

Bio 102 Practice Problems

Chromosomes, Karyotyping and Sex Linkage

Multiple choice: Unless otherwise directed, circle the one best answer:

1. A sex-linked trait is a trait:
 - A. That can be inherited only by males
 - B. That can be inherited only by females
 - C. Whose gene is located on the X-chromosome
 - D. Whose gene is located on the Y-chromosome
 - E. Which cannot be accurately identified using a pedigree
2. A sex-linked gene:
 - A. is located on the X or Y chromosome
 - B. is not inherited according to Mendel's principles
 - C. is not passed from father to son
 - D. is required for development as a male or female
 - E. shows a phenotype only in males
3. All of the following are similarities between the behavior of genes and the behavior of chromosomes EXCEPT:
 - A. Alleles segregate in gamete formation, and homologous chromosomes separate in meiosis I
 - B. Dominant alleles determine the individual's phenotype, and sister chromatids determine the individual's karyotype.
 - C. An individual has two alleles of each gene and two chromosomes in each homologous pair
 - D. Alleles of different genes segregate independently, and different homologous pairs separate independently in meiosis.
 - E. Offspring receive one allele of each gene from each parent, and offspring receive one chromosome from each homologous pair in the sperm and egg.

Short answer (show your work or thinking to get partial credit):

1. Colorblindness is sex-linked and recessive in humans. A woman with normal vision but whose father was color blind marries a man with normal vision whose father was also color blind. What type of vision can be expected in their offspring?

Let X^C represent an X chromosome carrying the dominant normal allele; X^c represents an X chromosome carrying the recessive colorblind allele. Y represents the Y chromosome, which lacks this gene (and all sex-linked genes that we will discuss).

The man with normal vision is X^CY ; his father doesn't matter (because he inherited Y from his father)

The woman's father was X^cY , so the woman got the colorblind allele from him and is X^CX^c .

Now, the cross is $X^CX^c \times X^CY$. So:

for daughters, $\frac{1}{2} X^CX^C$, normal; $\frac{1}{2} X^CX^c$, normal but carriers

for sons, $\frac{1}{2} X^CY$, normal; $\frac{1}{2} X^cY$, colorblind

2. A man and a woman, both of normal vision, have the following offspring:

(1) A color-blind son who then has a daughter with normal vision

(2) A daughter with normal vision who later has one color-blind son and one normal son

(3) A daughter with normal vision who later has five sons, all normal

What are the probable genotypes of the original couple, their children, and their grandchildren?

We know that there are color-blind alleles in this family, and we know they didn't come from the father, who has normal vision and must be X^CY , so they came from the mother, who must be heterozygous, X^CX^c .

In #1, the color-blind son can only be X^cY , and his daughter gets X^c from him so must be X^CX^c .

In #2, the normal daughter has a color-blind son, so she must be X^CX^c . The normal son is X^CY and the color-blind son is X^cY .

In #3, the daughter with normal vision has five normal sons, so while there's an outside possibility she's heterozygous (and very lucky), she's most likely X^CX^C . All her sons are X^CY .

3. A man's maternal grandmother had normal vision and his maternal grandfather was color-blind. His mother is color-blind, and his father has normal vision.

- a. What are the genotypes of his parents and these two grandparents?

His color-blind mother can only be homozygous X^cX^c ; his father can only be X^CY .

His mother's father is color-blind, so obviously he's X^cY .

His mother's mother had normal vision, but in order to produce a color-blind daughter, she must have contributed an X^c allele, so she must be X^CX^c .

- b. Will this man be color-blind or normal?

The man gets his X chromosome from his mother, and she has only X^c to give, so he's X^cY and colorblind.

- c. If he marries a woman who has the same genotype as his sisters, what will the phenotypes of their children be?

The man's sisters get X^c from the mother and X^C from the father, so they're heterozygous.

If he marries a woman of this genotype, the cross is $X^cY \times X^CX^c$. Daughters will get X^c from him but could get either allele from their mother, so $\frac{1}{2}$ of the daughters will be colorblind and $\frac{1}{2}$ will have normal vision but be carriers. Sons can get either allele from the mother, so $\frac{1}{2}$ will be colorblind and $\frac{1}{2}$ normal.

4. Suppose a baby is born with 47 chromosomes, instead of the normal 46: 22 pairs of autosomes, plus two X chromosomes and one Y chromosome. List two different ways that nondisjunction could have resulted in this particular chromosome combination.

- (1) Nondisjunction in meiosis I in the mother: XX gamete produced, fertilized by Y from father
- (2) Nondisjunction in meiosis II in the mother: XX gamete produced, fertilized by Y from father
- (3) Nondisjunction in meiosis I in the father: XY gamete produced, fertilized by X from mother
(Can't be meiosis II in the father, because X and Y have already separated at this point, so only an XX or YY gamete could be produced by nondisjunction, resulting in an XXX or XYY offspring after fertilization.)

5. Colorblindness is a recessive sex-linked trait in humans. A colorblind man marries a woman with normal vision whose father is colorblind. What fraction of this couple's sons would you expect to be colorblind? What fraction of the daughters? Will you expect any of the sons or daughters to be carriers?

Let X^c represent the recessive colorblind allele; X^+ represents normal. The woman's father is colorblind, meaning that his only X is X^c , so she inherited the trait but doesn't show it; she must be heterozygous, X^cX^+ . The man is colorblind, so he's automatically X^cY .

Daughters will all get X^c from their father and either X^c or X^+ from their mother; half of them should be normal but carriers and the other half colorblind.

Sons will all get Y from their father and either X^c or X^+ from their mother; half of them should be normal and the other half colorblind. Sons can't be carriers.

6. A yellow-bodied male fruit fly from a pure-breeding line is crossed with a normal female (also pure-breeding). What genotypes and phenotypes will you expect in the F_1 and F_2 generations if the recessive yellow-body phenotype is due to an autosomal gene? What will you expect if the gene is sex-linked?

If yellow is autosomal and recessive, then:

Let y = yellow and $+$ = normal.

Pure-breeding yellow male is yy ; pure-breeding normal female is $++$.

All F_1 offspring will be $+y$ (heterozygous) and phenotypically normal (yellow is recessive).

In the F_2 , $\frac{1}{4}$ will be $++$ and $\frac{1}{2}$ $+y$, so $\frac{3}{4}$ are phenotypically normal. Males and females come out the same.

If yellow is sex-linked and recessive, then:

Let X^y = yellow and X^+ = normal; as usual, Y represents the Y chromosome.

Yellow male can only be X^yY ; pure-breeding normal female must be X^+X^+ .

F_1 males will be X^+Y and phenotypically normal; F_1 females will be X^+X^y and phenotypically normal but carriers. (Notice that for any sex-linked trait, you have to specify genotypes for males and females separately, even if their phenotypes are the same.)

The F_2 cross is then $X^+Y \times X^+X^y$, so for male offspring, $\frac{1}{2}$ will be X^+Y (phenotypically normal) and $\frac{1}{2}$ will be X^yY (yellow); for female offspring, all will be phenotypically normal but $\frac{1}{2}$ will be X^+X^y carriers and $\frac{1}{2}$ will be homozygous, X^+X^+ .

7. A female fruit fly that has red eyes and short wings is crossed with a male with brown eyes and long wings. All the F_1 flies have red eyes and long wings. A cross between F_1 males and females produces the following offspring:

75 long, red females
23 short, red females
39 long, red males
37 long, brown males
14 short, red males
10 short, brown males

Are the eye-color and wing-shape genes autosomal or sex-linked? Which alleles are dominant? What are the genotypes of the parents?

From the F_1 , we can see that red eyes and long wings are the dominant alleles.

If we add up long and short, we see a 3:1 ratio for both males and females (75:23 for females, 76:24 for males). This suggests that the eye color gene is autosomal. Furthermore, the 3:1 ratio tells us that the F_1 flies were all heterozygous, so the parents must have been pure-breeding.

For red and brown, however, we see only red females and a 1:1 ratio of red to brown for males (53:47). The difference between the sexes suggests sex linkage. Half the males inherited the red allele from their mothers, and half inherited brown, so the mother was heterozygous. The females all got the red allele from their father.

Now, if we let L =long, l =short, X^R =red and X^r =brown, the F_1 flies must have been $X^R X^r Ll$ and $X^R Y Ll$.

Since the original parents were a red short female and a brown long male, and we have shown that they were pure-breeding, their genotypes must have been $X^R X^R ll$ and $X^r Y LL$.

8. A pregnant woman and her husband are concerned about the possibility their unborn child could have a genetic disorder. They decide to have amniocentesis and karyotyping performed to look for chromosomal abnormalities. It turns out that the child has 47 chromosomes: 23 normal autosomal pairs plus one X chromosome and two Y chromosomes.

- a. Will this child be a male or a female? Explain.

This is a male: Y determines "maleness," so any individual with a Y is male.

- b. This chromosomal abnormality resulted from a nondisjunction event during meiosis. Did the nondisjunction occur in the father or in the mother (assuming both have normal karyotypes)? Explain your reasoning briefly.

Clearly, this had to happen in the father! Mom had only X chromosomes to give, so no matter what might have gone wrong there, she could never have given a Y. Both Y chromosomes must have come from the father.

- c. Did the nondisjunction event occur during meiosis I or meiosis II? Explain your reasoning.

In meiosis I, the X and Y chromosome pair up as if they were a normal homologous pair. If nondisjunction occurred there, you would get an XY gamete or a gamete with no sex chromosome at all, which is not what happened.

Instead, the nondisjunction must have happened in meiosis II, where the two sister chromatids of the Y chromosome could fail to separate, giving a YY gamete.

9. A man with hemophilia (failure of blood to clot due to a recessive, sex-linked allele) has a daughter whose phenotype is normal. She marries a man whose phenotype is also normal.

a. What is the probability that a son will have hemophilia? Explain.

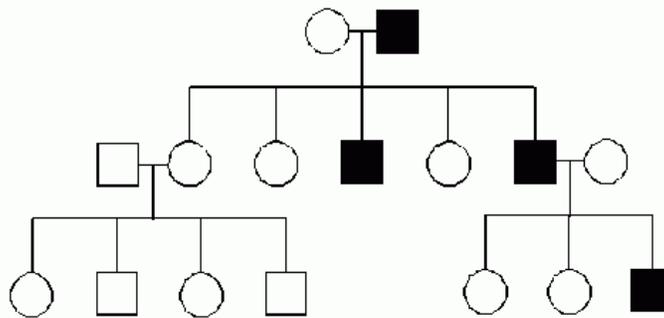
The woman's father had hemophilia, so if we use X^h for this recessive trait, her father was X^hY . She inherited her father's X (as all daughters do), but she doesn't show the trait, so she must have gotten a normal allele (X^+) from her mother and her genotype is X^+X^h (heterozygous). Her husband is normal and since he's hemizygous he must be X^+Y . Therefore, this couple's sons will get Y from dad and either X^+ or X^h from mom, meaning that there is a 50/50 chance of getting X^h and having hemophilia (X^hY).

b. What is the probability that a daughter will have hemophilia? Explain.

None. The daughters will get X^+ from their father, so even if they get X^h from their mother, they won't have hemophilia. However, they have a 50/50 chance of being carriers

10. For the trait shown in each of the following human pedigrees, state whether the most likely mode of inheritance is dominant or recessive and whether the trait is sex-linked or autosomal. Assume the traits are rare. Base your decision only on the information given, and briefly state your evidence.

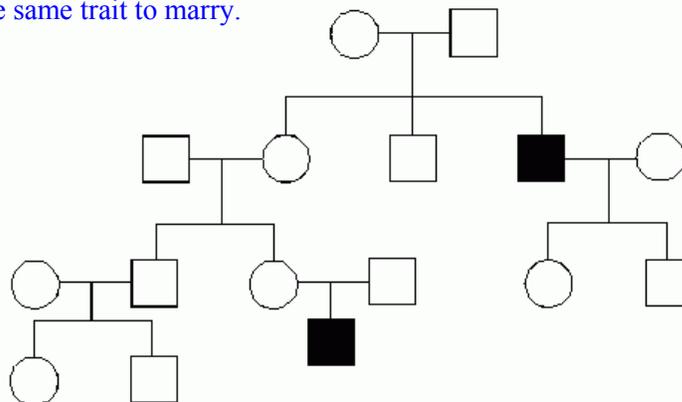
a.



This trait is autosomal and dominant. Evidence: it's dominant because every affected child has an affected parent. Given that it's dominant, then it has to be autosomal because the sons could not inherit a sex-linked trait from their fathers! Don't be fooled by the fact that it happened to occur only in males!! It can't be sex-linked and dominant because of the male-male inheritance.

It could be sex-linked and recessive, if we assume that both of the mothers of the affected children are carriers of the same trait. However, we are instructed to assume that the trait is rare, so it's pretty unlikely that two different men who have a rare trait will happen to find two different women who are carriers of the same trait to marry.

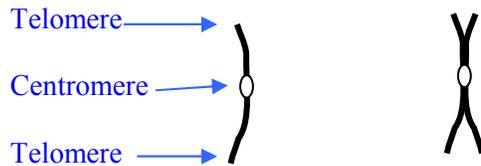
b.



This trait is sex-linked and recessive. Evidence: it's recessive, because the affected individuals each have two unaffected parents. It could be autosomal, but then we'd again have to assume that two different carriers of a rare trait both married other carriers of the same rare trait. Given that it's rare, it is more likely that this is a sex-linked trait, so that the sons get it directly from their mothers and only one affected family is involved.

11. Chromosome structure.

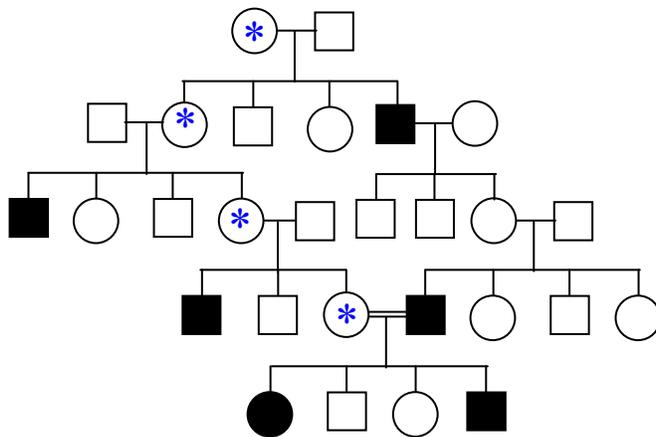
- a. Draw a chromosome with one chromatid and also draw a chromosome with two chromatids. Label the centromere and telomeres on the chromosome with one chromatid.



- b. In all the karyotypes we looked at in class, the chromosomes had two chromatids. Why?

The cells from which the chromosomes were isolated for karyotyping had been grown on a dish and arrested in metaphase (by adding the drug colchicine). When cells are in metaphase, it is after S phase (when the DNA duplicates and chromosomes change from having one chromatid to two) and it's before anaphase (when the two sister chromatids separate, regenerating chromosomes with one chromatid).

12. The pedigree below is for a rare human genetic disease. Decide whether the most likely mode of inheritance is dominant or recessive, and whether the trait is autosomal or sex-linked. The double line indicates a marriage between two related individuals. **Give specific evidence** to support your conclusions.



Dominant or Recessive?

Recessive. Affected children of unaffected parents is clear evidence.

Autosomal or Sex-linked?

Sex-linked.

If it's autosomal, then all the people marked with the * must be carriers. this is very unlikely if it's rare.

If it's sex-linked, then the males who show the trait got one copy from their mothers, and no outsiders have to be carriers. The only female who has the disease got it from a carrier mother and an affected father, who are related to each other.

13. A child with Klinefelter syndrome (XXY) is born to a normal couple who have no prior history of the disease.

- a. Could this child's chromosomal abnormality be due to nondisjunction during meiosis in the father? Explain your answer briefly, and if you say yes, also illustrate your answer with a diagram to show how this would occur.

Yes: the X and Y could have failed to separate in meiosis I, producing an XY sperm that could fertilize an X-bearing egg from the mother.

- b. Could this child's chromosomal abnormality be due to nondisjunction during meiosis in the mother? Explain your answer briefly, and if you say yes, also illustrate your answer with a diagram to show how this would occur.

Yes: the two X chromosomes could have failed to separate in either meiosis I or meiosis II, producing an XX egg that could be fertilized by a Y-bearing sperm from the father.

14. You have been given a vial containing fruit flies that have beautiful emerald-green eyes (a very rare mutation, indeed!) instead of the usual red eyes. You know that these flies are pure-breeding, but you don't have any other information about this trait. You would like to know whether the green-eye allele is dominant or recessive and sex-linked or autosomal. Diagram a cross (or two crosses, if necessary) that you could do to gain this information and show how the results will be different if green is sex-linked vs. autosomal and if it is dominant vs. recessive. Assume that the dominant allele will be completely dominant and don't forget to define your symbols.

Cross a green female with a pure-breeding wild-type male:

If recessive and autosomal:

R = red, r = green

rr female x RR male

all offspring red (Rr)

If recessive and sex-linked:

X^R = red, X^r = green

X^rX^r female x X^RY male

all male offspring X^rY , green

all female offspring X^RX^r , red

If dominant and autosomal:

G = green, g = red

GG female x gg male

all offspring green (Gg)

If dominant and sex-linked:

X^G = green, X^g = red

X^GX^G female x X^gY male

all male offspring X^GY , green

all female offspring X^GX^g , green

If all offspring are green, then we will have to do one more cross, such as continuing to the F_2 :

If dominant and autosomal:

Gg x Gg

Offspring $\frac{3}{4}$ G-, green; $\frac{1}{4}$ gg, red

If dominant and sex-linked:

X^GX^g x X^GY

male offspring $\frac{1}{2}$ X^GY , green; $\frac{1}{2}$ X^gY , red

Female offspring all X^G- , green

15. In jackalopes, fur color is determined by a pair of alleles showing incomplete dominance; heterozygous individuals with one brown and one white allele have gray fur. The fur color gene is autosomal. A sex-linked gene determines antler length.

A brown female jackalope with short antlers is crossed with a white male that has long antlers. All of the offspring, both males and females, have short antlers. Give the genotypes, phenotypes and expected ratios that will result from a cross between two of these offspring. Don't forget to define your symbols carefully.

Let's deal with antler length first. This is a sex-linked gene, so a male can only have one allele. Therefore, the male with long antlers has only a long allele, and all of the female offspring will inherit it. Because all the female offspring get a long allele yet have short antlers, we know that short must be dominant over long.

Now we can define symbols for antler length: X^S = short (dominant), and X^s = long (recessive). Note that we must have the X to show that this is a sex-linked trait!

OK, how about fur color? Here, we have lots of given information, so the symbols are easy. Remember that incomplete dominance is usually shown with capital superscripts, so that we can use one letter to represent one gene. Let's use C for the color gene, and our two alleles are then C^B = brown and C^W = white (the heterozygous C^BC^W genotype produces gray).

Now we have a brown female jackalope with short antlers. The only way to be brown is to have the genotype C^BC^B . However, she could be X^SX^s or X^SX^S and have short antlers (dominant phenotype). Looking at her offspring, however, we see that they all have short antlers. Since we know the females get a long allele from their father, they must all get a short allele from the mother, so she must be homozygous: X^SX^S .

The white male with long antlers can only be $C^WC^W X^sY$.

All of the female offspring will get X^S from the mother and X^s from the father, so they're X^SX^s and have short antlers. They all get C^B from the mother and C^W from the father, so they're C^BC^W and have gray fur.

All of the male offspring will get X^S from the mother and Y from the father, so they're X^SY and have short antlers. They all get C^B from the mother and C^W from the father, so they're C^BC^W and have gray fur.

16. Two different genes are important in development of *Drosophila* (fruit flies). A mutant allele of the *Bar* gene (B) produces very narrow (bar-like) eyes; this allele shows incomplete dominance, and heterozygous flies have kidney-shaped eyes. A mutant allele of the *scalloped* gene (s) produces wings with wavy edges; this allele is completely recessive to wild-type. Both genes are sex-linked.

- a. Using standard *Drosophilagenetic* notation and considering all the facts above, what would be the best symbols to use for the wild-type and mutant alleles of each gene?

Symbols must include the X chromosome, since these are sex-linked traits, and a lower-case letter should be used for a recessive trait. For fruit flies, it's also standard to use a (+) for wild-type (whether dominant or recessive). So:

For eyes, X^B (Bar) and X^{B^+} (wild-type) would be good, clear symbols

For wings, X^s (scalloped) and X^{s^+} (wild-type) would be clear

- b. A female fly with bar eyes and normal wings mates with a male that has normal eyes and scalloped wings. Both flies are from pure-breeding lines. Give the genotype and phenotype of their male and female offspring, again using symbols that give the most possible information (be sure your symbols take into account *all* the facts!).

These genes are both located on the same chromosome, so your genotypes need to clearly show this. One way to show this would be: $X^{B^+ s^+} X^{B^+ s^+} \times X^{B^+ s} Y$, giving female offspring that are all $X^{B^+ s^+} X^{B^+ s^+}$ (all kidney-eyed and normal-winged) and male offspring that are all $X^{B^+ s^+} Y$ (all bar-eyed and normal-winged). Or, you could show the linkage more explicitly using the horizontal line to separate the two chromosomes:

$$\frac{B^+ s^+}{B^+ s^+} \times \frac{B^+ s}{Y} \rightarrow \text{all males } \frac{B^+ s^+}{Y}, \text{ all females } \frac{B^+ s^+}{B^+ s^+}$$

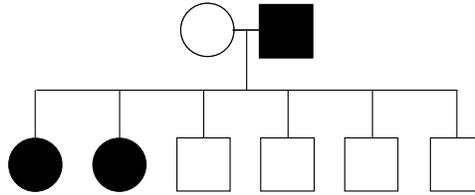
- c. The male and female flies whose genotypes you provided above now mate to produce an F_2 generation. Assuming that no crossing-over happens between these two genes, give the genotypes, phenotypes and ratios expected for their offspring.

If there is no crossing-over, then there are only two gametes for each parent:

	$B^+ s^+$	Y	
$B^+ s$	$\frac{B^+ s}{B^+ s^+}$	$\frac{B^+ s}{Y}$	males: ½ scalloped ½ bar
$B^+ s^+$	$\frac{B^+ s^+}{B^+ s^+}$	$\frac{B^+ s^+}{Y}$	females: ½ kidney ½ bar

17. An exceedingly rare genetic condition known as congenital generalized hypertrichosis leads to excessive facial and body hair.

- a. Suppose you are a genetic counsellor whose client, a woman, has this condition. In the course of your interview, you learn that her sister and her father have similar symptoms, but neither her mother nor any of her four brothers has the disease. Draw a pedigree for this family.



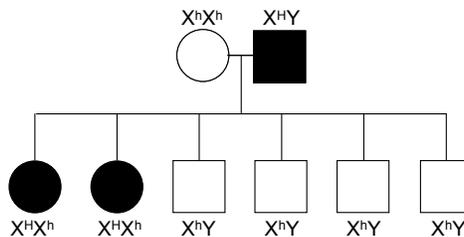
- b. Based on the available information, decide whether this disease is dominant or recessive and whether it is sex-linked or autosomal. Give specific evidence to support your conclusions.

First, it's very odd that the affected father passed the trait to his only two daughters but not to any of four sons. While it's possible that this could be a coincidence, it seems very unlikely and much more likely that the trait is located on his X chromosome and therefore goes to all his daughters but none of his sons. Sex-linked is therefore the best answer.

Now, if the father has the trait on his X chromosome and it's very rare, then it's very unlikely that the mother is also a carrier. This means the daughters are getting the trait when they receive one hypertrichosis allele on the X chromosome from the father. Thus, the trait must be dominant.

- c. Assign symbols to the alleles and assign genotypes to each of the individuals in your pedigree.

As usual, symbols should use one letter for one gene and should show dominance; also, we need to show that this is a gene on the X chromosome. If we want to use "H" for hypertrichosis (you can use any letter you want), then the symbols can be X^H for the dominant hypertrichosis allele and X^h for the recessive normal allele (or you could also use X^+ for the "wild-type" normal allele). Genotypes are:



- d. If your client marries a man with no family history of the disease, what is the probability that she will have an affected child?

If there's no family history of the dominant disease, then the man must be X^hY , and she is heterozygous, X^HX^h . That means there is a 50% chance that either a son or a daughter will inherit the dominant disease allele from her and will have the trait.

18. You have pure-breeding mutant fruit flies that have narrow eyes and short wings. You decide to make a cross between mutant females and wild-type males (pure-breeding, round eyes, long wings) and another cross between mutant males and wild-type females. Here are the results:

mutant ♀ × wild-type ♂
 males: narrow eyes, long wings
 females: oval eyes, long wings

mutant ♂ × wild-type ♀
 males: round eyes, long wings
 females: oval eyes, long wings

a. Explain how the wing-length character is inherited: dominant or recessive, sex-linked or autosomal.
 Short wings appear to be recessive (F₁ flies have long wings) and autosomal (no difference between the sexes or between these reciprocal crosses).

b. Explain how the eye-shape character is inherited.
 Here, we see the appearance of a third phenotype, intermediate between the other two (oval eyes), and we see a difference between sexes in the reciprocal crosses. So, this looks like a sex-linked trait showing incomplete dominance.

c. Diagram a cross between the narrow-eyed male offspring and the oval-eyed female offspring. Include genotypes and phenotypes of parents and offspring, as well as the expected ratios of the offspring. Assume independent assortment if your model involves more than one gene.

Symbols: X^N = narrow eyes (sex-linked, incomplete dominance)
 X^R = round eyes
 L = long wings (autosomal, dominant)
 l = short wings (recessive)

The narrow-eyed F₁ males must be X^NY Ll, and the oval-eyed females are X^NX^R Ll.

X^NY Ll (narrow, long) × X^NX^R Ll (oval, long)

male offspring:

½ X^NY (narrow)
 ½ X^RY (round) ¼ ll (short)

¾ L- (long)



3/8 narrow, long
 3/8 round, long
 1/8 narrow, short
 1/8 round, short

female offspring:

½ X^NX^N (narrow)
 ½ X^NX^R (oval) ¼ ll (short)

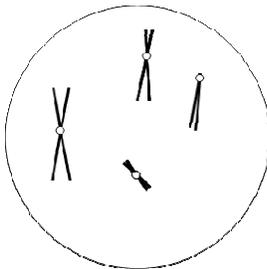
¾ L- (long)



3/8 narrow, long
 3/8 oval, long
 1/8 narrow, short
 1/8 oval, short

19. Fruit flies (*Drosophila melanogaster*) have one pair of sex chromosomes and three pairs of autosomes.

a. Below, show how a cell from the ovary of a female fly might look during prophase of meiosis II.



Four chromosomes, no homologous pairs (already separated), but sister chromatids are still attached.

b. Suppose nondisjunction affects the sex chromosomes of the cell you just drew. The resulting egg cells are then fertilized by sperm cells that carry Y chromosomes. What sex chromosomes will the offspring have, and will they survive?

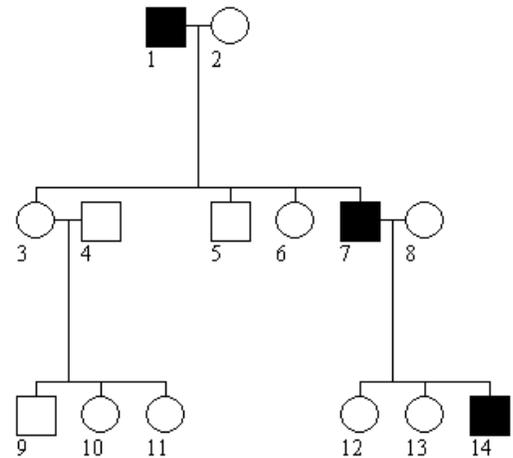
If the two sister X chromosomes fail to separate, the resulting gametes will get either XX or no sex chromosome. If an XX gamete is fertilized by a Y-bearing sperm, an XXY individual will result and survive. If a gamete with no sex chromosome is fertilized, a YO individual will result and die.

20. Wild-type *Drosophila* have round, red eyes and long, straight wings; the wild-type flies used by geneticists can be assumed to be homozygous for all genes. Suppose you find a male fly that has very narrow slit-shaped eyes and curly wings. In order to find out more about how these traits are inherited, you mate your fly with a wild-type female and then cross the F₁ offspring to get an F₂ generation. Here are the results for the F₂ generation:

<u>males</u>	<u>females</u>
300 round eyes, curly wings	300 round eyes, curly wings
100 round eyes, straight wings	100 round eyes, straight wings
300 slit eyes, curly wings	300 oval eyes, curly wings
<u>100 slit eyes, straight wings</u>	<u>100 oval eyes, straight wings</u>
800 total	800 total

- a. Is the curly wing trait dominant or recessive? Give specific evidence to support your answer.
 The curly allele is dominant. ¾ of all the offspring are curly, and ¼ are straight, the 3:1 ratio expected in the F₂ generation; this shows that both homozygous and heterozygous flies that inherit the curly allele get curly wings.
- b. Is the wing-shape gene an autosomal gene or a sex-linked gene? Give specific evidence to support your answer.
 The wing shape gene is autosomal. We see the same 3:1 ratio for both males and females.
- c. Is the slit eye trait dominant or recessive? Give specific evidence to support your answer.
 The slit eye allele is incompletely dominant. Half the females are heterozygous, and in those flies, we see the appearance of a third, intermediate phenotype.
- d. Is the eye-shape gene an autosomal gene or a sex-linked gene? Give specific evidence to support your answer.
 The eye shape gene is sex-linked. The fact that there's a difference between females and males is a big clue; it then turns out that only females get the heterozygous phenotype.

21. Suppose you are a researcher studying pancreatic cancer, which has a very high fatality rate and is usually incurable. While cancer is not inherited, there are specific alleles of certain genes that are associated with increased *risk* of cancer. In a particular family, you find that an unusually large number of individuals have gotten pancreatic cancer, and you produce the pedigree shown at right. Based on the pedigree, is increased risk of pancreatic cancer a dominant trait or a recessive trait, and is it autosomal or sex-linked? Give specific evidence to support your conclusions. (Assume that a cancer-susceptibility allele will be relatively rare in the population.)



This looks like a dominant trait. It appears in every generation, and since it is rare we can assume that the outsiders who marry into the family are not carrying the allele.

If it is dominant, then it is clear that individual #1 is heterozygous for the allele and #2 does not have it (or she'd show it). Therefore, #7 could only have gotten the allele from #1, and since sex-linked genes (on the X chromosome) can't go from father to son, it must be autosomal.

True or False? Read carefully: a question is false unless it is completely true!

- T** **F** 1. Nondisjunction is an error in meiosis which can affect only three different human chromosomes, plus the sex chromosomes.
- T** **F** 2. Sex-linked recessive traits can be seen only in men, because they result from genes located on the X chromosome.
- T** **F** 3. In preparing cells for karyotyping, colchicine is added to stimulate cell division stop cell division at metaphase, since this is the only time chromosomes become visible.
- T** **F** 4. Nondisjunction of all chromosome pairs in meiosis could result in polyploidy, but polyploid individuals cannot survive.
- T** **F** 5. The Y chromosome is responsible for male development in mammals, so even aneuploid mammals with a Y chromosome (XXY, XXXY, XXXXY, etc.) would be male.
- T** **F** 6. A karyotype can be used to detect genetic disorders such as Down syndrome, sickle-cell anemia or Tay-Sachs disease in an unborn child.
- T** **F** 7. Often, more males than females in a pedigree show a sex-linked trait because sons who inherit only a single recessive allele from their fathers will show the recessive phenotype.
- T** **F** 8. Amniocentesis and karyotyping permit an unborn child to be screened for genetic disorders such as Tay-Sachs disease and sickle-cell anemia.
- T** **F** 9. Polyploidy is invariably fatal.
- T** **F** 10. Nondisjunction for all chromosome pairs in meiosis I could result in polyploidy; however, polyploid organisms never survive.
- T** **F** 11. Klinefelter syndrome could result from nondisjunction occurring during meiosis II in a male.
- T** **F** 12. Genetic disorders such as Down syndrome, Turner's syndrome or Tay-Sachs disease could be detected in unborn children by amniocentesis and karyotyping.

Matching:

1. Each of the individuals below made a significant contribution to our understanding of the molecular nature of the gene. Match each one with his or her major contribution. One letter per blank, use the letters once only.

- | | |
|-----------------------------|--|
| <u>H</u> Martha Chase | a. Used x-ray analysis to investigate the structure of DNA |
| <u>F</u> Matthew Meselson | b. Showed how codons could represent amino acids |
| <u>A</u> Rosalind Franklin | c. Found that genes affect individual steps in metabolic pathways |
| <u>G</u> Frederick Griffith | d. Demonstrated that genes are located on chromosomes |
| <u>C</u> George Beadle | e. Showed that the transforming principle was DNA |
| <u>D</u> T. H. Morgan | f. Provided experimental evidence of semiconservative replication |
| | g. Observed an inheritable change in <i>Streptococcus pneumoniae</i> |
| | h. Found that DNA was essential for virus replication |
| | j. Showed that enzymes are missing in inborn errors of metabolism |