

## F12 Study Sheet for Quiz #1

Quiz #1 is scheduled for Tuesday Oct. 23 and will be worth between 15 and 25 pts.

### *The quiz will cover:*

- All information contained in any assignment or handout related to Mendel Revisited including lecture on models, basic probability, Chi Square analysis, allele nomenclature, etc

**→ For this quiz, you will be given the chi square equation and a p value table.**

1. A couple discovers that they are both heterozygous for the same two recessive disease states: cystic fibrosis and progeria (premature aging). The mutated genes are located on different autosomes.

a. What is the probability that their first child will have one (either cystic fibrosis or progeria) but not both, of the diseases?

b. Their first kid is has cystic fibrosis, but not progeria. The parents want to know that probability that she (the kid) is free of the progeria allele -- in other words is homozygous for the wild-type allele of the progeria gene?

2. After genetic testing using DNA based techniques, a married couple (from a somewhat inbred population) discovers that they are both heterozygous for the same three recessive autosomal disease states (AaBbCc) The mutated genes are located on different autosomes.

**NOTE: Don't even think about setting up a Punnet square to work this problem. Remember that if the genes are assorting individually, you can figure out genotype/phenotype frequencies for each gene and then use the product & sum rules of probability to figure out overall probabilities for all three genes taken together. Show your work and circle your answer.**

a. What is the probability that their first kid is AABbCC?

b. They have a kid and she is phenotypically normal (shows no signs of disease). The parents want to know that probability she is AABbCc.

3. Recall that one of the goals of the Mendel Revisited Experiment was to determine if the *unc?* gene was on the same chromosome as the *dpy-11* gene. As you discovered, assessing linking by scoring progeny ratios in the F2 was dicey

business because the validity of your result rested on the correct scoring of each animal on the plate and any systematic biases in assessing phenotype led to very wonky data. *C. elegans* researchers would get around this problem of assessing linkage using the following protocol:

1. F2 hermaphrodites that are Dpy but not Unc are picked 1/plate
2. The self progeny of each F2 are scored for the presence of Unc animals and the genotype at the unc gene inferred

***If the unc and dpy genes are assorting independently, what fraction of Dpy non-Unc F2 animals will be heterozygous for the unc mutation? Very briefly explain your answer.***

**4 . ESSAY** Your mom is an avid reader of the New York Times. In the Jan 16, 2003 issue she sees the following article:

*From Worm Genes, Human Obesity Clues by Nicholas Wade*  
“Biologists at MIT have laid a new basis for studying human obesity by identifying almost all the genes that regulate fat storage and metabolism in a small animal, the laboratory roundworm.  
The finding should provide leads to the many unknown genes that regulate fat storage in people and to the defects in the genes that underlie many obesity cases.....”

Your mom is an astute individual, but she is a bit mystified with respect to the relationship between worms and humans and wonders how the statement in the second paragraph can be justified. She phones you (or maybe sends an email) and asks you to explain all of this to her.

***Briefly discuss the rationale for studying an organism like C. elegans. In your essay be sure to include the following:***

- explicit definition of a model organism
- two specific advantages of studying a model organism
- an explicit discussion of why what we learn about one organism can often be applied to understanding a different organism
- Please take time to organize your thoughts before you start writing

5. You perform the following cross with truebreeding parents:

Parents: *Lon* ♂ X *Rol Bli* ♀

Rol = roller Bli = blister Lon = long

Here is how a *C. elegans* researcher would write out the cross

*lon-1* (e31) X ♂ X *rol-3* (s754) V; *bli-15*(ct75) X ♀

Define single letter allele symbols for each gene and write out the ***genotypes of the parents*** and the ***genotypes and phenotypes of the progeny of this cross***. All mutant alleles are recessive to the wild-type allele.

Be sure to:

- The X following *lon-1* and *bli-15* indicates that both genes are X-linked
- respect conventions concerning allele designations
- ***indicate the genotype of each gene for each animal and indicate the sex***
- list both ***cross and self progeny***
- take your time and triple-check your work

*allele symbols:*

PARENTS *Lon* ♂ X *Rol Bli* ♀



↘ *self*

***F1 cross progeny:***

6. In a hunt for new mutations in *C. elegans*, you isolate a very short, fat mutant. You name this mutant strain Piggy. You do the following experiment with true-breeding strains

Piggy hermaphrodites X wild-type males

↓


F1 wild-type cross progeny males and hermaphrodites

↓ self F1 hermaphrodites



F2 30 wildtype  
2 piggy

- a) You conclude that the Piggy phenotype results from a combination of mutations in two different genes (that assort independently). You also propose that a worm homozygous for a recessive mutation in only one of these genes has a wild-type phenotype. **Why did you propose that two genes were involved in specifying this trait? Define simple allele symbols for each gene and explain your conclusion using genotypes.**
- b) Your somewhat contentious lab partner argues that your data doesn't eliminate the possibility that the Piggy phenotype is a single gene trait. In other words, he thinks that you have come up with a complicated explanation without sufficient experimental evidence. **What is your response to your lab partner's criticism? Do the appropriate chi square calculation and state explicitly in words what your calculation tells you.**
- c) **State explicitly what you can and cannot conclude from your analysis.** ***HINT: systematically work through various possible explanations for why expected and observed data don't match.***

R = red flower      r = white flower

F1 Rr plant 

↓  
self

F2 RR, Rr and rr progeny  
 and 

F2 plants are selfed to deduce genotype

An Rr  plant is selected and selfed

↓  
10 progeny plants are examined

- If only ten progeny of each red F2 plant are scored, what is the likelihood that an Rr plant will be misclassified as an RR plant? *Show your calculation.*
- What part (which data collection) of the Mendel Revisited Exercise is this question relevant to?
- Did Mendel fudge his data? Huh?* When assessing the ratio of F2 homozygotes (for the dominant allele) to heterozygotes, Mendel always reported the expected 2:1 ratio. *But if he only scored 10 progeny of each F2, the ratio of Rr to RR would turn out to be 1.7 to 1 instead of 2 to 1. Do the appropriate calculations to show where the 1.7 to 1 comes from. Show all intermediate steps*