

# Bio 111 Handout for Genetics 1

This handout contains:

- Today's iClicker Question
- Handout for today's lecture

## Bio 111 iClicker Question #1 (Genetics 1)

In Mendel's experiments with round and wrinkled peas that I described in this lecture, 3/4 of the  $F_2$  offspring produced in cross 2 were round. Suppose you randomly picked two of those round  $F_2$ 's and crossed them. What are the possible resulting offspring of this cross?

- A. 3:1 round:wrinkled      or      all round      or      all wrinkled
- B. 3:1 round:wrinkled      only
- C. all round only
- D. 3:1 round:wrinkled      or      all round
- E. 3:1 round:wrinkled      or      all wrinkled

1. Figure out your answer and select the appropriate letter (A-E).
2. Turn on your iClicker by pressing the "ON/OFF" button; the blue "POWER" light should come on. If the red "LOW BATTERY" light comes on, you should replace your batteries soon.
3. When I say "start", transmit your answer as follows:
  - a. Press the button corresponding to the answer you've selected (A thru E).
  - b. The "STATUS" light will flash green to indicate that your answer has been received. If the "STATUS" light flashed red, your answer was not received; you should re-send it until you get a green "STATUS" light.

The answer will be available on the course website (<http://intro.bio.umb.edu/111-112>) later today.

# Bio 111 Genetics Definitions:

**Character** = a heritable feature of an organism (ex. eye color, pea shape, etc.)

**Trait** = one of many forms of a character (ex. blue eyes, brown eyes; round peas, wrinkled peas, etc.)

**Gene** = a particle of inheritance. "Seed shape in peas is controlled by one gene." In the simplest cases, each gene controls one **character** (ex. a gene for eye color or pea shape), and each **character** is controlled by one gene.

**Allele** = an alternative form of a gene. The different alleles of a **gene** control different traits of that **character**.

(ex. "The seed shape gene in peas has two alleles, each conferring a different trait:  
R - round and r - wrinkled")

**wild-type allele** = the allele most commonly found in nature

**mutant allele** = an altered form of a gene that is different from wild-type

**Genotype** = the alleles present in an organism (ex. "RR", "Rr", "rr")

**homozygous** = both alleles are the same type (ex. "RR rr") a.k.a "true-breeding"

**homozygote** = an organism that is homozygous

**heterozygous** = both alleles are different (ex. "Rr")

**heterozygote** = an organism that is heterozygous

**haploid** = having only one allele of each gene; sometimes abbreviated "N". Gametes (eggs, sperm, etc.) are haploid and would therefore have genotypes like "r" or "R" but not "RR".

**diploid** = having two alleles of each gene; sometimes abbreviated "2N". Most cells of an individual are diploid and would therefore have genotypes like "RR", "Rr", etc.

**Phenotype** = the observable characteristics of an organism (ex. "round peas" or "wrinkled peas")

**dominant** = the phenotype observed in the heterozygote

types of dominance

(ex. A = red and a = white, so the homozygotes are AA - red and aa - white)

- **simple dominance** = the heterozygote looks like one of the homozygotes (ex. if A is simply dominant to a, then Aa would be red)

- **incomplete dominance** = the heterozygote's phenotype is in between the homozygotes (ex. Aa would be pink - in between red and white).

- **co-dominance** = the heterozygote looks like both homozygotes (ex. Aa would have patches of red and patches of white). The inheritance of blood type involves co-dominance.

**recessive** = the phenotype masked in the heterozygote

# Bio 111 Handout for Genetics 2

This handout contains:

- Today's iClicker Questions
- Handout for today's lecture

## iClicker Question #2A - at the start of lecture

Consider Campbell figure 14.5. Suppose you crossed an  $F_1$  purple-flowered plant with a white-flowered plant from the P generation. What would be the expected offspring?

- A. all purple
- B. all white
- C. 3:1 white:purple
- D. 3:1 purple:white
- E. 1:1 white:purple

As you enter the class, beam in your answer as described below. Answers will be accepted until I have finished with the daily announcements.

## iClicker Question #2B - at the end of lecture

Fred is blood-type B, his mom is type AB, and his dad is type A. From this information, what can you conclude?

- A. I don't know
- B. the individuals listed above cannot be Fred's parents
- C. Fred is  $I^B i$  and his dad can only be  $I^A i$
- D. Fred is  $I^B i$  and his dad can be  $I^A i$  or  $I^A I^A$
- E. Fred is  $I^B I^B$

When I say "start", beam in your answer as described below; you will have two minutes to enter your answer.

### **Beaming in your answers**

1. Figure out your answer and select the appropriate letter (A-E).
2. Turn on your iClicker by pressing the "ON/OFF" button; the blue "POWER" light should come on. If the red "LOW BATTERY" light comes on, you should replace your batteries soon.
3. Transmit your answer as follows:
  - a. Press the button corresponding to the answer you've selected (A thru E).
  - b. The "STATUS" light will flash green to indicate that your answer has been received. If the "STATUS" light flashed red, your answer was not received; you should re-send it until you get a green "STATUS" light.

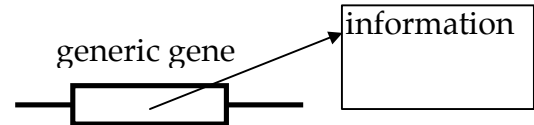
# Bio 111 Gene & Allele Revisited

Take the case of round & wrinkled peas.

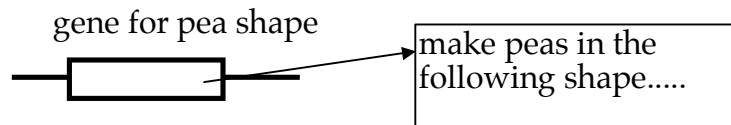
- The **gene** is responsible for the **character** - that is, the pea's shape.
- The **allele** is responsible for the **trait** of the character  
- that is, round ("R" allele) or wrinkled ("r" allele).

Another way to look at this:

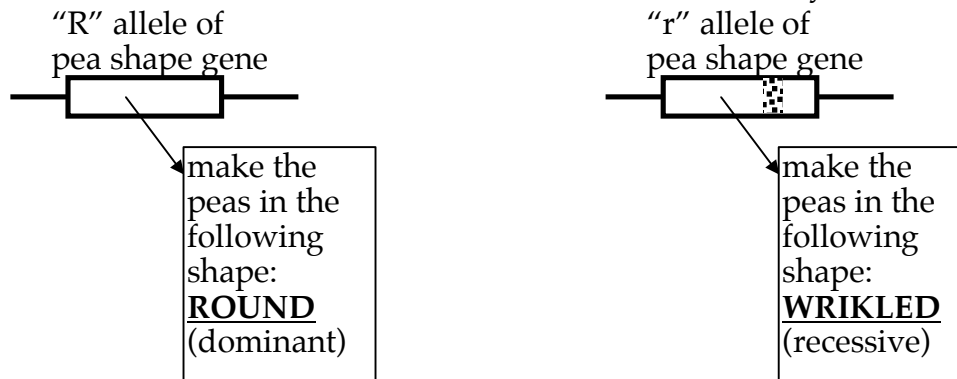
1) A gene is a place for information to be kept:



2) All the alleles of the gene for pea shape have the same basic structure:



3) The different alleles differ in some of the information they contain.



4) So what are Mendel's "particles of inheritance" - the things that everyone has 2 of (one from mom & one from dad)? They are genes and alleles - actually, they are alleles of a gene. To use the example: An Rr individual might say, "I have two copies of the pea shape gene. I got one copy of this gene, in the R form, from my father and I got another copy, this one in the r form, from my mother." or, "I have two copies of the pea shape gene. I got one copy of this gene, the R allele, from my father and I got another copy, this one the r allele, from my mother."

5) A gene is not "made of alleles" (in the sense that a building is made of bricks), it is a place where alleles can be found. The gene contains information; different forms of that information are alleles.

6) The combination of alleles present in an organism (its **genotype**) determines its appearance (**phenotype**) by a set of rules that can be listed as follows:

allele	contribution to phenotype	this information is equivalent to		genotype	phenotype
R	round peas (dominant)	→		RR	round
r	wrinkled peas (recessive)			Rr	round
				rr	wrinkled

(Thanks to the members of the Fall 1998 Development Group for these ideas.)

# Bio 111 Handout for Genetics 3

This handout contains:

1. Today's iClicker Questions
2. Handout for today's lecture

## iClicker Question #3A - at the start of lecture

Mitosis can be described as “duplicate the chromosomes and divide once”. Which of the following phrases best describes meiosis?

- A. duplicate the chromosomes and divide once
- B. do not duplicate the chromosomes and divide once
- C. duplicate the chromosomes and divide twice
- D. duplicate the chromosomes and divide three times
- E. none of the above

As you enter the class, beam in your answer as described below. Answers will be accepted until I have finished with the daily announcements.

## iClicker Question #3B - at the end of lecture

Which of the following diagrams correctly represents the chromosomes of a cell with genotype Aa in interphase of mitosis (before the chromosomes have been replicated)?

Diagram 1

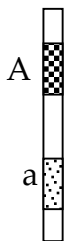


Diagram 2

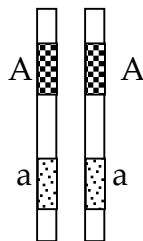


Diagram 3

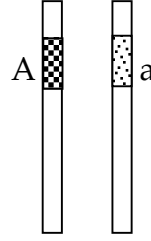
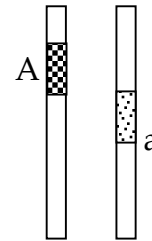


Diagram 4



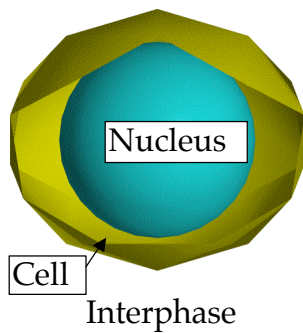
- A. Diagram 1
- B. Diagram 2
- C. Diagram 3
- D. Diagram 4
- E. I don't know / more than one / none of the above.

When I say “start”, beam in your answer as described below; you will have two minutes to enter your answer.

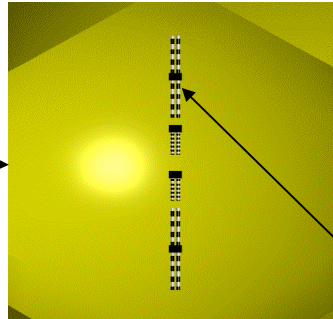
### Beaming in your answers

1. Figure out your answer and select the appropriate letter (A-E).
2. Turn on your iClicker by pressing the “ON/OFF” button; the blue “POWER” light should come on. If the red “LOW BATTERY” light comes on, you should replace your batteries soon.
3. Transmit your answer as follows:
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  - b. The “STATUS” light will flash green to indicate that your answer has been received. If the “STATUS” light flashed red, your answer was not received; you should re-send it until you get a green “STATUS” light.

## Mitosis: Original Observations

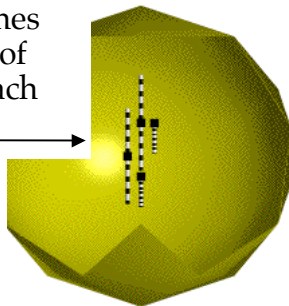


Chromosomes duplicate, become visible, and line up.

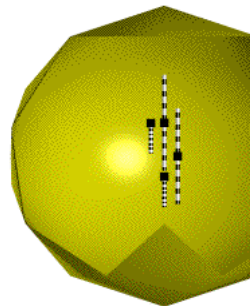


- (1) There are two different kinds of chromosomes: long and short.
- (2) There are two pairs of short ones and two pairs of long ones.

Pairs of chromosomes split. One member of each pair goes to each daughter cell.



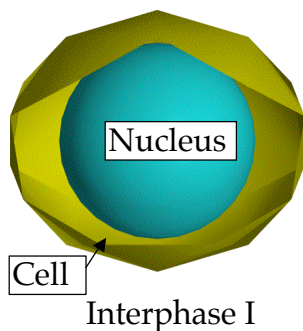
Daughter cell #1



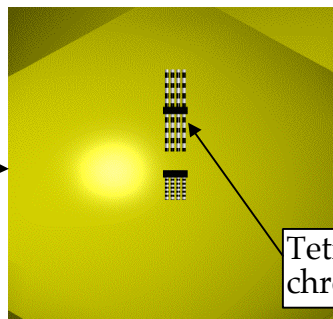
Daughter cell #2

- (1) Each cell now has two separate copies of each type of chromosome. That is, two "longs" and two "shorts".
- (2) These cells are identical to the starting cell.

## Meiosis: Original Observations



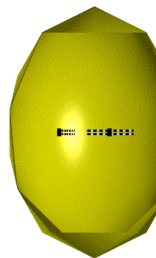
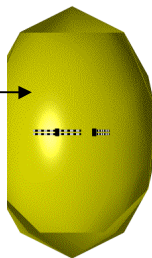
Chromosomes duplicate, become visible, and line up. But, they line up differently!



- (1) All the long ones line up together.
- (2) All the short ones line up together.

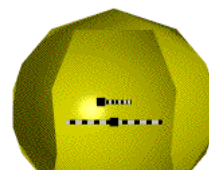
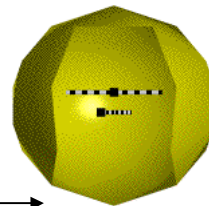
First division of meiosis (Meiosis I)

The tetrads split in Meiosis I.

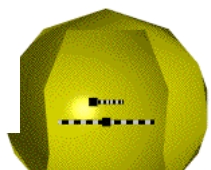
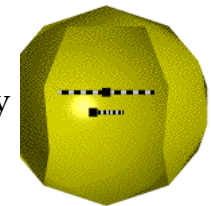


Each cell gets two copies of each type of chromosome.

Both cells divide one more time. (Meiosis II)



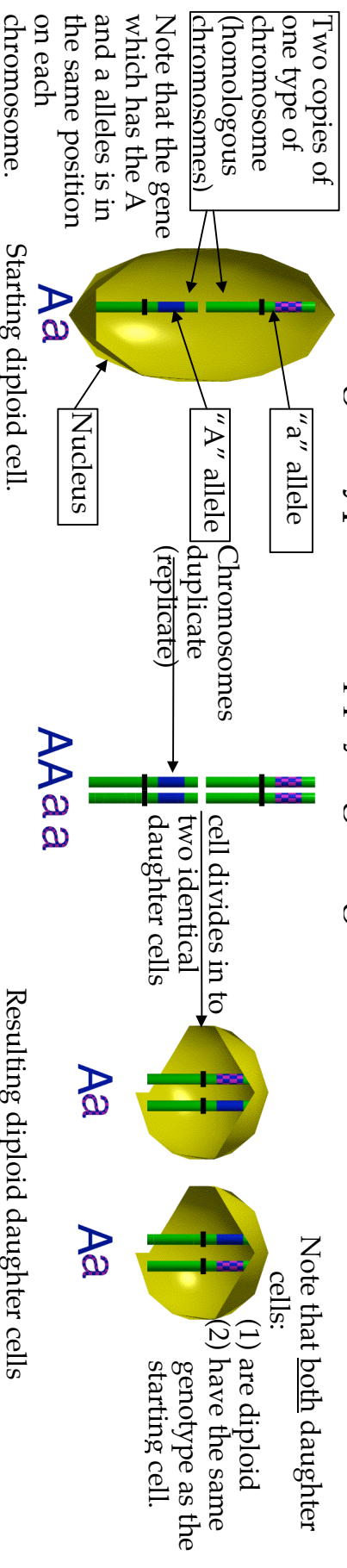
Each cell has only one copy of each type of chromosome: one long and one short (haploid).



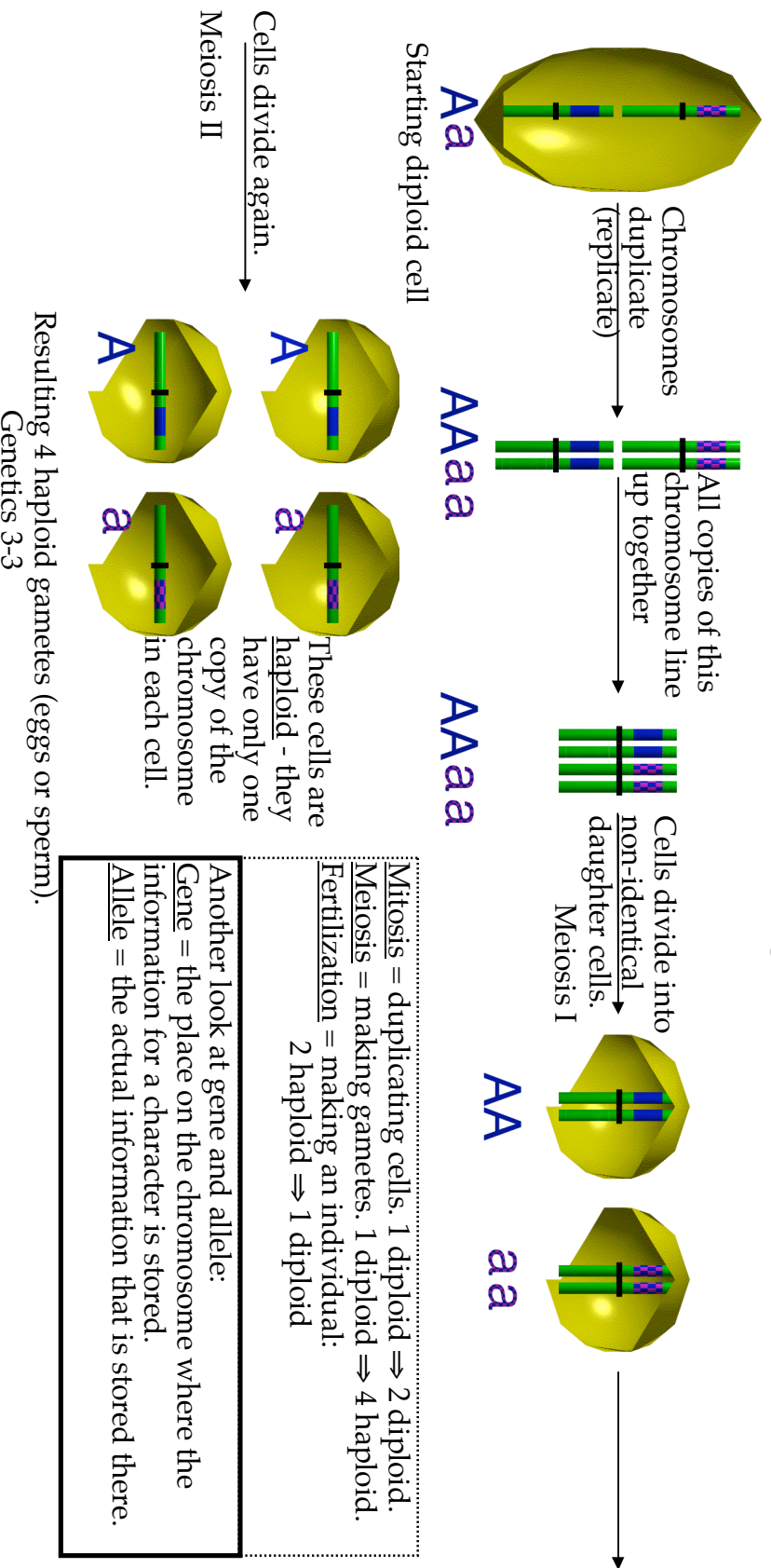
Product: 4 haploid gametes (eggs or sperm)



## Mitosis of a cell with genotype Aa – applying the genetic model:



## Meiosis of a cell with genotype Aa – applying the genetic model:



# Bio 111 Chromosomes & Punnett Squares

After talking to several students about the “two alleles make a gene” misconception, here is a clarification.

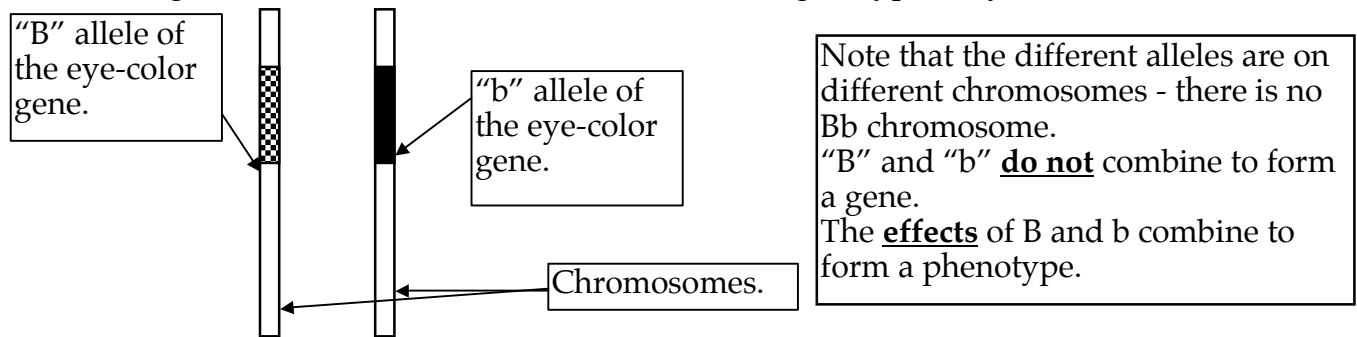
- Suppose eye color were controlled by one gene - this gene has two forms: the following 2 alleles:

allele   contribution to phenotype

B   blue eyes (dominant)

b   white eyes (recessive)

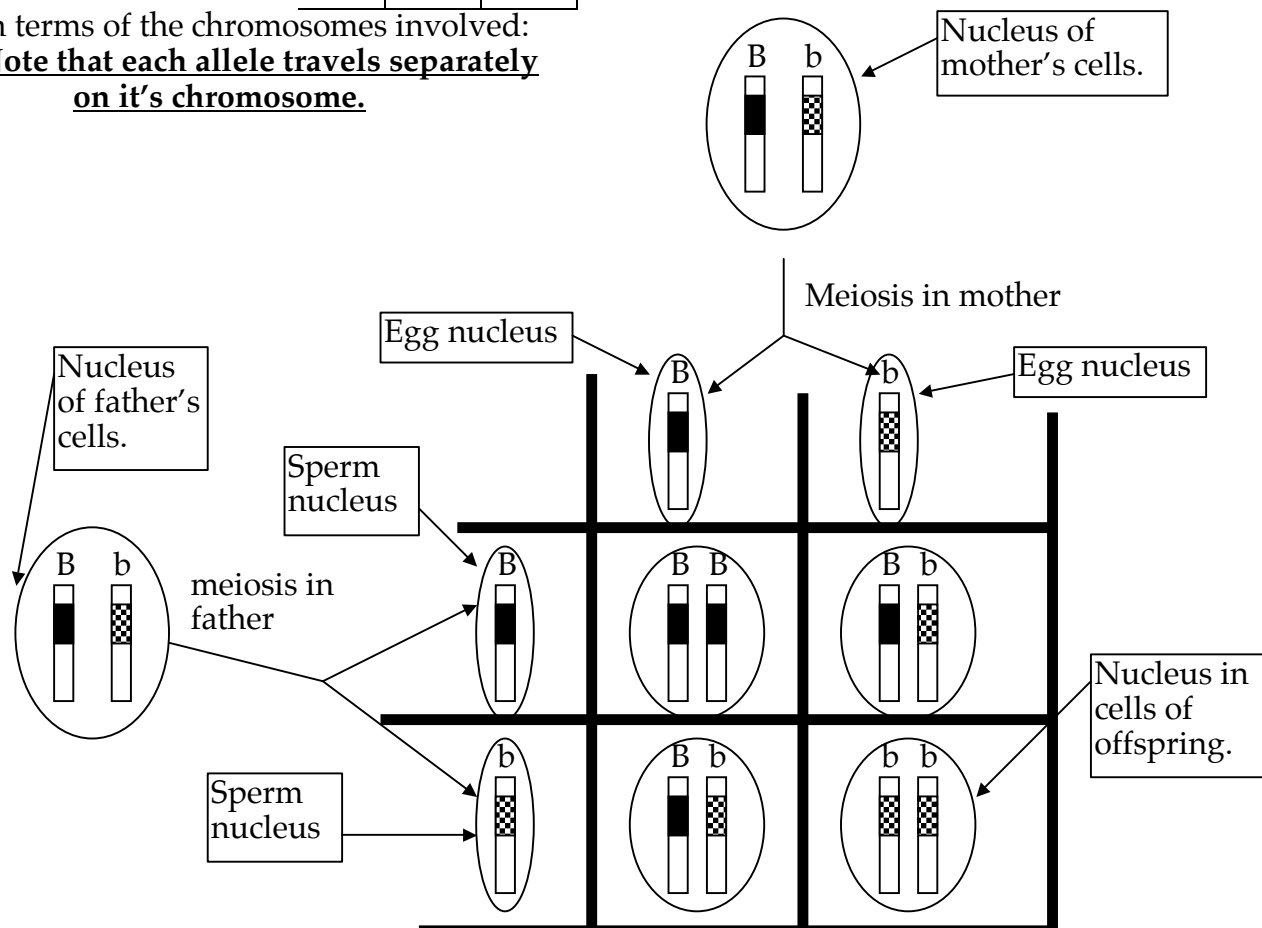
Looking at the chromosomes of an individual with genotype Bb, you'd see:



If two Bb individuals crossed, the resulting Punnett Square would look like:

	B	b
B	BB	Bb
b	Bb	bb

In terms of the chromosomes involved:  
**Note that each allele travels separately on it's chromosome.**





# Bio 111 Handout for Genetics 4

This handout contains:

1. Today's iClicker Questions
2. Handout for today's lecture

## iClicker Question #4A - before lecture

Consider figure 15.10 in Campbell. What would be the expected offspring of an affected mother and a normal father?

- A. all normal
- B. all males normal; all females affected
- C. all males affected; all females normal
- D. all affected
- E. none of the above

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## iClicker Question #4B - after lecture

Can a normal (non-hemophiliac) father have a hemophiliac daughter?

- A. Yes.
- B. No.
- C. I don't know.

When I say "start", beam in your answer as described below; you will have two minutes to enter your answer.

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### **Beaming in your answers**

1. Figure out your answer and select the appropriate letter (A-E).
2. Turn on your iClicker by pressing the "ON/OFF" button; the blue "POWER" light should come on. If the red "LOW BATTERY" light comes on, you should replace your batteries soon.
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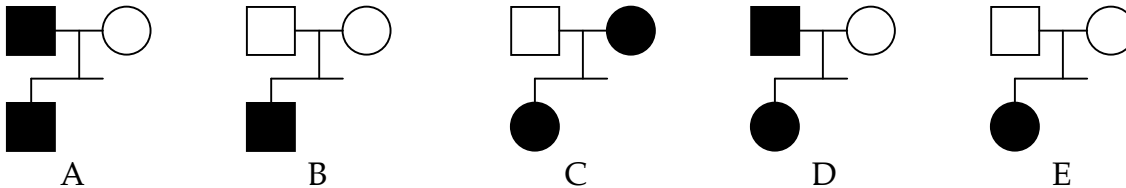
# Bio 111 Handout for Genetics 5

This handout contains:

1. Today's iClikr Questions
2. Handout on counting unrelated carriers

## iClikr Question #5A - before lecture

Which of the following pedigrees corresponds to this description: "An affected father and unaffected mother have an affected daughter"?

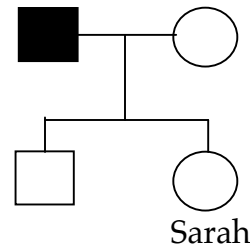


## iClikr Question #5B - after lecture

Consider the following pedigree that shows a genetic disease that is inherited in a sex-linked recessive manner.

allele   contribution to phenotype

$X^A$	normal (dominant)
$X^a$	diseased (recessive)
Y	none



What is Sarah's genotype?

- A.  $X^A X^a$
- B.  $X^A X^A$  or  $X^A X^a$
- C.  $X^A X^A$ ,  $X^A X^a$ , or  $X^a X^a$
- D.  $X^A X^a$  or  $X^a X^a$
- E. I don't know.

### Beaming in your answers

1. Figure out your answer and select the appropriate letter (A-E).
2. Turn on your iClikr by pressing the "ON/OFF" button; the blue "POWER" light should come on. If the red "LOW BATTERY" light comes on, you should replace your batteries soon.
3. Transmit your answer as follows:
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## Bio 111 Counting “Unrelated Carriers”

In lecture, I talked about “counting unrelated carriers” - finding out how many people had to bring in a disease allele to explain a particular pedigree. This was useful in the case where more than one mode of inheritance was *possible* but you were asked to determine which was *more likely*. There are many ways to figure this out; this handout describes one possible way. Thanks to Silas for suggesting this.

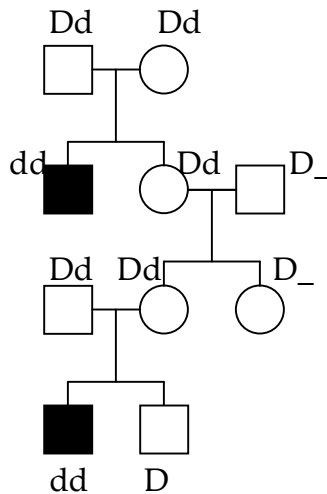
More precisely, we are counting the number of people who had to bring a disease allele into the family. Why is this important? Because, since any given genetic disease is usually quite rare, individuals with a disease allele must also be rare. Therefore, a mode of inheritance that requires more of these rare individuals is less likely than one that requires fewer.

Put another way, we are trying to account for each of the disease alleles found in individuals in the pedigree and to count the number of individuals who brought these alleles into the family.

Here is one way to do it:

1. Work out the pedigree and find out the *possible* mode(s) of inheritance. If only one is possible, this technique is not necessary. If more than one is possible, continue.
2. Fill in the genotypes of all individuals in the pedigree.
3. Starting with the affected individual at the lowest part of the pedigree, star all the individual(s) that gave this individual a disease allele.
4. Work your way up the pedigree from each starred individual. If their disease allele can be traced to another person in the pedigree, erase their star and move it to the source of the disease allele.
5. When you have reached the top of the pedigree, you will have accounted for all the disease alleles. The number of starred individuals is the number of ‘unrelated carriers’.

Here is an example; suppose you saw the following pedigree:



This pedigree is not consistent with Autosomal Dominant inheritance.

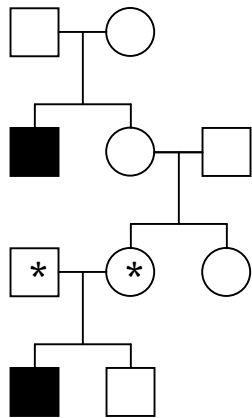
It is consistent with Autosomal Recessive and Sex-Linked Recessive.

Start with Autosomal Recessive:

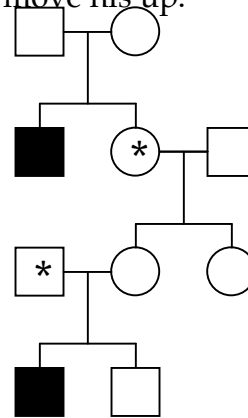
<u>allele</u>	<u>contribution to phenotype</u>
D	normal (dominant)
d	diseased (recessive)

The genotypes of individuals in the pedigree are shown at the left.

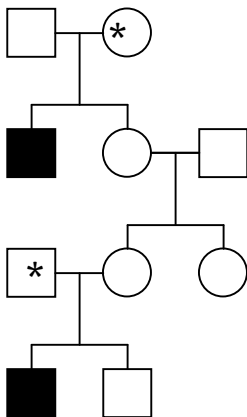
a) Step (3). Start with the son at the bottom of the pedigree. He got his d's from his parents. Star them.



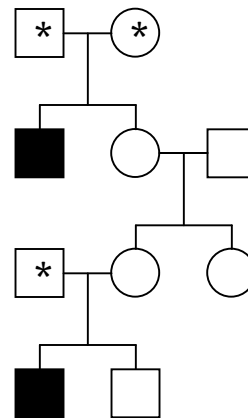
b) Step (4). Where did the \*ed mother get her d? From her mother. To show this, move her star up. We don't know where the father got his \*, so don't move his up.



c) Step (4) again. Where did the mother from (b) get her d? From her mother or father. So, you should star one or the other of the parents at the top of the pedigree. Because we don't know anything about the parents of the top generation, it doesn't matter which one we star, so star the mother for now (we may revise this later).



d) Step (3). Now you must account for the disease alleles in the other affected male. They came from his parents. Note that you have already \*ed his mother; there is no need to \* her twice. Since you have no information about the parents of the \*ed individuals, you can't move their start. Therefore, you are done.

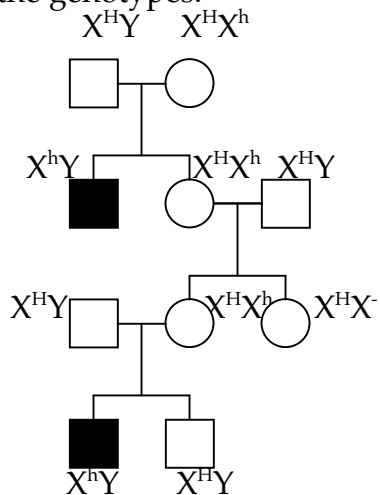


Now you have accounted for all of the d's in the pedigree. Three unrelated carriers are required to explain this pedigree as autosomal recessive.

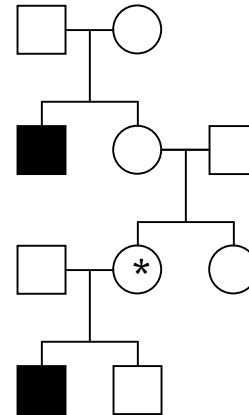
On the next page, we work it through for Sex-Linked Recessive:

allele	contribution to phenotype
$X^H$	normal (dominant)
$X^h$	disease (recessive)
Y	none

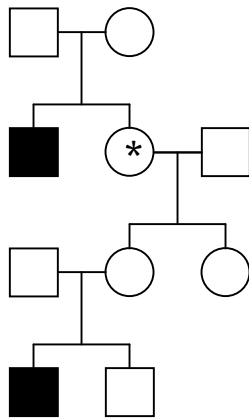
Here are the genotypes:



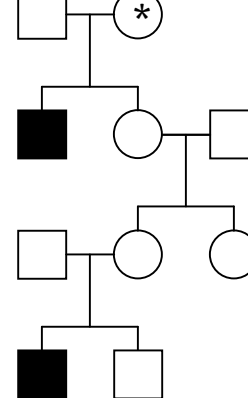
e) Step (3). Start with the affected son at the bottom of the pedigree. He got his  $X^h$  from his mother. Star her.



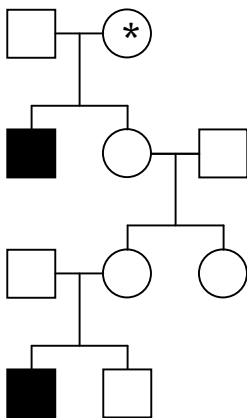
f) Step (4). The \*ed mother in (e) got her  $X^h$  from her mother. Move the \* up to her.



g) Step (4) again. The \*ed female from (f) got her  $X^h$  from her mother. Move the \* up to her. Because we don't know where she got her  $X^h$ , we cannot move her \*.



h) Step (3). Start from the other affected son. He got his  $X^h$  from his mother. She is already \*ed. There is no more information in the pedigree, so you are done.



You have accounted for all the  $X^h$ 's. There is only one unrelated carrier required to explain this pedigree as sex-linked recessive.

Since an autosomal recessive explanation of **this pedigree** requires 3 unrelated carriers and a sex-linked recessive explanation of **this pedigree** requires only one of these rare individuals, sex-linked recessive is the *most likely mode of inheritance*.



# Bio 111 Handout for Genetics 6

This handout contains:

1. Today's iClicker Questions
2. Yet more genetics practice problems & solutions.

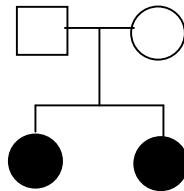
## iClicker Question #6A - before lecture

Consider Question 1.1.2 from Chapter 1 of APAIB (page 22). Which of the following is a good answer for part (a) (i)?

- |  |  |
|--|--|
| (A) <u>Allele</u> <u>Contribution to Phenotype</u><br>X <sup>D</sup> normal (dominant)<br>X <sup>d</sup> cystic fibrosis (recessive)<br>Y none | (B) <u>Allele</u> <u>Contribution to Phenotype</u><br>D normal (dominant)<br>d cystic fibrosis (recessive) |
| (C) <u>Allele</u> <u>Contribution to Phenotype</u><br>X <sup>D</sup> cystic fibrosis (dominant)<br>X <sup>d</sup> normal (recessive)<br>Y none | (D) <u>Allele</u> <u>Contribution to Phenotype</u><br>D cystic fibrosis (dominant)<br>d normal (recessive) |
- (E) none of the above

## iClicker Question #6B - after lecture

Consider the following pedigree for a rare genetic disease. Based on this pedigree, what is the most likely mode of inheritance for this disease?



- A. Autosomal recessive
- B. Sex-linked recessive
- C. Autosomal dominant
- D. Autosomal recessive and Sex-linked recessive are equally likely
- E. I don't know / none of the above.

### Beaming in your answers

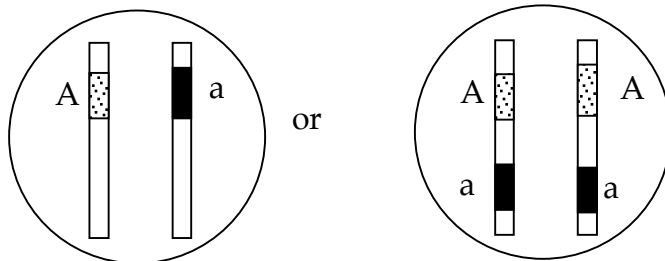
1. Figure out your answer and select the appropriate letter (A-E).
2. Turn on your iClicker by pressing the "ON/OFF" button; the blue "POWER" light should come on. If the red "LOW BATTERY" light comes on, you should replace your batteries soon.
3. Transmit your answer as follows:
  - a. Press the button corresponding to the answer you've selected (A thru E).
  - b. The "STATUS" light will flash green to indicate that your answer has been received. If the "STATUS" light flashed red, your answer was not received; you should re-send it until you get a green "STATUS" light.

# Bio 111: Yet More Genetics Practice Problems

These were taken from Exam I Fall 1999.

## Question 1: Chromosomes, Mitosis, Meiosis, & Fertilization (20 points)

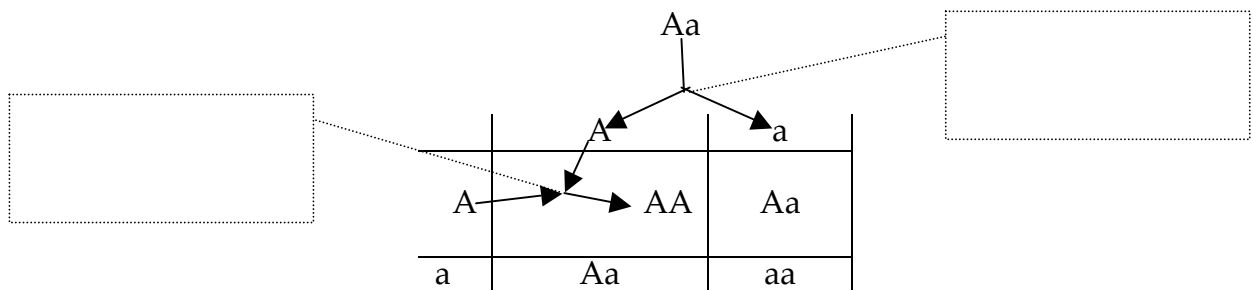
a) Which of the following is a correct representation of the chromosomes of an individual whose genotype is Aa? Circle the correct one and explain your reasoning. (5 pts)



Explanation:

b) Using diagrams like those in part (a) draw the chromosomes of a **hemophiliac human male** at **metaphase of mitosis**. You need not show all 46 chromosomes, only those that are involved in hemophilia. Be sure to indicate the location of the gene for hemophilia as well as the alleles present. (6 pts)

c) Which processes are represented by the arrows indicated in the diagram below. Fill in the boxes with the appropriate term (choose from mitosis, meiosis, replication, fertilization). (4 pts)



d) What is incorrect about the following Punnett Square? state what is wrong and explain why it is wrong. (5 pts)

	I <sup>A</sup>	I <sup>B</sup>	i
I <sup>A</sup>	I <sup>A</sup> I <sup>A</sup>	I <sup>A</sup> I <sup>B</sup>	I <sup>A</sup> i
I <sup>B</sup>	I <sup>B</sup> I <sup>A</sup>	I <sup>B</sup> I <sup>B</sup>	I <sup>B</sup> i
i	I <sup>A</sup> i	I <sup>B</sup> i	ii

### Question 3: Blood type (15 points)

Fred was separated from his parents at birth; his blood type is A. (5 pts each)

a) Fred thinks that Jane (blood type A) and Peter (blood type AB) may be his parents. Given the information so far, is it possible that Fred is Peter and Jane's son? Circle your answer and explain your reasoning.

Yes

No

Explanation:

b) Jane's parents are both type AB. Given the information so far, is it possible that Fred is Peter and Jane's son? Circle your answer and explain your reasoning.

Yes

No

Explanation:

c) Fred has a child with blood type O. Given the information so far, is it possible that Fred is Peter and Jane's son? Circle your answer and explain your reasoning.

Yes

No

Explanation:

### Question 5: Flies (25 points)

You are studying a strange (and hypothetical) population of flies; female flies are XX; male flies are XY. At first, you find flies in three colors: red, green, and blue. You do some crosses to find out how color is inherited:

- a)      Cross 1:
- |                |                |
|----------------|----------------|
| green female X | red male       |
|                | $\downarrow$   |
|                | 1/2 red female |
|                | 1/2 green male |

Based on this information only, give a genetic model of color inheritance in these flies:

- i) Where is the gene that controls color in these flies? autosome X-chromosome  
(circle one) (2 pts)

- ii) Define appropriate allele symbols and give their contribution to phenotype: (2 pts)
- | <u>allele</u> | <u>contribution to phenotype</u> |
|---------------|----------------------------------|
|---------------|----------------------------------|

- iii) Give the genotypes of: female parent \_\_\_\_\_ male parent \_\_\_\_\_  
(2 pts each)

- b) Cross 2:  $\frac{1}{2}$  green female X  $\frac{1}{2}$  blue male  
 $\downarrow$   
 $\frac{1}{2}$  blue female  
 $\frac{1}{2}$  green male

Based on the information in part (b) only, give a genetic model of color inheritance in these flies:

- i) Where is the gene that controls color in these files?   autosome      X-chromosome  
(circle one) (2 pts)

- ii) Define appropriate allele symbols and give their contribution to phenotype: (2 pts)
- | <u>allele</u> | <u>contribution to phenotype</u> |
|---------------|----------------------------------|
| $A$           | dominant                         |
| $a$           | recessive                        |

- iii) Give the genotypes of: female parent \_\_\_\_\_ male parent \_\_\_\_\_  
(2 pts each)



c)      Cross 3:

red female	X	blue male
		$\downarrow$
		1/2 purple female
		1/2 red male

Based on the information in parts (a), (b), and (c), give a genetic model of color inheritance in these flies:

- i) Where is the gene that controls color in these flies?    autosome    X-chromosome  
(circle one) (2 pts)
- ii) Define appropriate allele symbols and give their contribution to phenotype: (3 pts)  
allele                      contribution to phenotype

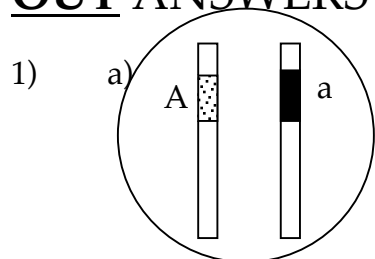
iii) Give the genotypes of: female parent \_\_\_\_\_ male parent \_\_\_\_\_  
(2 pts each)





## SOLUTIONS:

DO NOT LOOK AT THESE UNTIL YOU HAVE **WRITTEN OUT** ANSWERS TO THE PROBLEMS.

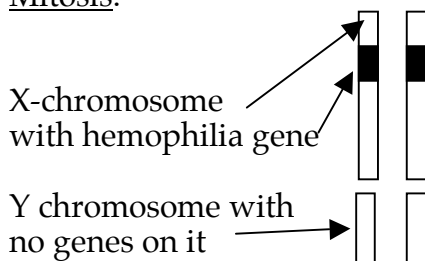


There are several reasons why this is correct:

- (1) each chromosome has one copy of each gene
- (2) the genotype of this cell is Aa; the other would be AAaa

b) (always 2 copies of both chromosomes at these stages)

Mitosis:



c)  $Aa \Rightarrow A + a$  is meiosis

$A + A \Rightarrow AA$  is fertilization

d) This implies that the parents' genotypes were both  $I^A I^B i$  - which would imply that the parent had three copies of the blood-type gene ('triploid'). Although there are 3 different alleles for blood type, an individual can carry a maximum of 2.

3) a) **Yes**. If Jane is  $I^A \_$  and Peter is  $I^A I^B$ , they can have a son with type A blood; he will be either  $I^A I^A$  or  $I^A i$ , depending on Jane's genotype.

b) **Yes** If Jane's parents are both  $I^A I^B$ , then they can have a type A child; her genotype will be  $I^A I^A$  so that Fred's genotype must then be  $I^A I^A$ .

c) **No** To have a type O child, Fred must have an i allele. However, from part (b), he does not - therefore, Jane & Peter cannot be his parents.

5) a) i) X-chromosome      ii) allele      contribution to phenotype  
 $X^R$       red dominant  
 $X^r$       green recessive  
 $Y$       none

iii) male =  $X^R Y$       female =  $X^r X^r$   
 b) i) X-chromosome      ii) allele      contribution to phenotype  
 $X^B$       blue dominant  
 $X^r$       green recessive  
 $Y$       none

iii) male =  $X^B Y$       female =  $X^r X^r$

c) i) X-chromosome  
 ii) allele      contribution to phenotype  
 $X^B$       blue dom. to green; inc. dom. to red  
 $X^R$       red dom. to green; inc. dom. to blue  
 $X^r$       green recessive  
 $Y$       none

iii) male =  $X^B Y$       female =  $X^R X^R$

# Bio 111 Handout for Genetics 7

This handout contains:

1. Today's iClicker Questions
2. Handouts for today's lecture

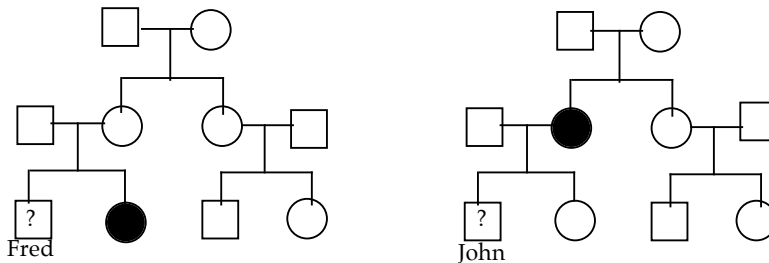
## iClicker Question #7A - before lecture

If my wife is a carrier for the X-linked recessive "disease" red-green colorblindness and I have normal color vision, what is the chance that my son will be red-green color blind?

- A. 0
- B.  $\frac{1}{4}$
- C.  $\frac{1}{2}$
- D.  $\frac{3}{4}$
- E. 1

## iClicker Question #7B - after lecture

Shown below are two pedigrees for a rare autosomal recessive genetic disease. Fewer than 1 in 1000 people are carriers for this disease.



Fred and John are as-yet unborn children of parents who are concerned that they may be affected with the genetic disease. Based on the above information, which individual, Fred or John, has a greater risk of being affected by the disease.

- A. Fred has a greater risk of being diseased
- B. John has a greater risk of being diseased
- C. Both have an equal risk of being diseased
- D. It is not possible to tell who has the higher risk of being diseased
- E. I don't know.

### Beaming in your answers

1. Figure out your answer and select the appropriate letter (A-E).
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# Bio 111 Probability Simulation

Consider an autosomal recessive genetic disease. That is:

the disease is controlled by one gene on an autosome with two alleles:

D - normal (dominant)

d - diseased (recessive)

If the father is Dd, there is a 1/2 chance that he will give a d allele to his offspring.

If the mother is Dd, there is a 1/2 chance that she will give a d allele to her offspring.

If the child gets a d from both mom and from dad, he or she will have the disease.

So, overall, if two carrier (Dd) parents have children, each child has a 1/4 chance of having the disease.

	D	d
D	DD	Dd
d	Dd	dd

We will simulate a family with 4 children.

For each child:

(1) flip a coin once to choose the allele that the **father** will contribute.

If it is **heads**, the allele is **D**; write the allele in the appropriate box.

If it is **tails**, the allele is **d**; write the allele in the appropriate box.

(2) flip a coin once to choose the allele that the **mother** will contribute.

If it is **heads**, the allele is **D**; write the allele in the appropriate box.

If it is **tails**, the allele is **d**; write the allele in the appropriate box.

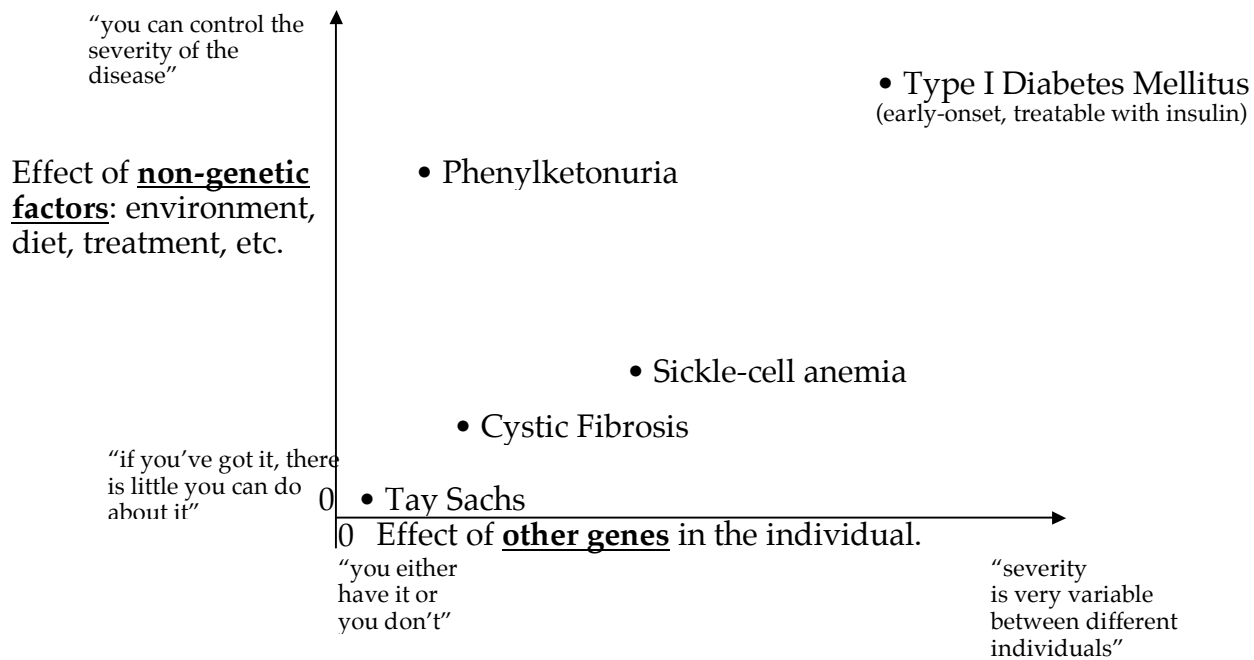
(you will flip the coin a total of 8 times)

	Genotype		Phenotype
	Allele from Father	Allele from Mother	
First Child			
Second Child			
Third Child			
Fourth Child			

# Bio 111 Misc. Facts about Genetic Diseases

## 1) Influence of other factors on severity of selected genetic diseases

(based on figure from "The Metabolic and Molecular Basis of Inherited Disease", Scrivner & al.)



## 2) Frequency of selected genetic diseases

### X-linked

Duchenne Muscular dystrophy	1 in 3000 males
Hemophilia	1 in 10,000 males

### Autosomal Recessive

Phenylketonuria	1 in 12,000 (average)
Tay-Sachs Disease	1 in 3,000 (Ashkenazi Jews)
	1 in 320,000 (American non-Jews)
Cystic Fibrosis	1 in 2000 (American Caucasian)
	1 in 17,000 (African-American)

### Autosomal Dominant

Marfan Syndrome	1 in 20,000
Achondroplaisic Dwarfism	1 in 50,000

