

- **Put your name and your GSI's name on all 9 pages. The pages will be separated for grading.**
- **No papers, notes, books, calculators, computers, telephones, mp3 players or other electronic devices may be used.**
- **You can leave your answers as fractions and sums.**
- **Write all answers in the space provided. You can use the backs of the pages for your own notes.**
- **Be sure to answer all parts of all 15 questions.**
- **The numbers in parentheses are the points for each part. The total is 150.**

1. Suppose that in flies, you find an autosomal mutant pd that causes polka-dot wings; pd^+ is the wild-type alleles. This mutant is partially penetrant in heterozygotes: 25% have polka-dot wings. It is completely penetrant in mutant homozygotes. Assume you cross a pure-breeding line of polka-dot flies with a pure breeding line of flies with wild-type wings.

a. (4) What is the fraction of flies in F_2 population with polka-dot wings?

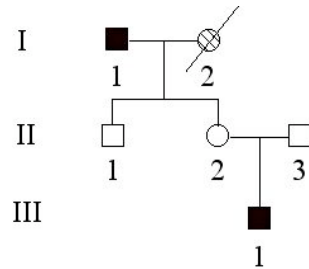
Answer: $3/8$

b. (4) What is the fraction of flies with polka-dot wings in the backcross of the F_1 to the polka-dot population?

Answer: $5/8$

2. (8) Humans and chimpanzees had a common ancestor 6 million years ago (6×10^6). Suppose there are 100 4-fold redundant sites in the sequence of α -globin. If the rate of neutral substitution is 2×10^{-9} per site per year, what is the expected number of differences between humans and chimpanzees would you expect to see at the 4-fold redundant sites of α -globin?

Answer: $D = 2\mu TL = 2(2 \times 10^{-9}) \times (6 \times 10^6) \times 10^2 = 2.4$.



3. In the above pedigree for mice, black indicates that the individual has blue fur, white indicates that the individual has the normal agouti fur, and the diagonal lines indicate that the fur color is unknown because of an unfortunate accident in the laboratory. Assume blue fur color is determined by an allele of a single gene.

a. (3) Based on the information in the pedigree, did I-2 have agouti or blue fur?

Answer: Agouti

b. (3) Does your answer to part a depend on whether the blue gene is X-linked?

Answer: No

c. (3) Suppose you could mate III-1 with a female from a pure-breeding line of agouti mice. Describe how you would determine whether the blue gene is X-linked.

Answer: Interbreed the hybrid offspring. If there are any blue females, then the blue gene is autosomal. If there are only blue males, then it is X-linked.

4. (8) Suppose that a gene is a suppressor of the expression of the wrinkled pea allele that Mendel studied; remember that round is dominant to wrinkled. The suppressor has two alleles S and s. ss peas are round regardless of the genotype of the round/wrinkled gene. The round/wrinkled phenotype is not affected in SS and Ss peas. You begin with two pure-breeding lines, one homozygous for s and round and the other homozygous for S and wrinkled. You then create the F₁ population by hybridizing the pure lines and the F₂ population by self-fertilizing individuals in the F₁. What are the expected proportions of round and wrinkled peas in the F₂ if these two genes are unlinked?

5. Suppose you have two guinea pigs, a male with smooth black fur and a female with rough white fur. You mate them and get 10 male and 10 female offspring (the F_1), all having rough black fur. You allow them to randomly mate. In the 125 offspring, you find 8 with smooth white fur, 25 with smooth black fur, 23 with rough white fur and 69 with rough black fur.

a. (5) How are fur color and texture inherited?

Answer: Two genes. Black is dominant to white; rough is dominant to smooth.

b. (5) What fractions of the four fur types would you expect to see if you mate a smooth white female from the F_2 with a male from the F_1 ?

Answer: Equal numbers of all four types.

6. (8) Suppose that the frequency of the S allele of the β -globin gene in a malarial region among adults is 0.2. If malaria were eliminated and, after that, 1/2 as many SS individuals as AA and AS individuals survive to adulthood, what would be the frequency of S in the next generation of adults if there is random mating in this population?

Answer: $(pq+q^2/2)/(p^2+2pq+q^2/2)=0.18/0.98$. $p=0.8$, $q=0.2$.

7. (10). Eosine-colored eyes in *Drosophila melanogaster* are caused by a recessive X-linked gene. In a pure-breeding eosine line, Bridges found a fly with cream-colored eyes. From this fly he obtained a pure-breeding line with cream-colored eyes. Bridges then crossed males from the pure-breeding population with cream-colored eyes with females from a pure-breeding wild-type line. All the F_1 had wild-type eyes. When the F_1 was interbred, he obtained 104 wild-type females, 52 wild-type males, 44 males with eosine-colored eyes and 14 males with cream-colored eyes. Assume these are in an 8:4:3:1 ratio.

Propose a genetic model to explain these results and assign phenotypes to each of the genotypes.

Answer: Let the eosine gene be eo^+/eo . The data fit a model of an autosomal gene. The eosine mutant is epistatic to the autosomal gene. Let the autosomal gene be a^+/a . eo^+/eo^+ and eo^+/eo (females) are wild-type regardless of the genotype at the autosomal gene.

8. The most common cause of cystic fibrosis is in people of European ancestry is caused by a recessive mutant allele, $\Delta F508$, which has a 2% frequency in Europe. Assume that the genotype frequencies of this gene are in Hardy-Weinberg proportions and that there are no other alleles that cause cystic fibrosis.

a. (3) If neither parent in a family has cystic fibrosis, what is the probability that the first child born in that family has cystic fibrosis?

Answer: $0.02^2=0.0004=1/2500$.

Also accepted: $(1/4)* [(2pq)/(1-q^2)]^2$; partial credit for $(1/4)* (2pq)^2$

b. (3) If the first child born to this couple has cystic fibrosis, what is the probability that the second child will have it also?

Answer: 1/4.

9. Suppose you are studying the inheritance of four phenotypes in *Drosophila melanogaster*. The mutant phenotypes are warped (wings), rumped (bristles), pale (wings), and raven (eyes). You begin with a warped, rumped pure-breeding line and a pale, raven pure-breeding line. You create two F₁ populations from these pure-breeding lines. In cross I, you breed warped, rumped females with pale, raven males and obtain warped rumped males and wild-type females. In cross II, you breed pale, raven females with warped rumped males and obtain pale, raven males and wild-type females. You then do a testcross of F₁ females from cross I with a pure-breeding warped, rumped, pale, raven population and obtain the following phenotypes among 1000 male offspring.

3 pale
428 pale, raven
48 pale, raven, rumped
23 pale rumped
22 warped, raven
2 warped raven, rumped
427 warped rumped
47 warped

a. (2) Are these 4 genes X-linked or autosomal?

Answer: X-linked.

b. (4) If each mutant phenotype is caused by a single gene, which two genes show no evidence of recombination between them?

Answer: There are no recombinants between the warped and pale genes.

c. (4) Describe what you can infer about the order of these four genes.

Answer: raven is between rumped and warped/pale. There is no way to determine whether warped or pale is closer to raven.

d. (4) What is the recombination fraction (RF) between genes carrying the raven and rumped mutants?

Answer: $RF = (48 + 2 + 3 + 47) / 1000 = 100 / 1000 = 0.1$

e. (4) What is your answer to part d if the data are for 1000 females instead of 1000 males?

Answer: RF is the same, 0.1.

10. The major allele causing cystic fibrosis in Europeans is $\Delta F508$, a 3-base deletion that results in the loss of the 508th amino acid of the CFTR protein.

a. (5) What is would the equilibrium frequency of $\Delta F508$ be if it were recessive in its effect on survivorship and individuals homozygous for $\Delta F508$ did not survive to adulthood? Assume that the population is at equilibrium under a mutation-selection balance with mutation rate 10^{-6} per generation and ignore reproductive compensation.

Answer: $\sqrt{10^{-6}}=10^{-3}=0.001=1/1000$.

b. (5) What would the relative fitness of normal homozygotes have to be for the equilibrium frequency to be 0.04 under balancing selection alone, again assuming that individuals homozygous for $\Delta F508$ do not survive to adulthood and ignoring reproductive compensation

Answer: $p=t/(s+t)=0.96$, when the relative fitnesses are written $1-s$, 1 , $1-t$. With $t=1$, $s=1/0.96-1$.

11. You want to test whether the genes for smooth/wrinkled peas and yellow/green peas are still unlinked. You create an F_1 population by hybridizing a pure-breeding that has yellow, smooth peas with a pure-breeding population that has green, wrinkled peas. All peas in the F_1 are yellow and smooth. You perform the backcross with the green, wrinkled population and find the following numbers of peas of each type in the offspring. The total is 160.

yellow, smooth	42
yellow, wrinkled	31
green, smooth	38
green, wrinkled	49

a. (6) What is the value of χ^2 you compute from these data?

Two answers are possible, but the correct df has to go with its χ^2 value. Otherwise, they lose the 2 for the df.

$$\text{Either: } \chi^2 = \frac{11^2 + 11^2}{80} = \frac{242}{80} \text{ with 1 df}$$

$$\text{Or } \frac{4 + 81 + 4 + 81}{40} = \frac{170}{40} \text{ with 3 df}$$

b. (2) How many degrees of freedom (df) would you use in finding the p value in a chi square?

12. You sample a population of *Drosophila melanogaster* and collect 200 individuals, 100 from location 1 and 100 from location 2, which is 500 miles away. In each group, you count the number of flies with scabrous eyes, a trait you already know is caused by a recessive autosomal allele. In location 1, you find 9 individuals with scabrous eyes and in location 2 you find only 1 individual with scabrous eyes.

a. (3) What is the frequency of the scabrous allele in these two populations if the genotypes are in Hardy-Weinberg equilibrium in each population?

Answer: location 1 - 0.3. location 2 - 0.1

b. (3) What fraction of the flies will have scabrous eyes if the two samples are mixed together?

Answer: $10/200=0.05$

c. (3) What is the expected fraction of flies with scabrous eyes in the mixed sample if the scabrous allele were in Hardy-Weinberg equilibrium?

Answer: $(0.3+0.1)^2/4=0.04$

13. You have several broccoli plants for which you know the genotypes of a self-incompatibility gene of the type discussed in class. Below are the genotypes of each plant.

Plant 1 = S_1S_3

Plant 2 = S_2S_3

Plant 3 = S_1S_4

a. (8) For each of the crosses below, list the genotypes of the progeny and the fractions of each genotype expected. In each cross, the first plant listed is the pollen donor and the second plant is the potential female parent.

Cross A: Plant 1 x Plant 2 \rightarrow $\frac{1}{2} S_1S_2$ $\frac{1}{2} S_1S_3$

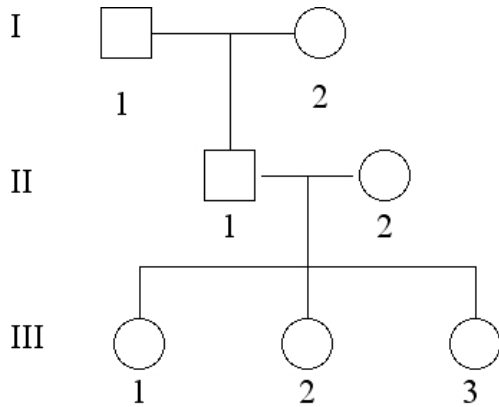
Cross B: Plant 1 x Plant 3 \rightarrow $\frac{1}{2} S_1S_3$ $\frac{1}{2} S_3S_4$

Cross C = Plant 2 x Plant 3 \rightarrow $\frac{1}{4} S_1S_2$ $\frac{1}{4} S_3S_4$, $\frac{1}{4} S_1S_3$ $\frac{1}{4} S_2S_4$

Cross D = Plant 1 x Plant 1 \rightarrow no progeny

b. (2) If you have a randomly mating population of broccoli plants in which there are 10 alleles of this self-incompatibility gene in equal frequency, what is the fraction of the offspring that will be homozygous?

Answer: 0. There can be no homozygotes



14. Assume that individuals in the above pedigree have been genotyped for two autosomal microsatellite loci, A and B. The genotypes are as follows. The subscripts indicate the different alleles of each locus. You can assume there is no genotyping error.

I-1 $A_1A_1B_1B_2$; I-2 $A_1A_2B_3B_4$

II-1 $A_1A_2B_2B_3$; II-2 $A_1A_3B_2B_3$

III-1 $A_1A_2B_2B_3$; III-2 $A_2A_3B_3B_3$; III-3 $A_1A_1B_2B_2$

a. (3) What is the haplotype phase of II-1?

Answer: A_1B_2/A_2B_3 .

b. (4) Which individual or individuals in generation III can you be sure inherited non-recombinant gametes from II-1?

Answer: III-2 and III-3. III-2 had to have inherited A_2B_3 from II-1 and III-3 had to have inherited A_1B_2 from II-1.

c. (2) For which individual or individuals in generation III can you **not** determine whether they inherited recombinant or non-recombinant gametes from individual II-1?

Answer: III-1. She could have inherited A_1B_2 (non-recombinant) from II-1 if the phase of II-2 is A_1B_3/A_2B_2 or A_1B_3 (recombinant) if the phase of II-2 is A_1B_2/A_2B_3 .

15. Suppose you catch a carp that has no scales and you want to determine whether this phenotype is inherited and, if so, how. You breed the scaleless carp to a carp from a pure-breeding population with a normal scale pattern. Among their progeny (the F_1 population) 1/2 have the wild-type scale pattern and 1/2 have a linear pattern you have not seen before.

a. (2) You mate two linear carp from the F_1 and find a 6:3:2:1 ratio of linear : wild-type : scaleless : scattered (another pattern you have not seen before). How many genes appear to be involved in determining scale pattern in this species?

Answer: 2

b. (2) You mate two wild-type carp from the F_1 and find a 3:1 ratio of wild-type to scattered. How many genes are appear to cause the difference between carp with the wild-type and scattered patterns?

Answer: 1

c. (4) You hybridize carp with the scattered pattern obtained from the cross described in part b with carp from a pure-breeding wild-type population. What fractions of carp with the different scale patterns would you expect to see in the offspring?

Answer: All wild-type.

d. (5) When you interbreed the scaleless carp you obtained from the cross in part a, you find a 2:1 ratio of scaleless to scattered carp. When the carp with the scattered pattern from the cross in part a are interbred, all their offspring have the scattered pattern. How can you explain these results?

Answer: Fish with the scattered are homozygous, say BB. Scaleless fish are heterozygous, say Bb. bb fish die.

e. (6) Describe the genetic basis of scale pattern and list the phenotype associated with each genotype.

Answer: There are 2 genes, say A/a and B/b.

Wild-type: AABB, AaBB

Scaleless: aaBb

Linear: AABb, AaBb

scattered: aaBB

dead: AAbb, Aabb, aabb