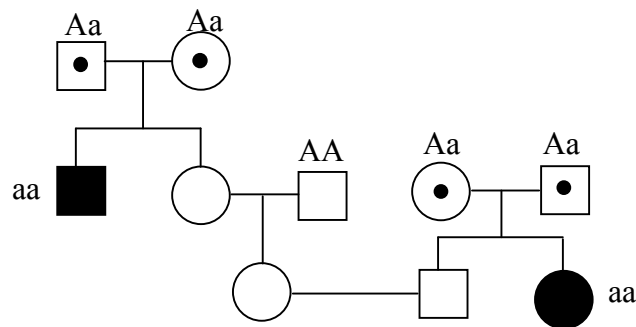


7.03 Exam 1 Review

SOLUTIONS

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?

Autosomal recessive

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

1/4

The only way to get an affected child is if both of the parents are carriers. The probability of getting an affected child from two carriers (Aa x Aa) is 1/4

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

1/18

2/3 - probability that Woman 1's mother is a carrier

1/2 - probability that Woman 1 is a carrier if her mom is a carrier

2/3 - probability that the husband is a carrier

1/4 - the probability they will have an affected child if they're carriers

$2/3 \times 1/2 \times 2/3 \times 1/4 = 1/18$

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

5/17

x = mom is a carrier y = one unaffected child

$p(x) = 2/3 \times 1/2 = 1/3$

$p(\text{not } x) = 2/3 \times 1/2 + 1/3 = 2/3$

For $p(y|x)$ we need to take into account the probability the father is a carrier or homozygous for the unaffected allele

$p(y|x) = 1/2 \times 2/3 \times 1/2 + 1/2 \times 2/3 \times 1/2 + 1/2 \times 2/3 \times 1/2 = 5/6$

$p(y|\text{not } x) = 1$

$p(x|y) = (5/6 \times 1/3) / (5/6 \times 1/3 + 1 \times 2/3)$

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
<i>his1</i> x wildtype	WT
<i>his2</i> x wildtype	WT
<i>his3</i> x wildtype	His ⁻

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

Dominance test: ***his1* – recessive**
***his2* – recessive**
***his3* – dominant**

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?

Complementation test: You get the mutant phenotype out so they don't complement. Therefore they should be in the same gene.

c) You decide to mate your *his1* and *his3* mutants and then sporulate 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	1	1	1	1	14	1	1	1	1
									0	1	2	3		5	6	7	8
X	X	X	X	X	X	○	X	X	X	X	○	X	○	X	X	X	X
X	X	X	○	X	X	X	X	X	○	X	X	X	X	X	X	X	X
X	X	X	X	X	X	X	X	X	X	X	X	X	○	X	○	X	X
X	X	X	X	X	X	X	X	X	X	X	X	○	X	X	X	X	X
P	P	P	T	P	P	NP	P	P	TT	P	TT	TT	NP	P	TT	P	P
D	D	D	T	D	D	D	D	D		D			D	D		D	D

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

PD = 11
TT = 6
NPD = 1

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

Yes (PD ≠ NPD). $(6+6 \times 1) / (2 \times 18) \times 100 = 33\text{cm}$

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.

same gene—they fail complementation since they don't give wt red eyes

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?

Recombination in the mother is the only way to get red-eyed flies. The recombination would have to take place between the apricot and white mutations. This crossover will give one wt chromosome and one double mutant chromosome. Since we have 6 red eyed (wt) flies we can assume that we have 6 double mutants, adding to 12 recombinant offspring in total.

Distance (cM) = $\frac{\# \text{ of Recombinants}}{\text{Total offspring}} \times 100 = \frac{12}{1000} \times 100 = 1.2 \text{ cM}$

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.

X-chromosome—the females and males exhibit different phenotypes

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

Different genes— they complement each other in the females

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.

