guinea pig.

c) B cells, by gene rearrangement of Ab genes (VDJ rearrangement). Also, T cells (by rearrangement of T cell receptor genes).

d) Neither siblings nor identical twins will produce the same antibodies, because the DNA rearrangement process that produces the antibody repertoire is a random event in each B cell. **Question 4** 

ii) 2/3. *B* is not affected, therefore must be either +/+ or +/-.

	+	cf
+	+/+	+/cf
Cf	+/cf	cf/cf

iii) Since both parents are carriers (+/cf), the probability of an affected child is 1/4.

iv) Again, both parents are carriers, the probability of having a child who is a carrier is 1/2.

b) i) Since the signal sequence would be unaffected by the mutation, the protein could be found in either the Golgi, the ER, or some membrane vesicle.

**ii**) *Deletion or mutation of the signal sequence would create a protein which would not be translated into the ER.* 

c)

i) Since gp19 prevents MHC I display, virus-infected cells which are expressing the wild-type disease X gene would not be attacked by the cellular immune system.

*ii)* 1) Digest pBR-Ad2-7 with NheI and HindIII.

2) Digest pCMV-disease X with SpeI and HindIII. Isolate the CFTR cDNA fragment.

3) Ligate the products of steps 1 and 2.

4) Cut the resulting plasmid with BamHI to obtain a linear fragment with disease X gene at the  $P_{E1}$  promoter.

### Question 5

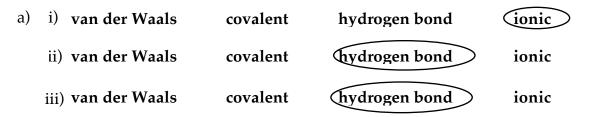
treat the substrate with DTT (a compound that disrupts disulfide bonds) and test the enzyme E activity again. This time the substrate is cleaved by enzyme E.

Why was enzyme E able to cleave the protein substrate only after the substrate was treated with DTT? *Enzyme X binds and cuts at specific sites. These site are not present on the exterior of the substrate when the substrate is properly folded. When the disulfide bonds within the substrate protein are disrupted, the 3 dimensional shape is altered, and the protein unfolds. This allows enzyme X access to sites that were previously protected within the substrate protein.* 

c) You conduct mutational studies of enzyme E. You examine the kinetics of the various mutants as compared to the normal enzyme. The data from your experiments is shown below.

ii) Which mutant has the same affinity for the substrate as the normal enzyme?  $mutant \ 2$ 

iii) Which mutant has the same catalytic activity as the normal enzyme?
 mutant 1
 Ouestion 5



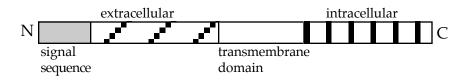
b) *i) Arg and Lys are both positively charged, thus the ionic interaction with the phosphate group is preserved. The side chains of both amino acids are also of similar size.* 

*ii) Tyr is much larger than Asp. Although Tyr can form a hydrogen bond, GDP will no longer fit into the binding pocket. The Tyr side chain is also much more hydrophobic than the Asp side chain.* 

- a) i) This would eliminate the disulfide bridge tethering the ligand-binding domain to the rest of the protein. The receptor would be inactive.
  - ii) This would prevent the differentiation of the R7 cell type.

b) *Leucine, alanine, isoleucine, valine, phenylalanine, glycine, tryptophan are all hydrophobic amino acids. The hydrophobic effect causes these amino acids to cluster away from water and stay in the interior of the plasma membrane.* 

d)



- e) i) Ras can activate several different proteins, each of which leads to a different signal transduction cascade. Different cells express different genes, and the specific protein that a given cell expresses will determine the outcome of Ras activation.
  - ii) You never see diferentiation of R7 cells. A loss-of function mutation in the boss/sev receptor would prevent signaling through Ras and activation of the differentiation pathway.
    - You see uncontrolled cell proliferation. *A gain-of function mutation in the EGF receptor such that it signals to Ras in the absence of growth factor would allow uncontrolled cell division.*

### Question 7

a) An autosome, because individual 6, a male, has 2 alleles.

**b**) *The mutant phenotype is recessive, because individuals 5 and 6 each have one copy of the mutant allele, m, and are both phenotypically normal.* 

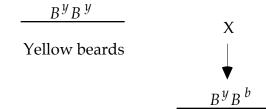
c) 1/4. Since individuals 3 and 4 already have an affected child, then they must both be heterozygotes.
 d) 5'...GCCAGCTG...3'

e) A mutation occurred which led to the production of a new mutant allele,  $m^*$ . This mutant allele has a recessive phenotype and its PCR product is cut by neither NheI nor PvuII. Individual 9 has the genotype  $m/m^*$ .

### Question 8

Below are two possible explanations for these results.

a) Possibility 1: Beard color is controlled by a single locus. Give the genotypes in the blanks below.

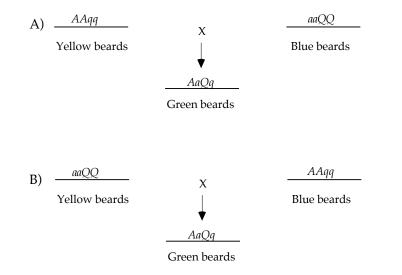


Blue beards

### Green beards

b) Possibility 2: Beard color is controlled by a pathway of two distinct enzymes encoded by the A and Q genes.

i) Give one genotype in each of the blanks below. Use A and Q to designate the wild- type alleles. Use a and q to designate the <u>loss-of-function</u> alleles.



ii) Given your answer to i) above, draw the pathway for beard color. Be sure to include at which step each of the genes functions.

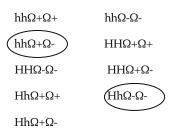
## Question 9

a) Autosomal Recessive

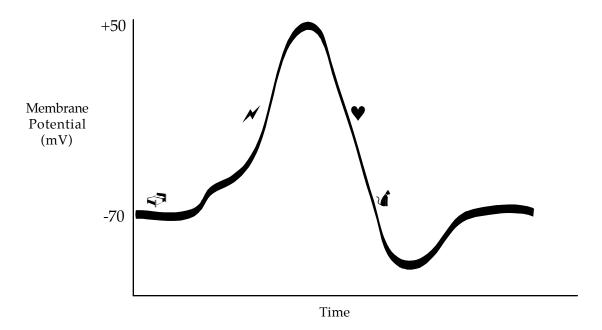
b)	individual	genotype
	2	88
	3	Gg
	4	Gg
	9	Gg
	10	GG or Gg

c)





The following is a plot of an action potential measured at a single spot along an axon. Four points are highlighted along the curve,  $\triangleleft$ ,  $\checkmark$ ,  $\checkmark$ ,  $\checkmark$ ,  $\checkmark$ ,  $\checkmark$ .



a) On the table below, identify which ion (Na<sup>+</sup>, K<sup>+</sup>, Ca<sup>++</sup>, Cl<sup>-</sup>) is undergoing the greatest net flow across the membrane at the points indicated and state the direction that the ion is moving (into the cell or out of the cell).

Point	Ion	Direction (in/out)
N	$Na^+$	in
•	$K^+$	out

b) The membrane potential is -70 mV at points  $\triangleleft$  and  $\checkmark$ , on the plot above. Which of the voltage-gated ion channels is closed at point  $\triangleleft$ , but open at point  $\checkmark$ ?

### voltage-gated K+ channel

c) What dictates the closing of the voltage-gated channel that is open at point  $\mathscr{M}$ ?

### TIME

d) There are at least three states in which the voltage-dependent Na<sup>+</sup> channel exists. At ♥ on the above plot, the majority of voltage-dependent Na<sup>+</sup> channels would be in which state? Circle the best answer.

Open

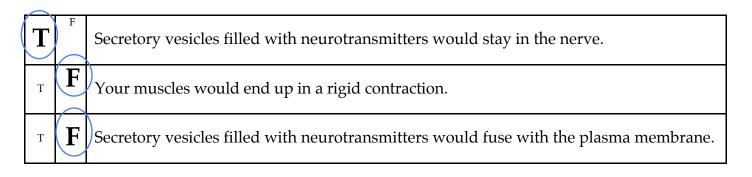
Closed

Inactivated

e) Circle the one correct statement below.

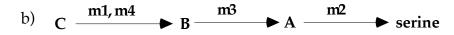
- If stimulated equally, neuron 1 is more likely to result in an action potential in cell W than neuron 2.
- If stimulated equally, neuron 2 is more likely to result in an action potential in cell W than neuron 1.
- If stimulated equally, neuron 1 and neuron 2 are equally as likely to result in an action potential in cell W.

f) If you were exposed to a toxin that irreversibly blocked voltage-gated Ca<sup>++</sup> channels, indicate whether the following statements would be TRUE or FALSE.



### Question 11

a) The m1 and m4 mutations are in the same gene. This is a complementation test. The diploid strain has the same growth phenotype as the haploid single mutants; the two mutations fail to complement (fail to produce the wild-type phenotype) in the double heterozygote. The m1 and m4 mutations must both inactivate the same gene (which codes for an enzyme essential for serine biosynthesis) so that the diploid double mutant has two mutant alleles of the same gene.

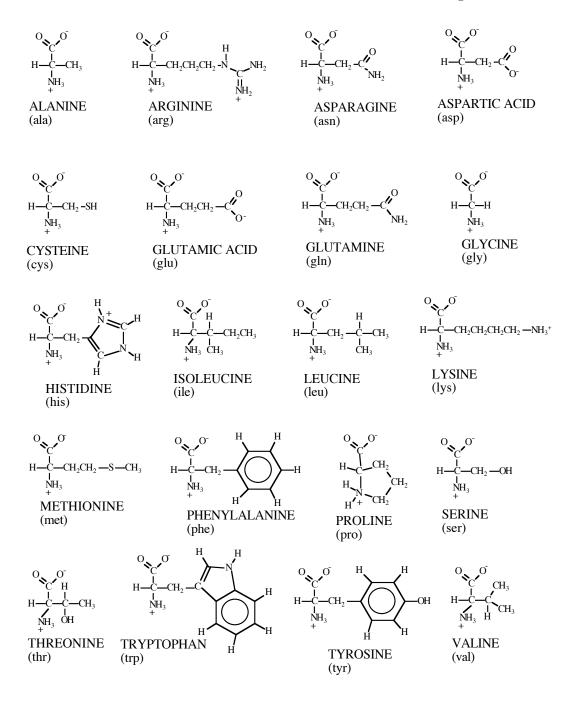


c) *i) This haploid mutant will grow on media supplemented with intermediate A.* 

ii) Intermediate C will accumulate when this haploid mutant is grown on minimal medium.

	U	C	A	G	
U	UUU phe (F)	UCU ser (S)	UAU tyr (Y)	UGU cys (C)	U
	UUC phe (F)	UCC ser (S)	UAC tyr (Y)	UGC Cys (C)	С
	UUA leu (L)	UCA ser (S)	UAA STOP	UGA STOP	А
	UUG leu (L)	UCG ser (S)	UAG STOP	UGG trp (W)	G
С	CUU leu (L)	CCU pro (P)	CAU his (H)	CGU arg (R)	U
	CUC leu (L)	CCC pro (P)	CAC his (H)	CGC arg (R)	С
	CUA leu (L)	CCA pro (P)	CAA gln (Q)	CGA arg (R)	A
	CUG leu (L)	CCG pro (P)	CAG gln (Q)	CGG arg (R)	G
А	AUU ile (I)	ACU thr (T)	AAU asn (N)	AGU ser (S)	U
	AUC ile (I)	ACC thr (T)	AAC asn (N)	AGC ser (S)	С
	AUA ile (I)	ACA thr (T)	AAA lys (K)	AGA arg (R)	A
	AUG met (M)	ACG thr (T)	AAG lys (K)	AGG arg (R)	G
G	GUU val (V)	GCU ala (A)	GAU asp (D)	GGU gly (G)	U
	GUC val (V)	GCC ala (A)	GAC asp (D)	GGC gly (G)	С
	GUA val (V)	GCA ala (A)	GAA glu (E)	GGA gly (G)	A
	GUG val (V)	GCG ala (A)	GAG glu (E)	GGG gly (G)	G
			,	, ,	

### STRUCTURES OF AMINO ACIDS at pH 7.0



# List of terms for Question 1.

### You may detach this page from the exam.

- A. allele
- B. amino acids
- C. autosomal gene
- D. carbohydrate
- E. cDNA
- F. competitive inhibitor
- G. diploid
- H. endoplasmic reticulum
- J. eukaryote
- K. G protein
- L. genotype
- M. haploid
- N. heterozygote
- O. homozygote
- P. K<sub>M</sub>
- Q. mitochondria
- R. non-competitive inhibitor
- S. nucleotides
- T. DNA ligase
- U. phenotype
- W. phospholipids
- X. plasma membrane
- Y. plasmid
- Z. polymerase chain reaction
- AA. primer
- BB. prokaryote
- CC. DNA polymerase
- DD. replication
- EE. repressor protein
- FF. sex-linked gene
- GG. transcription
- HH. translation