7.03 Exam 1 Review – 10/02/06

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1.) You are studying a completely penetrant human disorder that is rare in the general population but relatively common in certain regions. Woman #1, whose mother's brother was afflicted with this disorder, is trying to determine the probability that she and her husband would have an affected child. Woman #1's husband's sister ultimately died of the disease. Woman #1's father is from a low-risk population, but everyone else in the pedigree is from a region where the disorder is common. Besides the two individuals mentioned, no one else in this pedigree, including the parents of the affected individuals, is/was affected with the disease. All cases of this disorder are suspected to be caused by mutations in the same gene.

(a) Draw the pedigree of this family, indicating the genotypes of individuals where possible.

(b) Is this disorder recessive or dominant? Is it X-linked or autosomal?

(c) If the couple's first child is affected, what is the probability that the couple's second child will be affected?

(d) What is the probability that the couple's first child will be affected?

(e) If the couple's first child is not affected, what is the probability that Woman #1 is a carrier of the disease?

2.) You have isolated three mutants, *his1*, *his2* and *his3* that are unable to grow without the amino acid histidine. You mate each of these with a wild type strain of the opposite mating type and analyze the resulting diploid for its ability to grow without histidine. You get the following results:

Mating	Diploid's phenotype
his1 x wildtype	WT
his2 x wildtype	WT
his3 x wildtype	His^-

a) What does this tell you about each of the mutations?

b) You mate *his1* with a *his2* strain of the opposite mating type. The resulting diploid is unable to grow on histidine. What does that tell you about these two mutations?

c) You decide to mate your *his1* and *his3* mutants and then sporolate the resulting diploids. You analyze the resulting tetrads for their ability to grow without histidine:

X= No Growth ○ = Growth

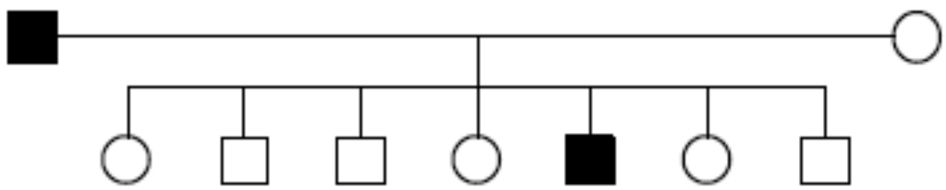
1	2	3	4	5	6	7	8	9	10	11	12	13	14	15	16	17	18
X	X	X	X	X	X	O	X	X	X	X	O	X	O	X	X	X	X
X	X	X	O	X	X	X	X	X	O	X	X	X	X	X	X	X	X
X	X	X	X	X	X	X	X	X	X	X	X	X	O	X	O	X	X
X	X	X	X	X	X	X	X	X	X	X	X	O	X	X	X	X	X

d) How many PD, TT and NPD are there?

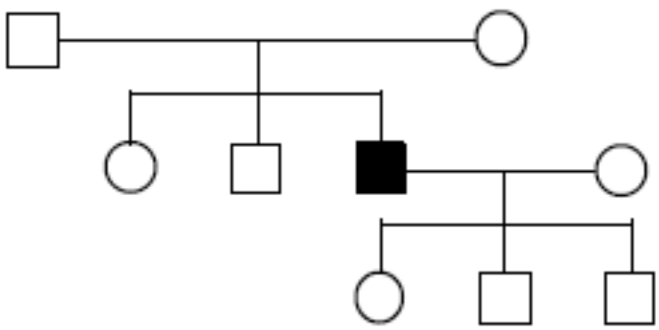
e) Are these two genes linked? If so then by how far?

3.) For each of the following pedigrees, list all modes of inheritance (the four options are: autosomal recessive, autosomal dominant, X-linked recessive, and X-linked dominant) that are consistent with the trait indicated by shaded circles and squares in the pedigree. Assume 100% penetrance and no spontaneous mutations. Also, assume that the trait is very rare in the general population.

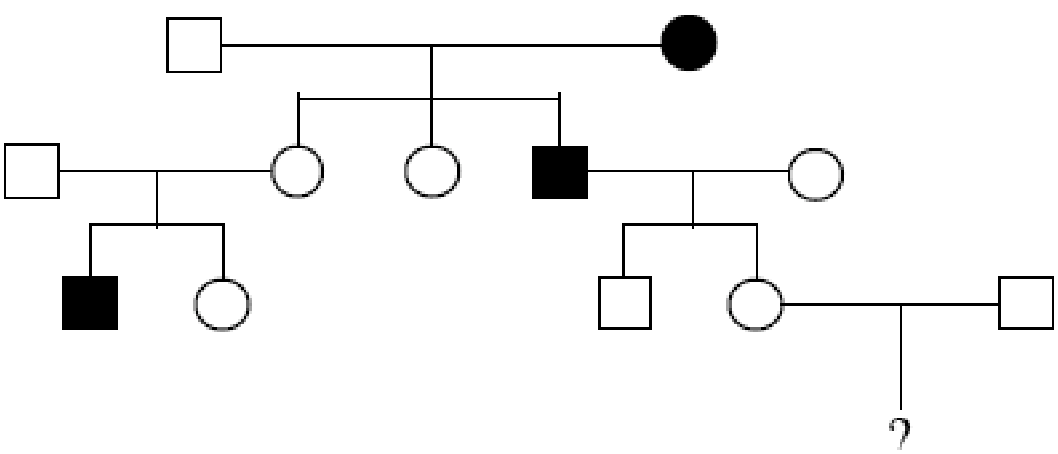
(i)



(ii)



(iii)



b) For each mode of inheritance that is consistent with the pedigree from part (iii): if the couple in question decides to have the child labeled with a question mark, what is the probability of that child showing the trait indicated by shaded circles and squares?

c) For each mode of inheritance that is consistent with the pedigree from part (iii): if identical twin daughters who do not show the trait are born to this couple, what is the probability that both children are carriers?

d) For each mode of inheritance that is consistent with the pedigree from part (iii): if fraternal twin daughters who do not show the trait are born to this couple, what is the probability that both children are carriers?

4.) Wild-type Drosophila have red eyes, and white eyes is an X-linked recessive phenotype caused by a single mutation. A new single mutation that gives the recessive phenotype of apricot colored eyes is isolated. A female from a true-breeding apricot-eyed strain is crossed to a male from a true-breeding white-eyed strain. All of the resulting F1 flies have apricot eyes.

(a) Are the white-eye and apricot-eye mutations in the same gene or in different genes? Explain your answer.

A collection of apricot-eyed F1 females from the cross described above are mated to males from a true-breeding white-eyed strain, and 1000 male progeny are examined. Among these progeny, only 6 flies have normal red eyes.

(b) What is the measured distance between the white-eye and apricot-eye loci in cM?

A new mutation is isolated that causes the recessive eye color "peach." A female from a true-breeding peach-eyed strain is crossed to a male from a true-breeding white-eyed strain. All of the resulting F1 females have normal red eyes and all of the resulting F1 males have peach eyes.

(c) Is the peach-eye mutation on an autosome or on the X-chromosome? Explain your answer.

(d) Are the white-eye and peach-eye mutations in the same gene or in different genes? Explain your answer.

A mutation that causes the recessive phenotype of crossveinless wings lies on the X-chromosome. A female from a true-breeding strain with apricot eyes and crossveinless wings is crossed to a male from a single mutant true-breeding strain with white eyes and normal wings. As expected, all of the F1 females from this cross have apricot eyes and normal wings. A large collection of these F1 females are crossed to wild-type males and 10,000 **male** progeny are examined. The observed phenotypes are as follows:

Normal wings	White eyes	4,418
Crossveinless wings	Apricot eyes	4,330
Normal wings	Apricot eyes	610
Crossveinless wings	White eyes	590
Normal wings	Red eyes	2
Crossveinless wings	Red eyes	50

(e) Draw a genetic map showing the relative order of the crossveinless, apricot and white loci.