Biol 321  Winter 2010   Take-home Assignment aka Quiz #4

40 pts total

☞ This assignment is due on Friday Feb 26 at 8:30am. No assignments will be accepted after this deadline

☞ Please read carefully through this quiz before Monday Feb 22. I will answer questions about it in class. I can’t take questions during office hours since I want the entire class to have access to the same information (from me). You can email me questions but I will only address them during class

☞ Rules for working this quiz: This is an open-note, open-book, open-internet quiz. You may discuss these questions with your fellow classmates but, you MUST write up your answers independently. In other words, if any answers look suspiciously similar I will divide the total possible points between the similar answers. Also please do not consult other faculty members about the quiz. You can google any term or topic that you want, but this should not be necessary for any of the questions.

☞ Please word-process the text of your answers and label each part of the assignment clearly. Diagrams should be neatly laid out and labelled, but it is fine if they are hand drawn. If I can’t easily read your answers, I will not grade them.
Problem 1. (10 pts.) GENE X ENVIRONMENT INTERACTIONS

A

B

Risk or likelihood of a particular phenotype. By definition the wild-type genotype with no environmental exposure has a risk of 1.0.

Part A: In 1-2 sentences summarize the main point(s) from Panel A.

Part B: In 1-2 sentences summarize the main point(s) from Panel B

Part C: State explicitly the main difference(s) between panels A and B. 1-2 sentences max.

Part D: Review text and lecture material on phenyketonuria. Do either of these graphs accurately depict the gene X environment interaction in this disease state (consider postnatal phenotype only). Defend your answer in a maximum of three sentences. Assume variant genotype means homozygous for a loss-of-function mutation. If you make any other assumptions, state them explicitly. Your answer should include a very brief statement describing the general features of the disease state.

Part E: Read about the Xeroderma Pigmentosum in the text. Answer the question in part D for this disease state.
2.  (15 pts.)

Antiviral drug works by causing a genetic meltdown

FIRST: Read carefully through Problem 8 on Assignment set #5. Then answer these questions.

Part A  Explain in detail why this ribavirin causes “genetic meltdown”. INCLUDE THE FOLLOWING:
1. What general class of mutagens does ribavirin belong to? No explanation here -- just the proper category: __________________
2. Using correct terminology describe the type of mutations that will occur at the RNA level. One sentence.
3. Include a diagram that shows how a mutation could occur in a segment of the genome with this sequence: 5’ GAACUCA 3’ (+ strand of the genome). NOTE: there is more than one possible mutagenic event. Just show one. Be sure to label the 5’ and 3’ ends of each strand and to indicate the + and – strands.
4. In three sentences or less explain why this compound is by definition a mutagen and compare to the natural bases which also can interconvert between two different forms.

Part B
1. What modification needs to occur to ribavirin before it can be used as a substrate for RNA polymerase? One sentence or a labelled diagram. No explanation needed.
2. Why does ribavirin specifically causes mutations in RNA viruses but not in human DNA? One sentence.
3. Speculate as to why this compound doesn’t interfere with transcription in the host cells. One sentence. Any reasonable idea will suffice.

Part C. The graph in problem 8 indicates that even in the absence of a mutagen, RNA viruses “operate” near the “outer limit” of mutation tolerance. How/why does a virus tolerate a normal error rate of $2 \times 10^{-4}$ mutations per nt? Compare this error rate to that of DNA replication in a prokaryotic or eukaryotic cell. What factors are important when considering what rate of mutation is “tolerable” for a virus or an organism? 4 sentences max.
Problem 3. (10 pts.) Examine table in problem 18 on Assignment Set 5. Then answer these questions.

a. Examine the table. What is meant by a neutral sequence variation? One sentence maximum.

b. Examine the data on PM7: Is this sequence variation a polymorphism? Briefly explain (one sentence). No credit if no explanation.

c. You want to set up a genetic screen to identify individuals at high risk for early onset breast cancer. What is the significance of the data in this table in the context of this goal? (Two sentences maximum.)

d. Is the sequence variation PM2 likely to result in a neutral missense mutation or silent (same sense) mutation? Briefly explain (one sentence). No credit if no explanation.

e. Is sequence variation PM3 likely to result in a neutral missense mutation or a silent (same sense) mutation? Briefly explain (one sentence). No credit if no explanation.

4. (5 pts.) Mutations that alter renal salt absorption alter blood pressure.

Autosomal dominant mutations in the mineralcorticoid receptor (MR gene) can cause either decreased or increased blood pressure. In some family groups, frameshift or nonsense mutations in the MR gene cause hypotension (decreased blood pressure). One family has been studied in which a missense mutation causes hypertension (increased blood pressure).

a. Which phenotype results from haploinsufficiency? No explanation required.

b. Which phenotype results from a gain-of-function mutation? No explanation required.

c. The gene therapy technique currently used in clinical trials involves the “addition” to somatic cells of a single wild-type copy of a gene. In other words, a single normal copy of the gene is inserted into the genome of the mutant somatic cell, but the mutated copy of the gene is not removed or replaced. Will this strategy work for individuals who are heterozygous for a gain-of-function dominant mutation? 1-2 sentence explanation