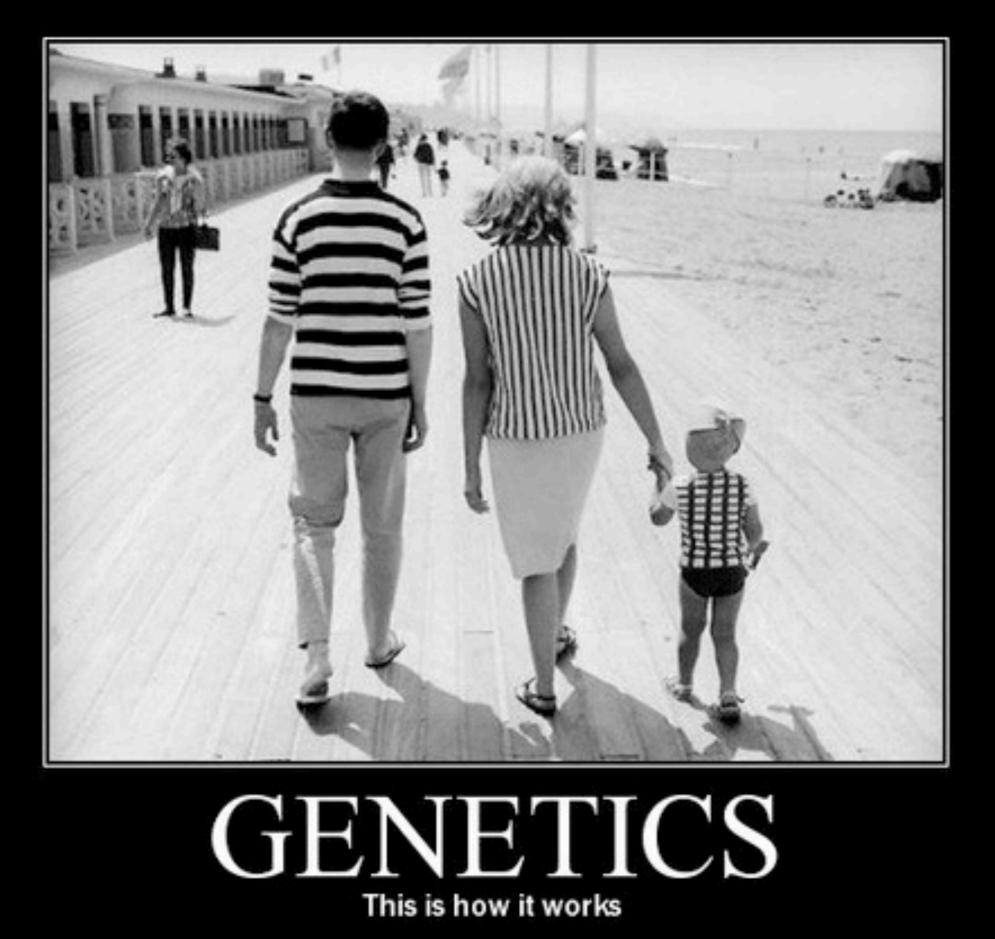
Big Idea 3: Living systems store, retrieve, transmit and respond to information essential to life processes.

Thursday, February 9, 17

Enduring understanding 3.A: Heritable information provides for continuity of life. Essential knowledge 3.A.3: The chromosomal basis of inheritance provides an understanding of the pattern of passage (transmission) of genes from parent to offspring.

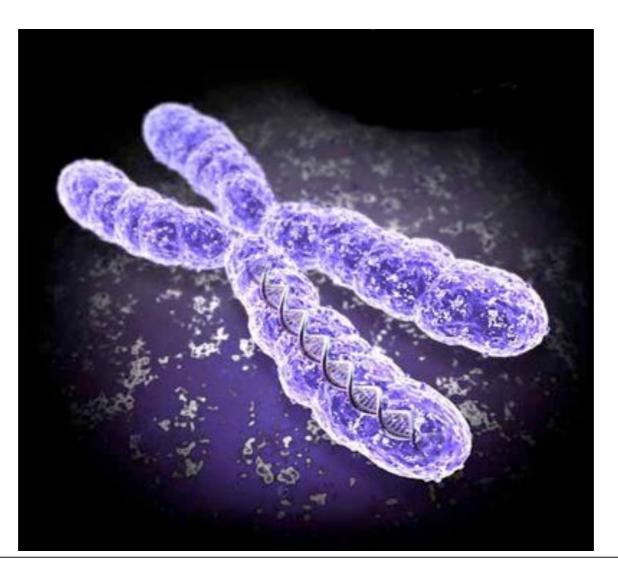
a. Rules of probability can be applied to analyze passage of single gene traits from parent to offspring.



motivateusnot.com

Transition to Genetics

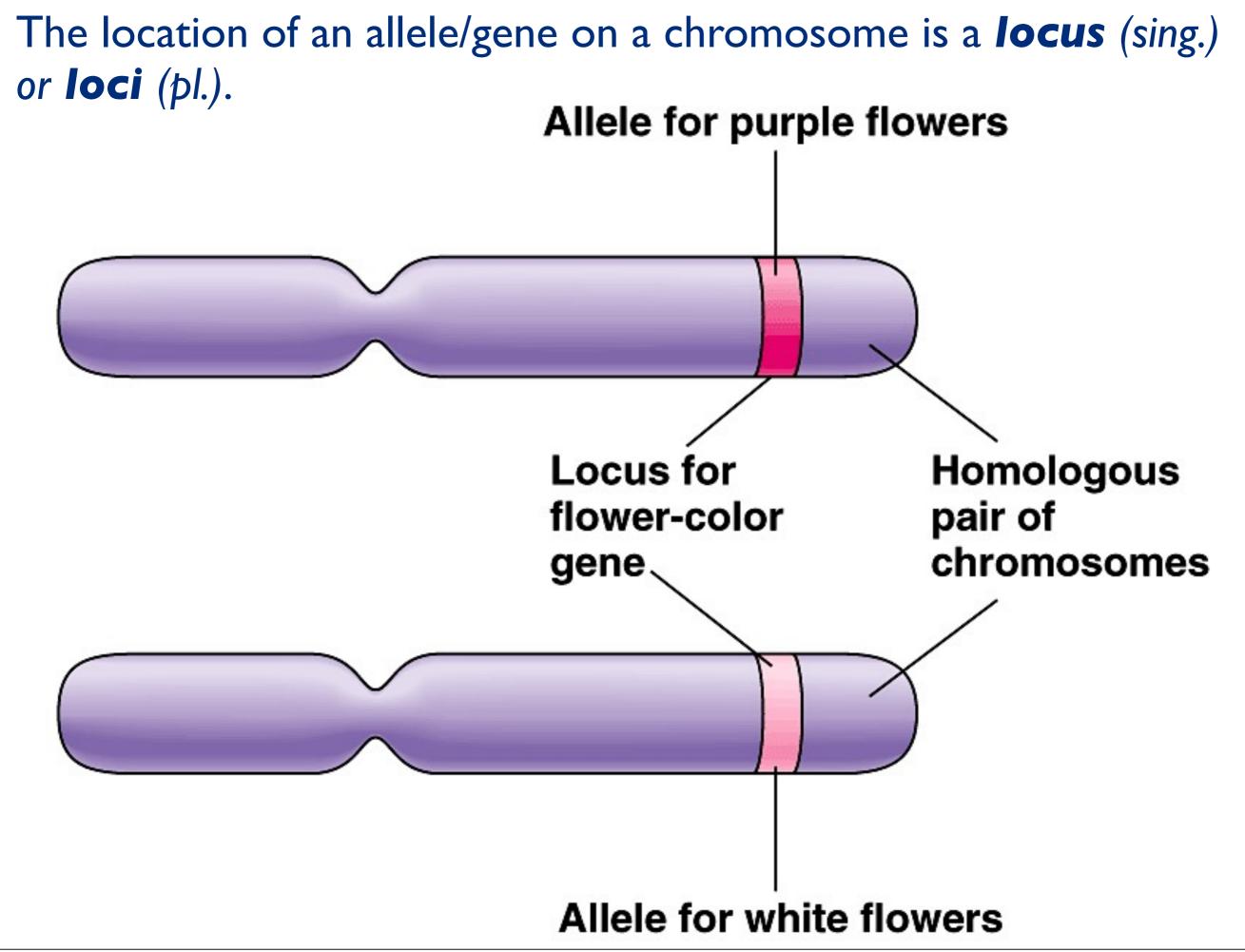
Main Idea: The principles of meiosis provide the foundation and framework for understanding the inheritance of traits.



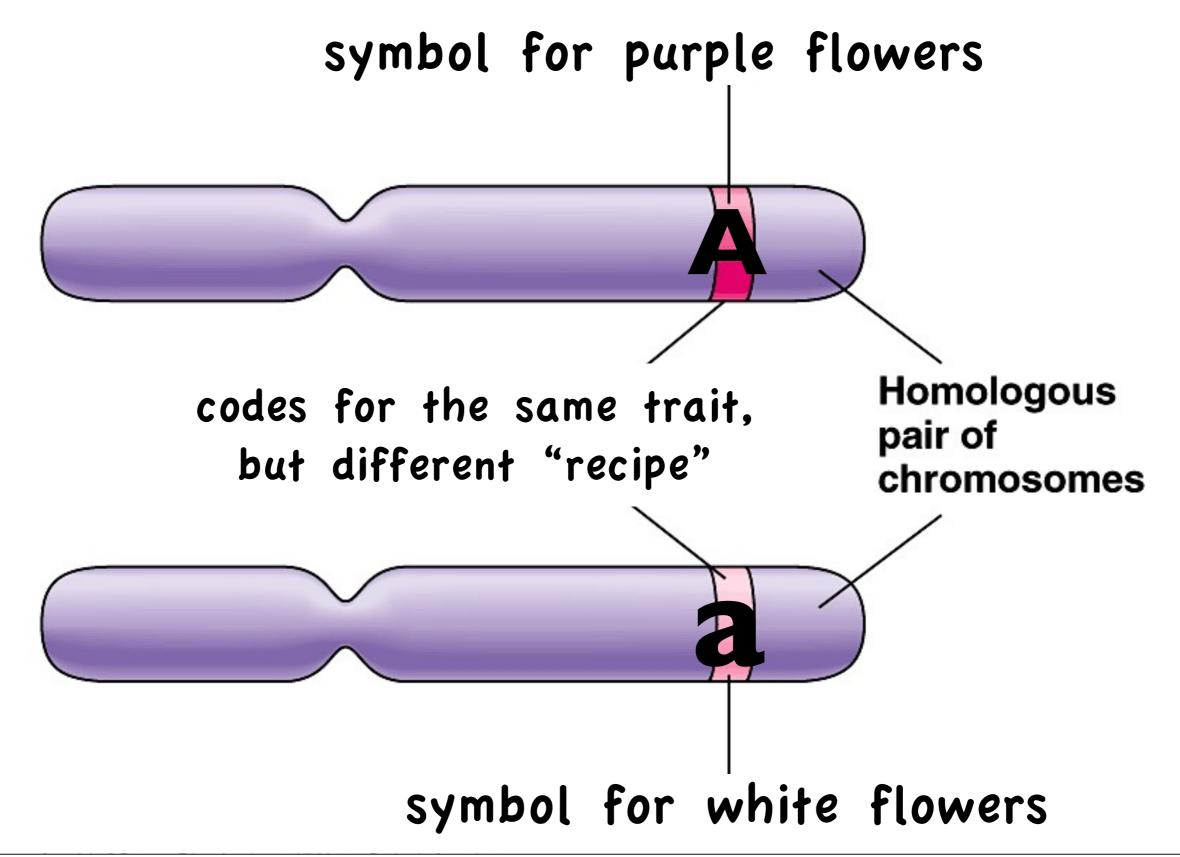
Alleles

- Alleles are alternate forms of genes.
- Genes are strings of nucleotides that make up DNA.
- DNA wraps around proteins to form chromosomes.
- During reproduction parents donate chromosomes (carrying the alleles) that determine the traits of their offspring.

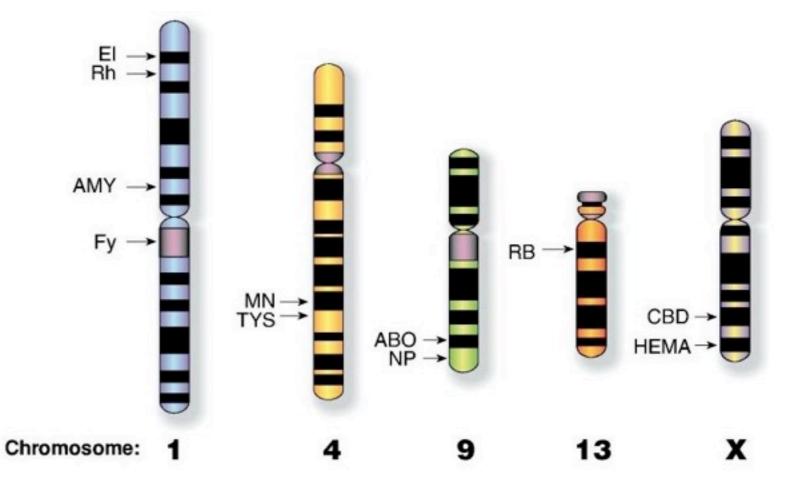
BUT, here is the key point! Parents do donate single alleles to their offspring rather single chromosomes. The chromosomes are packages of hundreds of alleles, to understand inheritance you have to understand the behavior of chromosomes, in other words meiosis!



We can not see genes so we use symbolic letters to represent the genes we can not see.

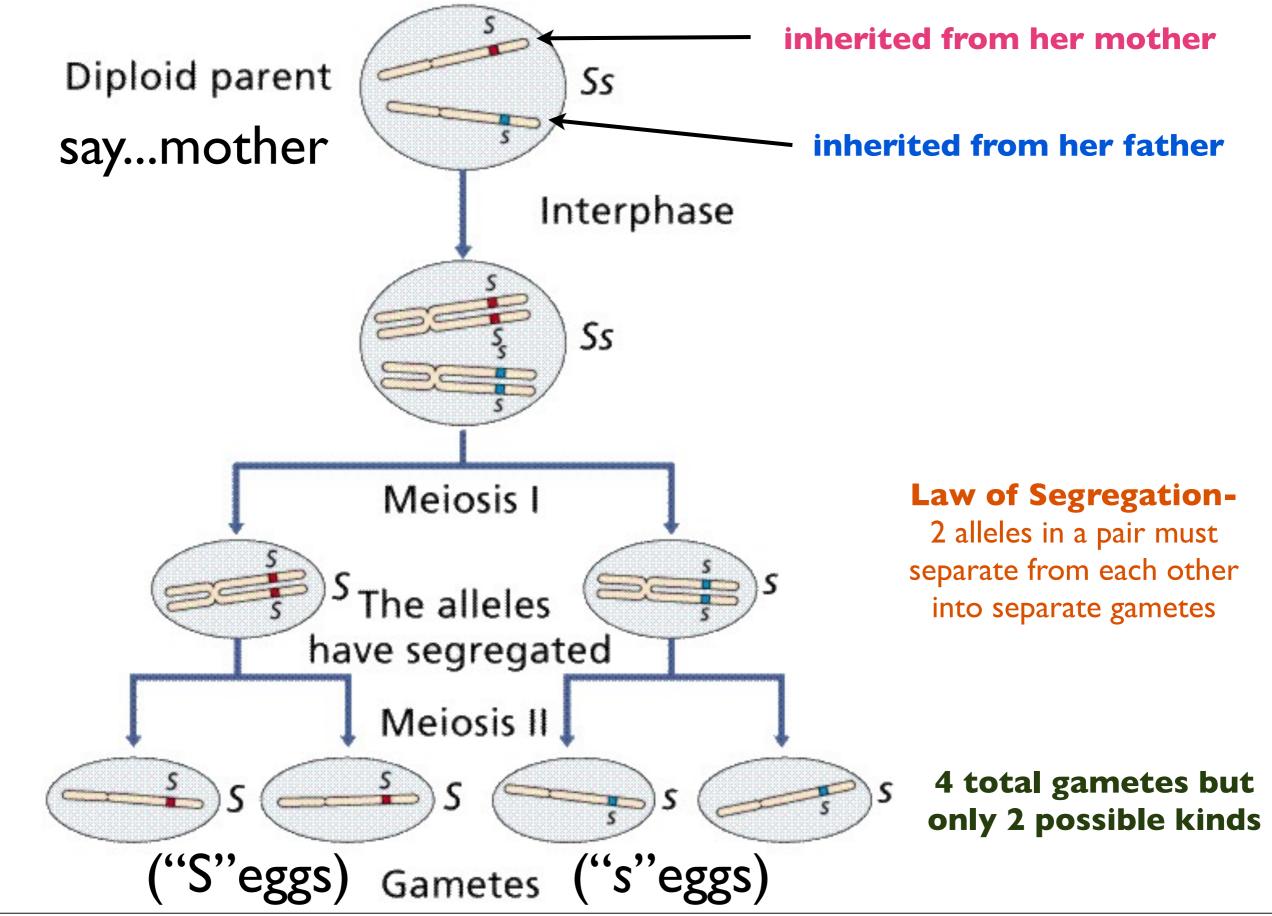


A Chromosome Carries Many Alleles/Genes

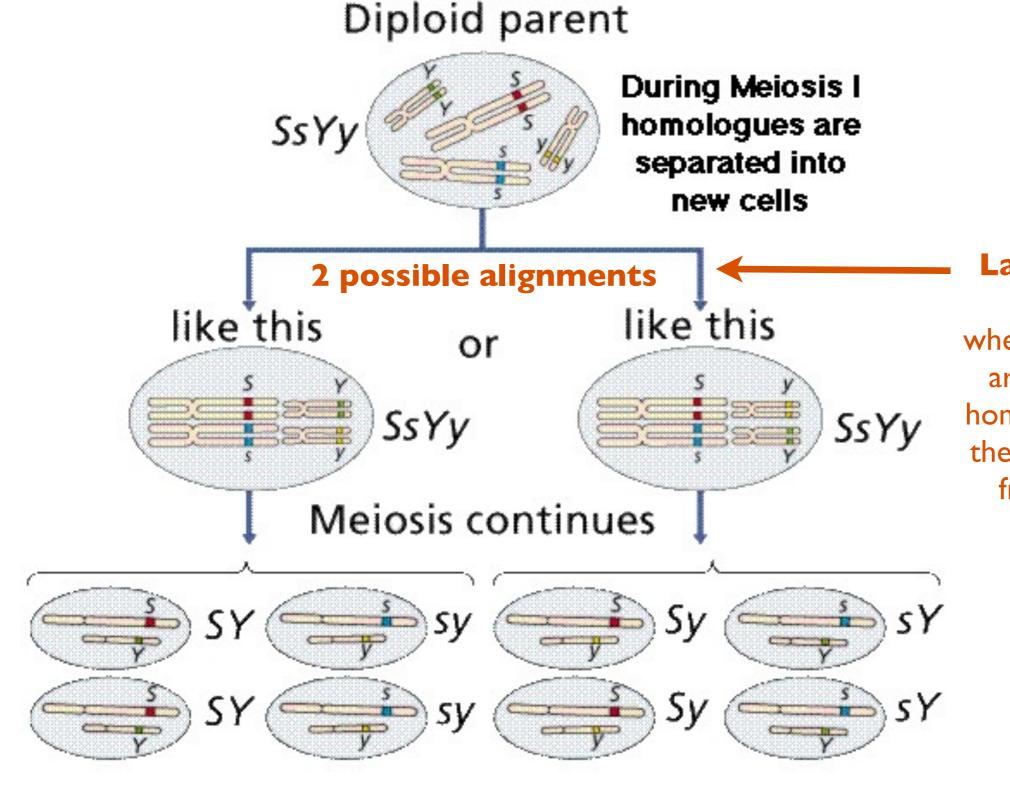


	GENE SYMBOLS
ABO	ABO blood type
AMY	Production of amylase enzyme
CBD	One form of colour blindness
EI	Shape of red blood cells
Fy	Duffy blood type
HEMA	Production of a blood clotting factor
NP	Structure of nails and kneecaps
Rh	Rhesus blood type
RB	Retinoblastoma (a cancer of the eye)
MN	MN blood type
TYS	Skin structure

Follow the chromosome, follow the traits!



We Can Follow Two Traits Simultaneously

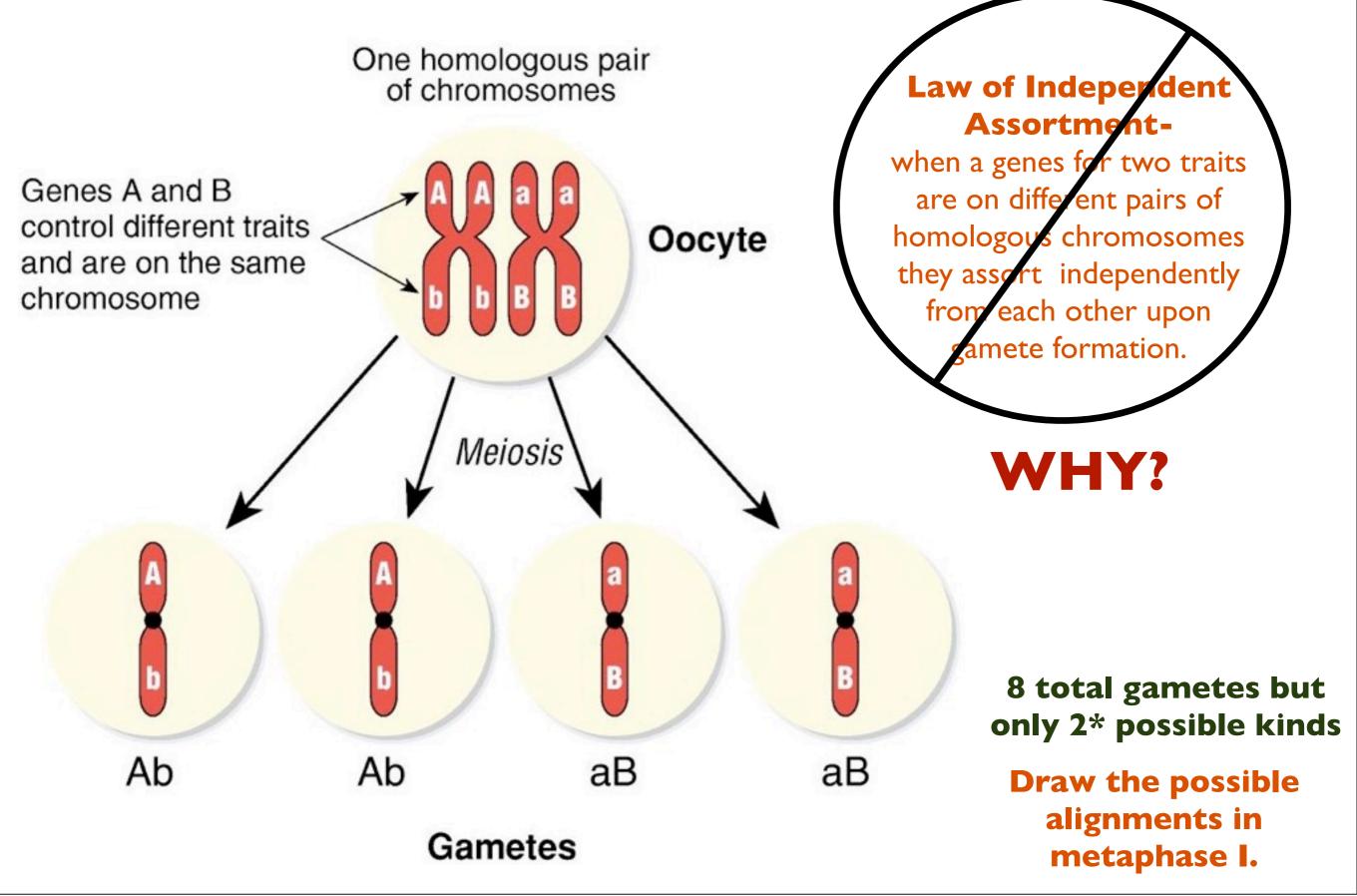


Law of Independent Assortment-

when a genes for two traits are on different pairs of homologous chromosomes they assort independently from each other upon gamete formation.

S assorts with Y or y s assorts with Y or y 8 total gametes but only 4 possible kinds

We Can Follow Two Traits Simultaneously

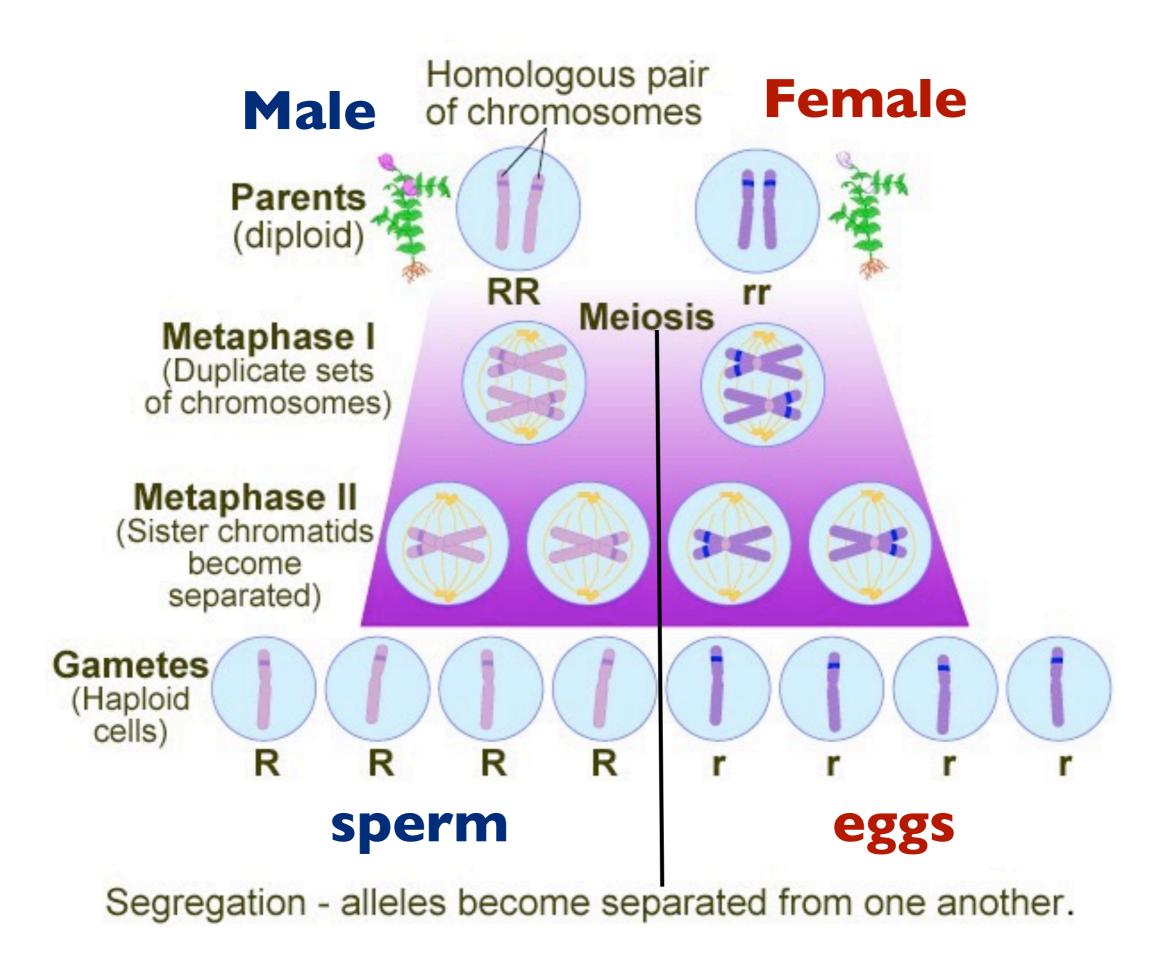


What do all of the last three slides have in common?

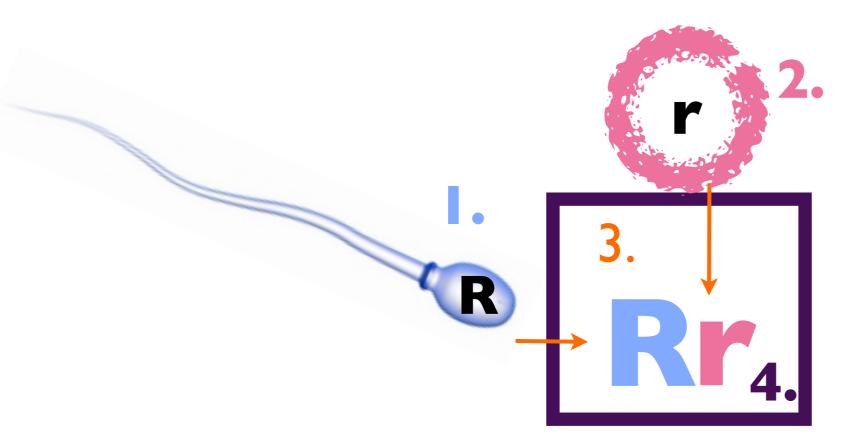
We know the possible gametes, the different types of sperm or eggs that "could" be produced by the parent.

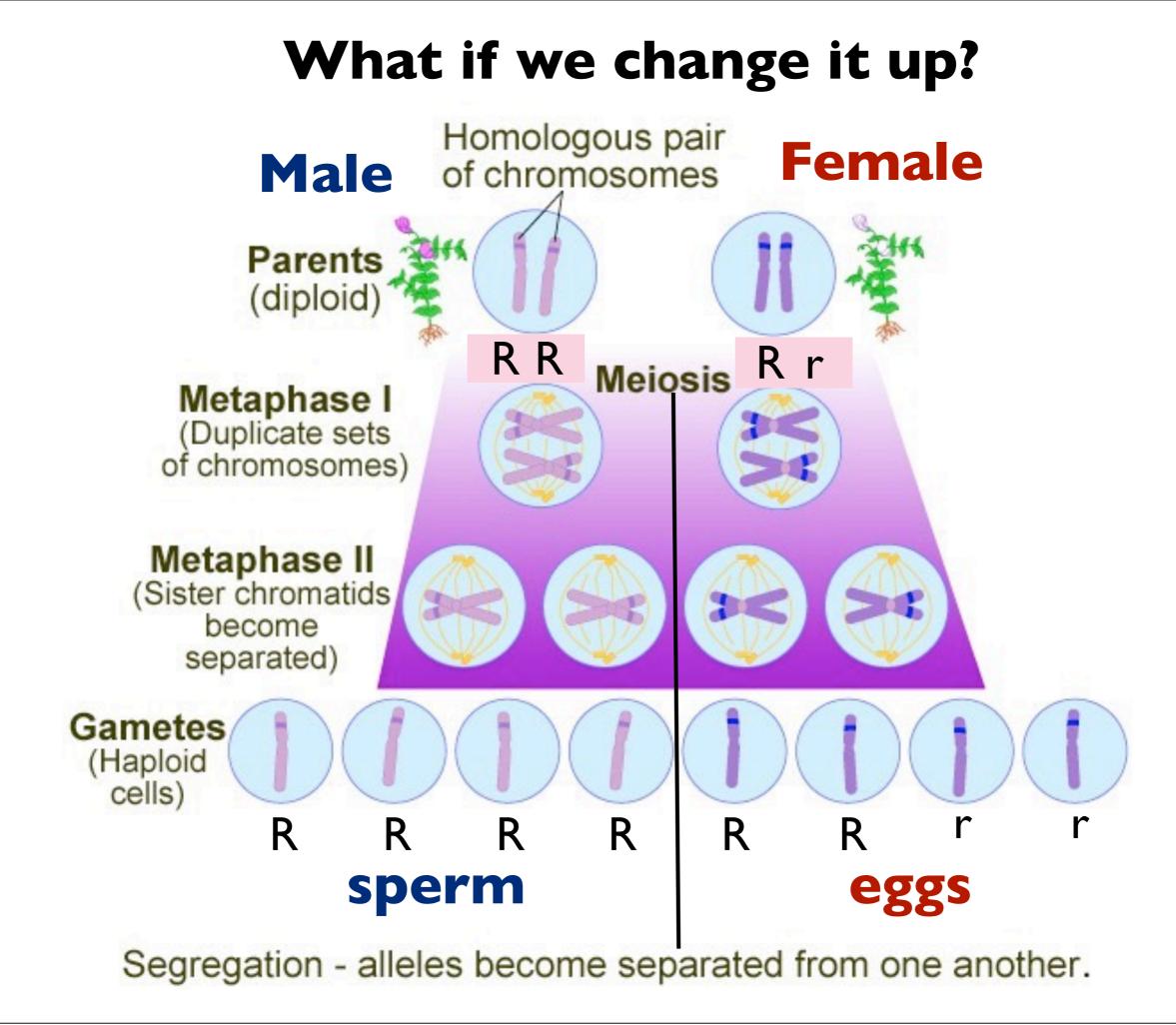
I.) We know that offspring result from the fusion of sperm with egg.

2.) If we know the types of possible sperm3.) and we know the types of possible eggs4.) then we can predict the possible offspring

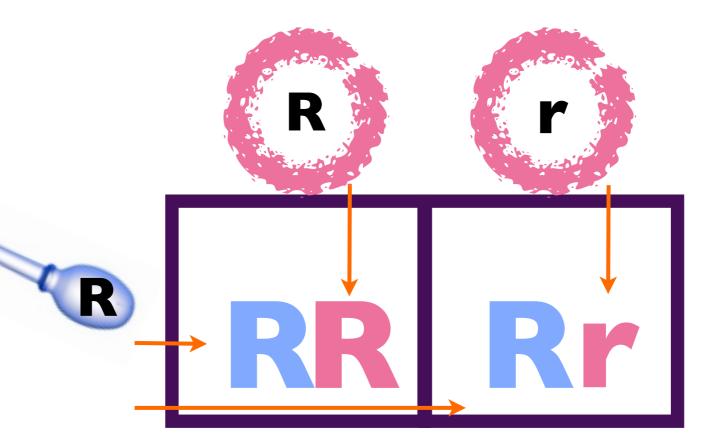


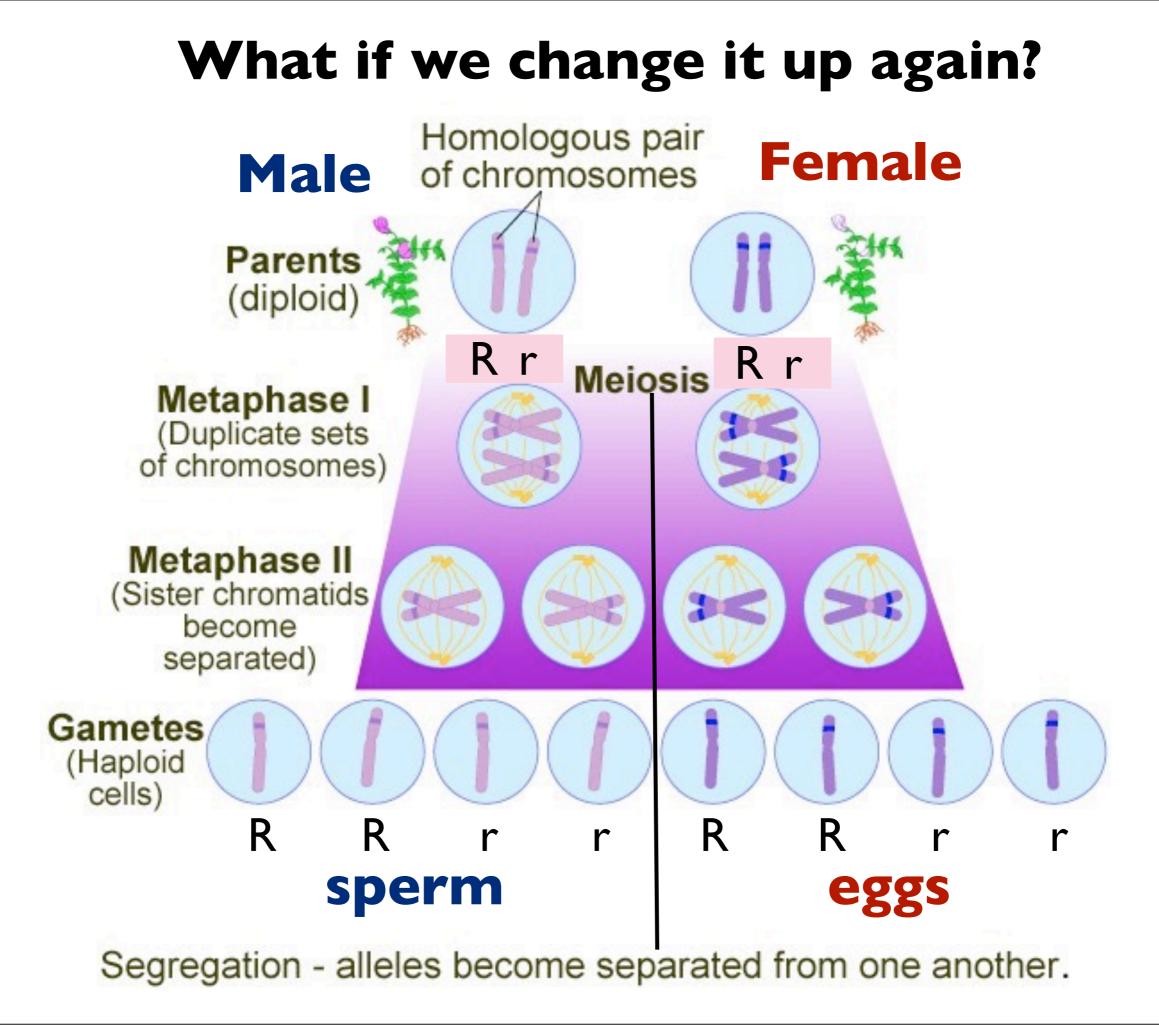
I.) What are the possible sperm? 2.) What are the possible eggs? 3.) What are the possible fertilizations? 4.) What are the possible offspring?



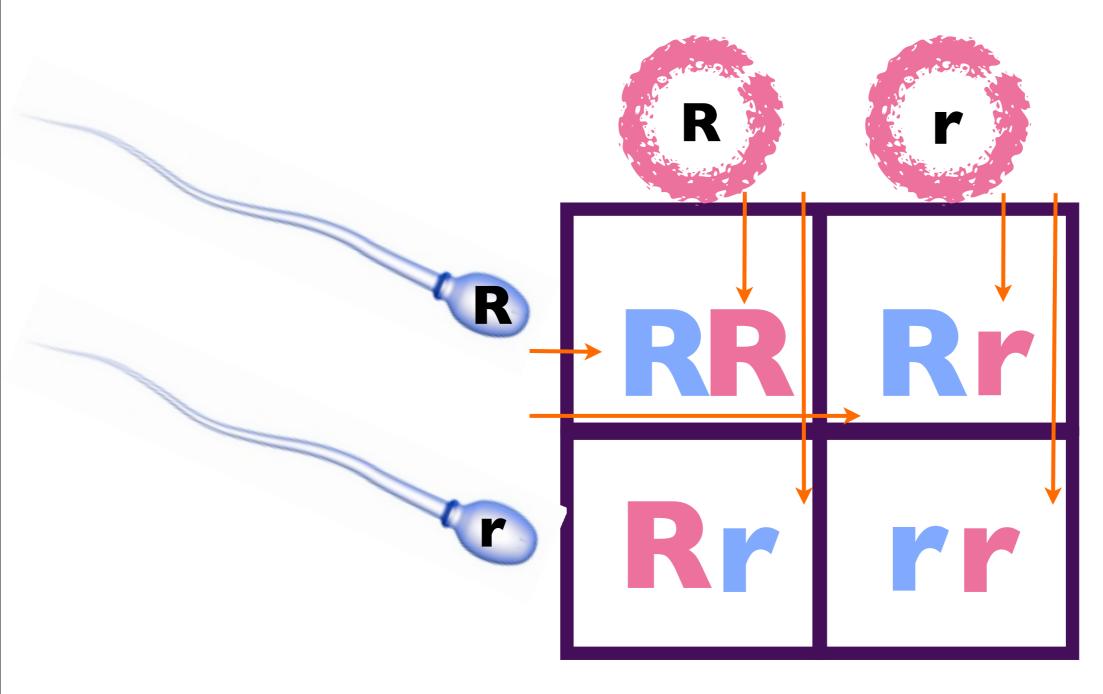


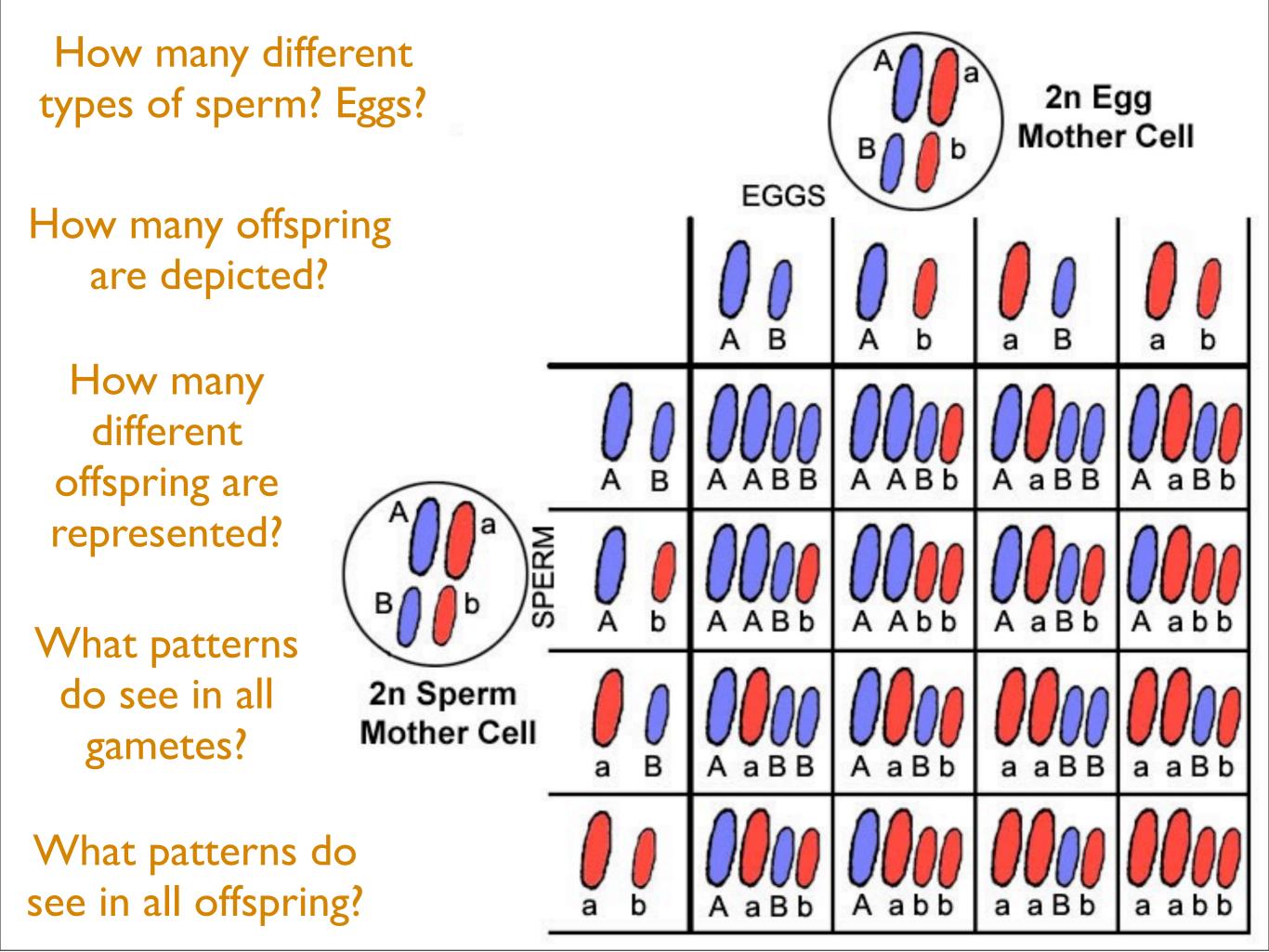
I.) What are the possible sperm? 2.) What are the possible eggs? 3.) What are the possible fertilizations? 4.) What are the possible offspring?





1.) What are the possible sperm? 2.) What are the possible eggs? 3.) What are the possible fertilizations? 4.) What are the possible offspring?





In Summary

- **Genetics** the study of *heredity* and hereditary variation.
- Heredity- the transmission of *traits* one generation to another.
- **Trait** one or more detectable variants in a genetic characteristic.
- **Characteristic** an observable feature that may vary among individuals.
- **Gene** a discrete unit of hereditary information consisting of a specific nucleotide sequence in DNA that is responsible for characteristics.
- **Chromosome** a cellular structure carrying genetic information (genes)

Transition to Genetics

BUT, here again is the key point! Chromosomes carry genes, genes control traits and genetics studies the transmission of these traits.

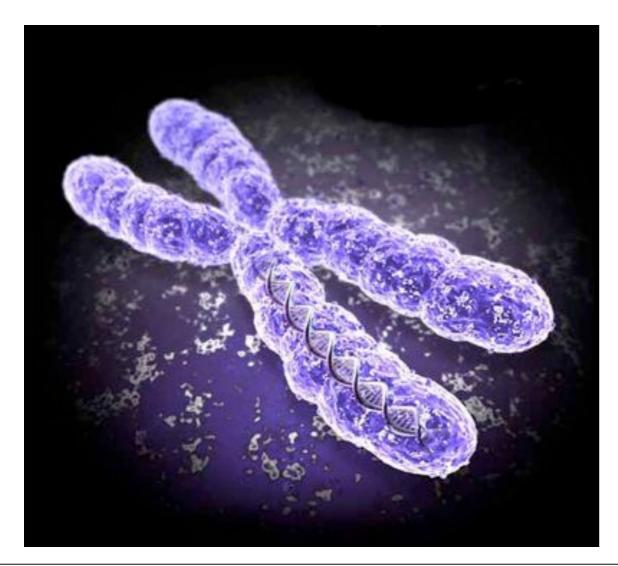
Understanding chromosomes transmission from one generation to another is an essential piece of knowledge in the "genetic puzzle"

It is amazing to think that our knowledge of genetics was born in a garden before we knew about chromosomes, genes and DNA.

Mendelian Genetics

II.

Main Idea: Laws of probability govern Mendel's laws of inheritance.





OK, I feel like we need to catch our breath and look at the big picture before we continue...lets review

- Genetics is the study of inherited traits.
- In a very general way genetics allows us to predict the pathway of traits into the future or allows us to track the pathway from which they came.
- Geneticists use punnet squares to look into the future and pedigrees to look into the past. (we will learn about pedigrees shortly)

- Punnet squares can be cumbersome and time consuming to use.
- As it turns out the Laws of Probability govern inheritance and thus we can use math to predict outcomes of future fusions between gametes.
- The Rule of Multiplication and the Rule of Addition are often less cumbersome and require far less time.
- Solving genetic problems mathematically will greatly increase your ability to quickly and correctly solve many of commonly asked questions in genetics.

Simple Probability

Probability of an event = occurring

the # of desired events

total # of events

52

Chance of flipping	=	I
"tails" on a coin.	_	2

Chance of picking an = "
"ace" in a deck of cards.

Chance of picking an |"ace of hearts" in a = $\frac{-1}{52}$ deck of cards.

Simple Probability Important lesson about probability!

Outcome of one event does not affect the outcome of second event when those outcomes are independent. The first toss of a coin has no effect on the second toss.

7

Chance of flipping "tails" on a coin.

Chance of flipping "tails" on a coin = for a second flip.

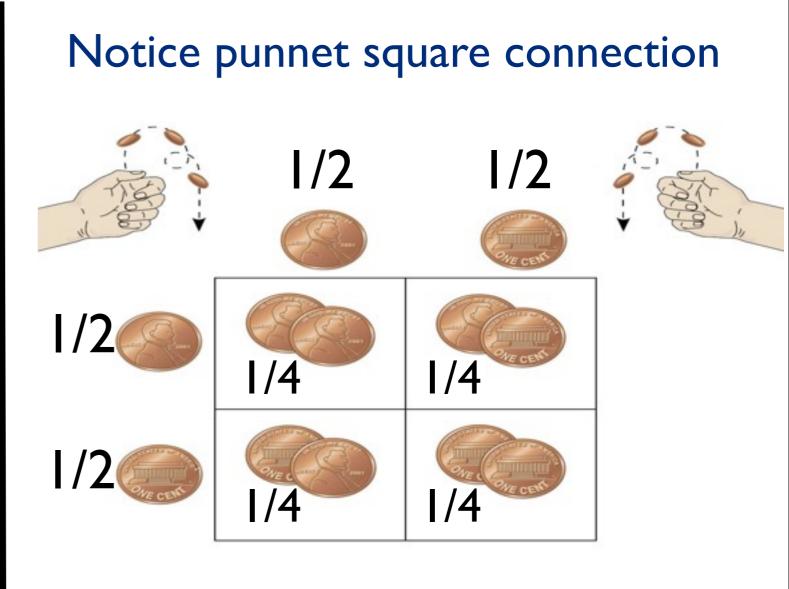
The segregation of alleles into gametes are also independent events, as we will see shortly.

The Rule of Multiplication

• The **Rule of Multiplication** states that to determine the probability of two or more independent events occurring together in some <u>specific combination</u>, we multiply the probability of one event by the probability of the other event.

Chance the coin lands on tails, on two consecutive flips.

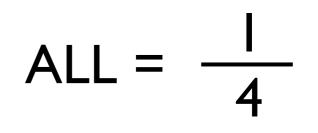
$$\frac{1}{2} \times \frac{1}{2} = \frac{1}{4}$$



The Rule of Multiplication

Notice...<u>If</u> the <u>order is specified</u> then you use the rule of multiplication.

Chance you flip tail, then another tail. Chance you flip tail, then a head. Chance you flip head, then a tail.



- Chance you flip head, then another head.
- In all four cases above the order is specified thus (1/2)(1/2)=1/4.

Chance you flip one tail and one head.

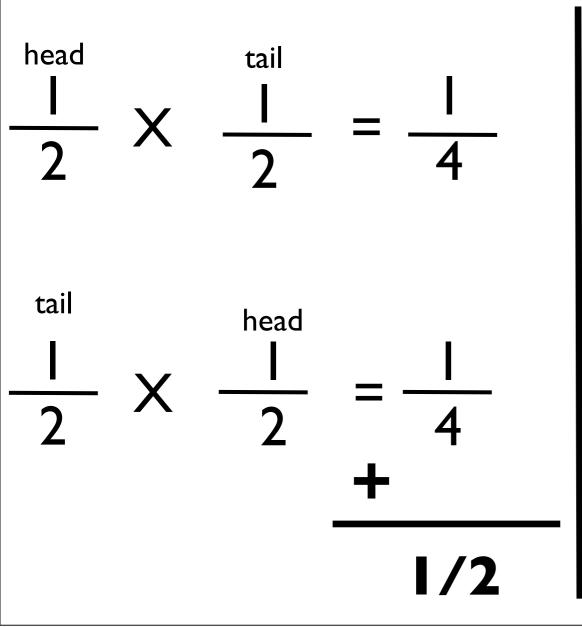
• Notice...The **order is NOT specified.** Now we need to use another rule along with the rule of multiplication.

... The Rule of Addition

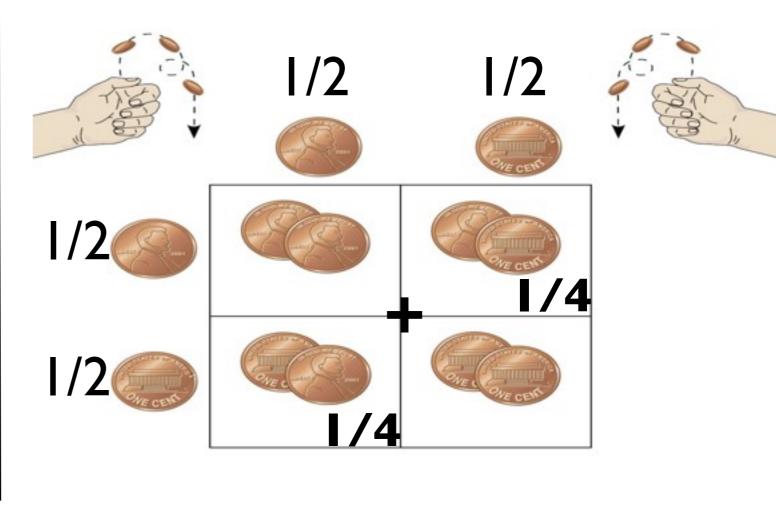
The Rule of Addition

• The **Rule of Addition** states that to determine the probability of two or more mutually exclusive events occurring together is calculated by adding their individual probabilities.

Chance you flip one tail and one head.



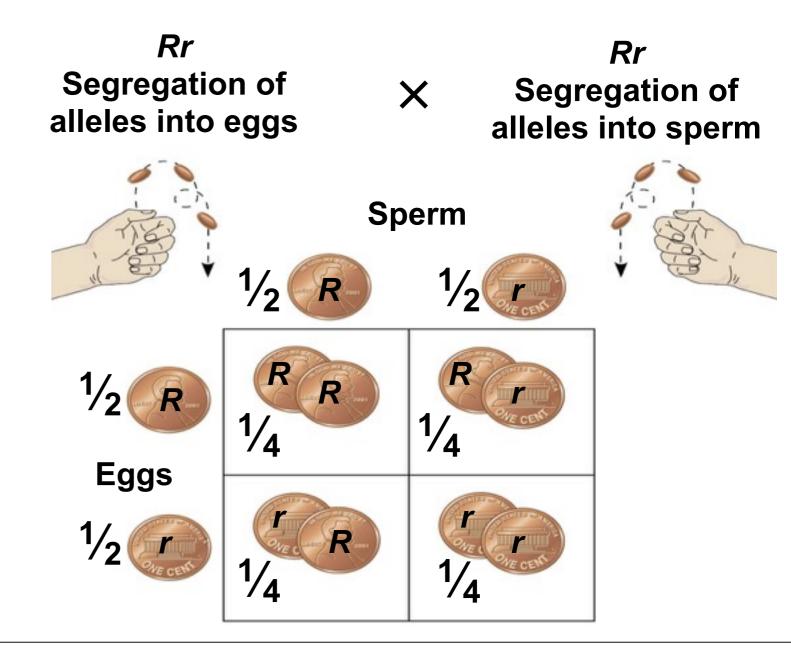
Notice punnet square connection



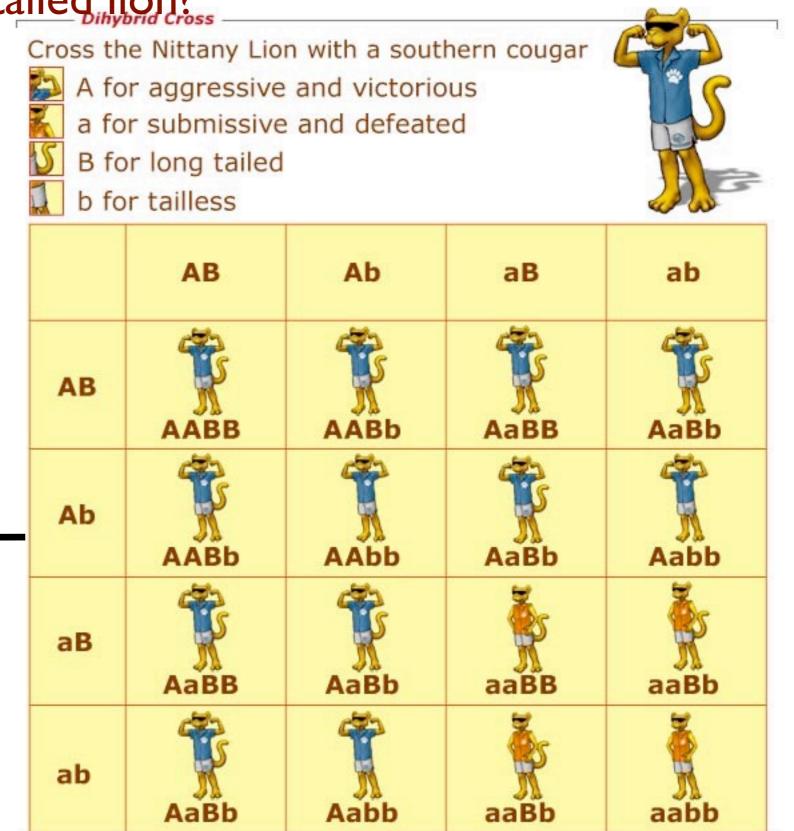
Math Applied to Genetics

Chance of homozygous recessive. (1/2)(1/2) = 1/4

- Chance of homozygous dominant. (1/2)(1/2) = 1/4
- Chance of heterozygous. (1/2)(1/2) + (1/2)(1/2) = 1/2

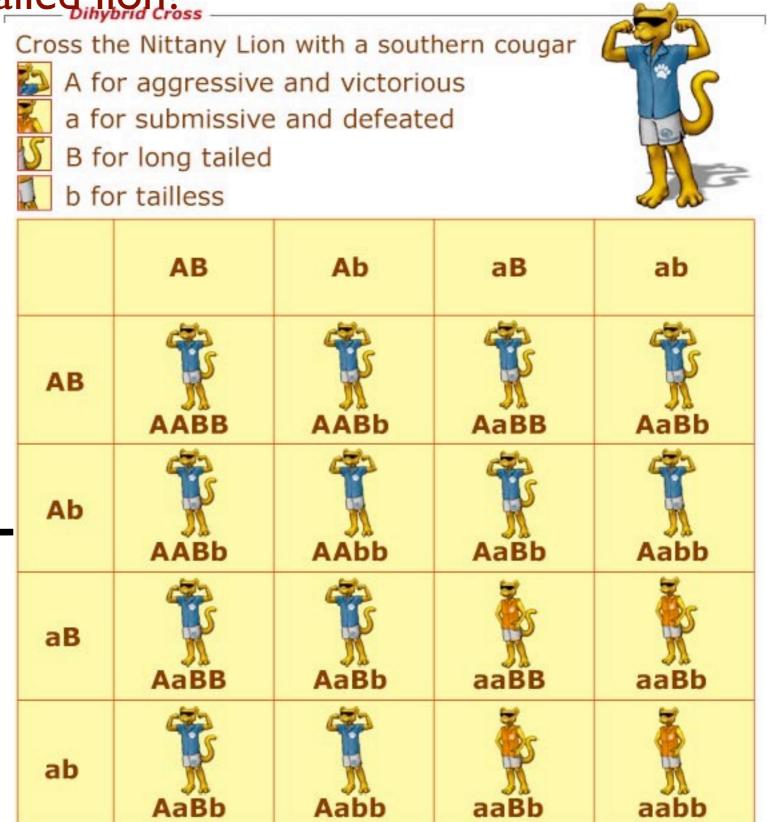


What is the probability that these parents produce a submissive long tailed lion?



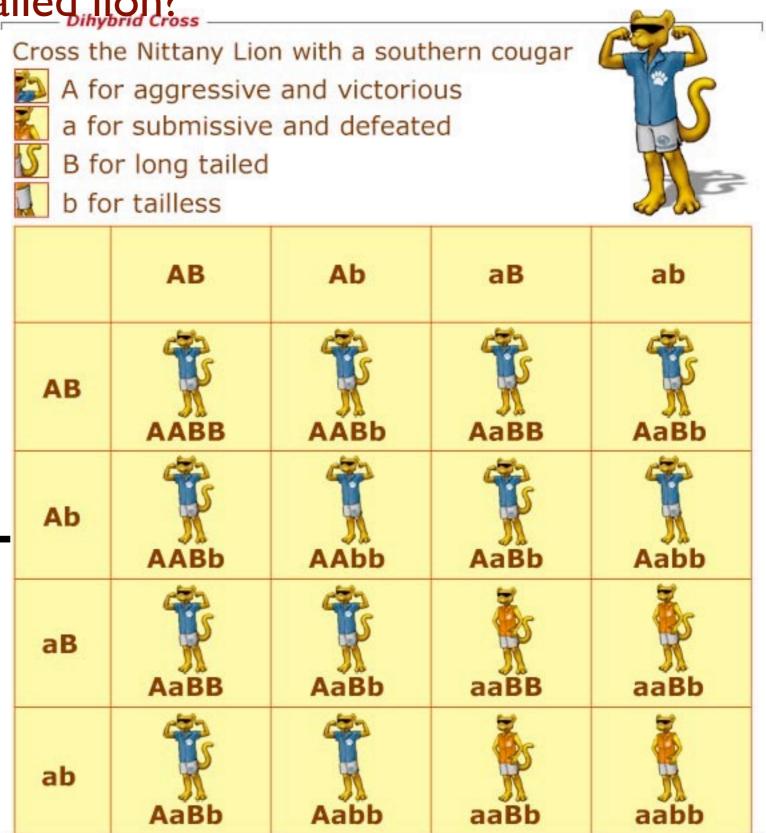
What is the probability that these parents produce a submissive long tailed lion?

Start by writing out the genotypes you "desire"



What is the probability that these parents produce a submissive long tailed lion?

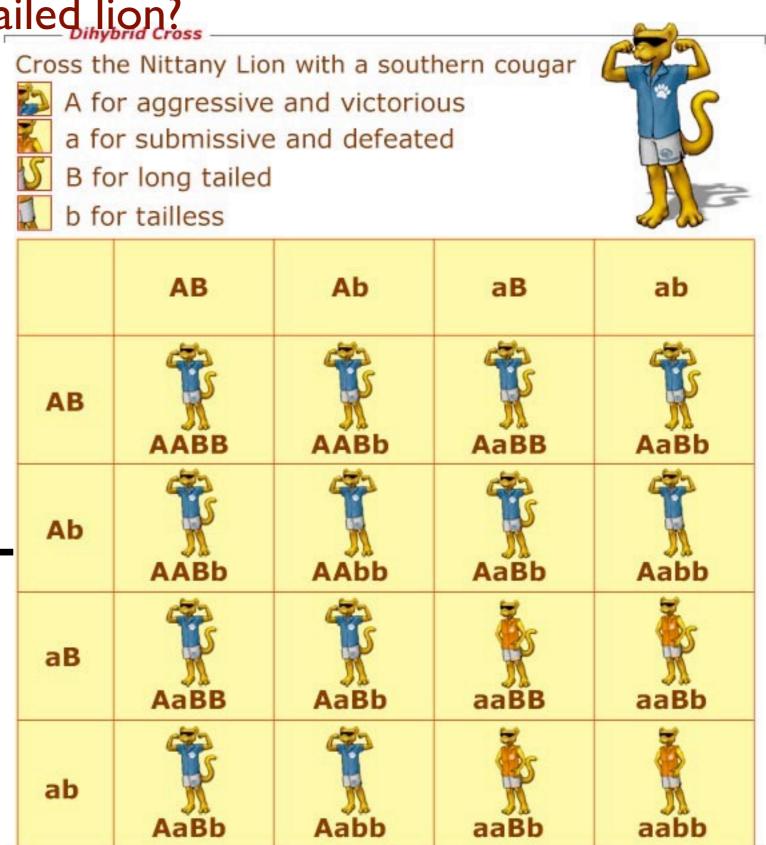
Start by writing out the genotypes you "desire" aaBB



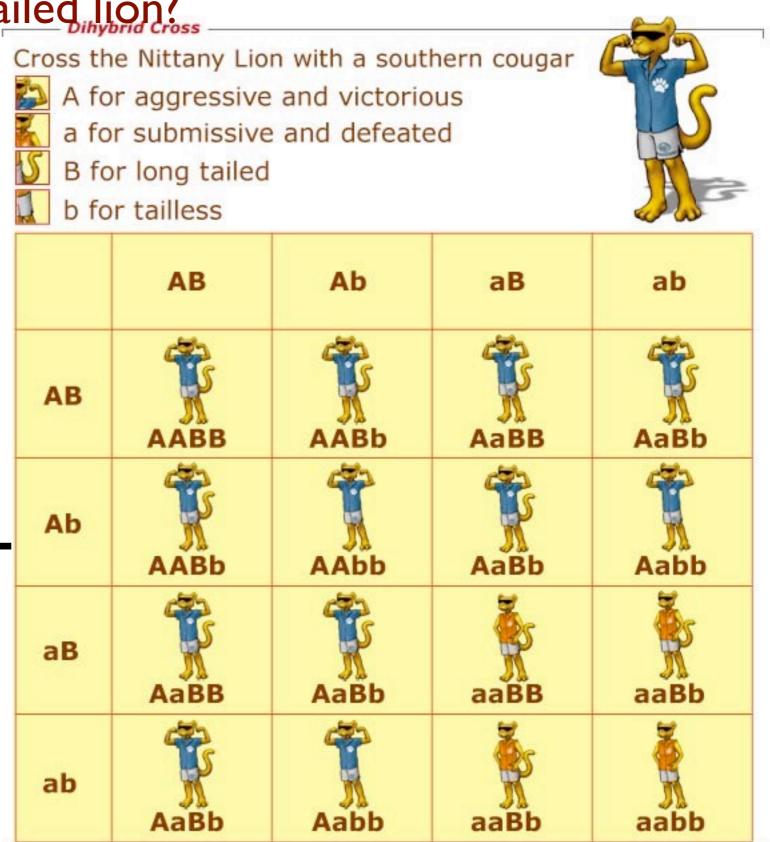
What is the probability that these parents produce a submissive long tailed lion?

Start by writing out the genotypes you "desire" aaBB

aaBb

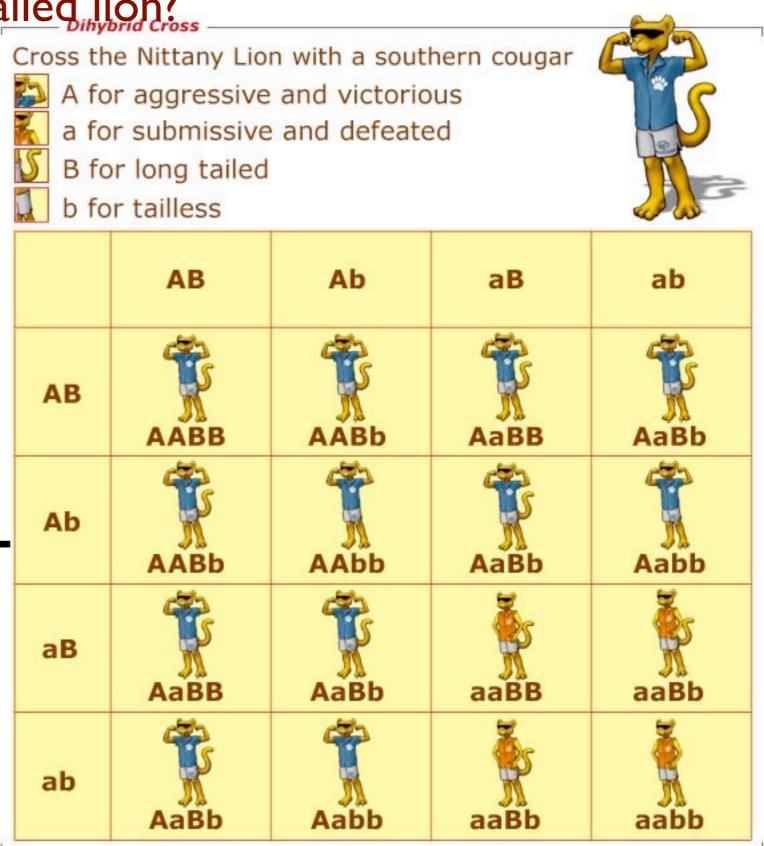


- What is the probability that these parents produce a submissive long tailed lion?
- Start by writing out the genotypes you "desire" **aaBB**
- (1/2)(1/2)(1/2)(1/2)=1/16



- What is the probability that these parents produce a submissive long tailed lion?
- Start by writing out the genotypes you "desire" aaBB
- (1/2)(1/2)(1/2)(1/2)=1/16

```
(1/2)(1/2)(1/2)(1/2)(2)=2/16
```

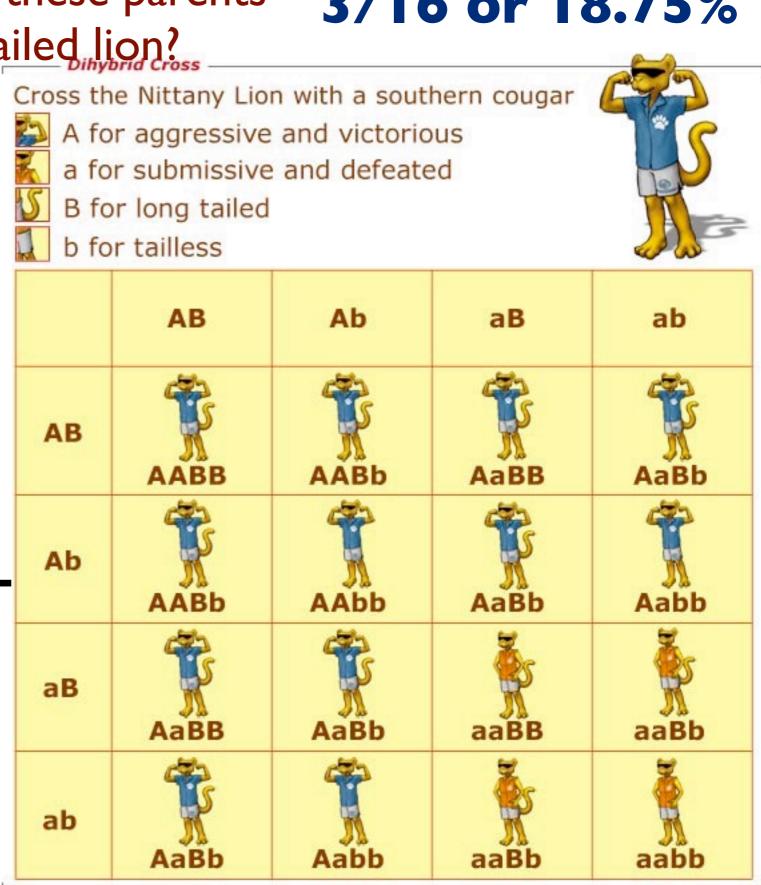


What is the probability that these parents 3/16 or 18.75% produce a submissive long tailed lion?

Start by writing out the genotypes you "desire" aaBB

(1/2)(1/2)(1/2)(1/2)=1/16

```
(1/2)(1/2)(1/2)(1/2)(2)=2/16
```

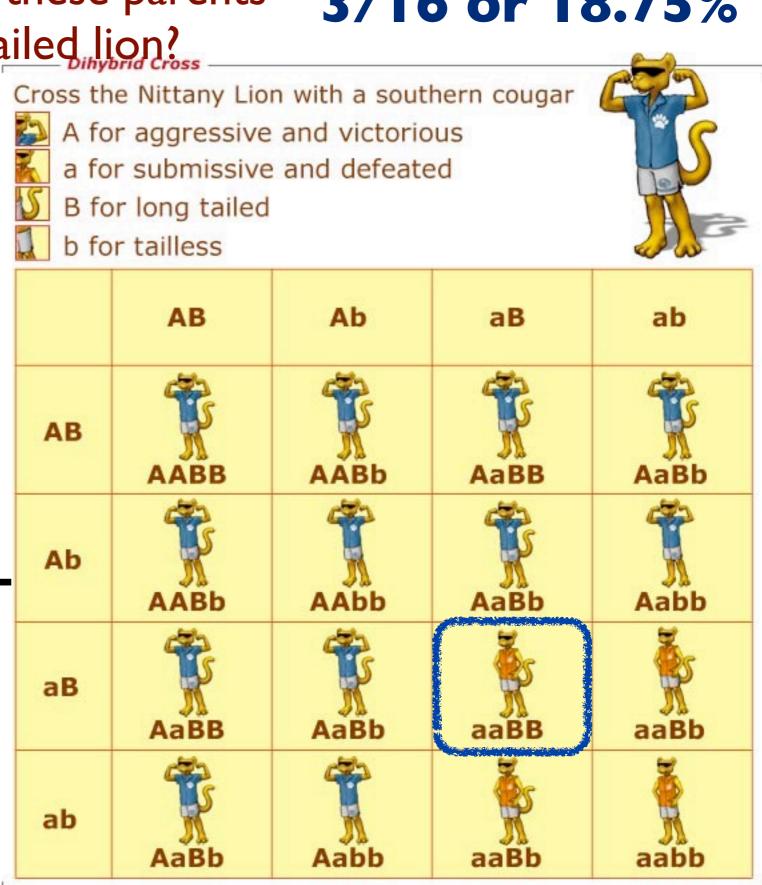


What is the probability that these parents 3/16 or 18.75% produce a submissive long tailed lion?

Start by writing out the genotypes you "desire" aaBB

(1/2)(1/2)(1/2)(1/2)=1/16

```
(1/2)(1/2)(1/2)(1/2)(2)=2/16
```

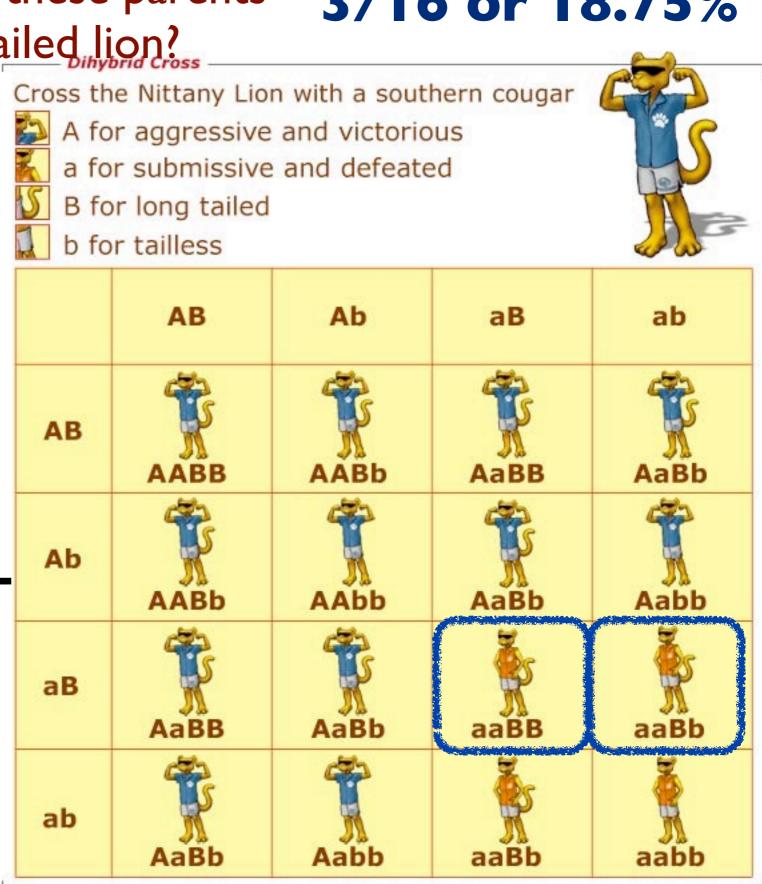


What is the probability that these parents 3/16 or 18.75% produce a submissive long tailed lion?

Start by writing out the genotypes you "desire" aaBB

(1/2)(1/2)(1/2)(1/2)=1/16

```
(1/2)(1/2)(1/2)(1/2)(2)=2/16
```

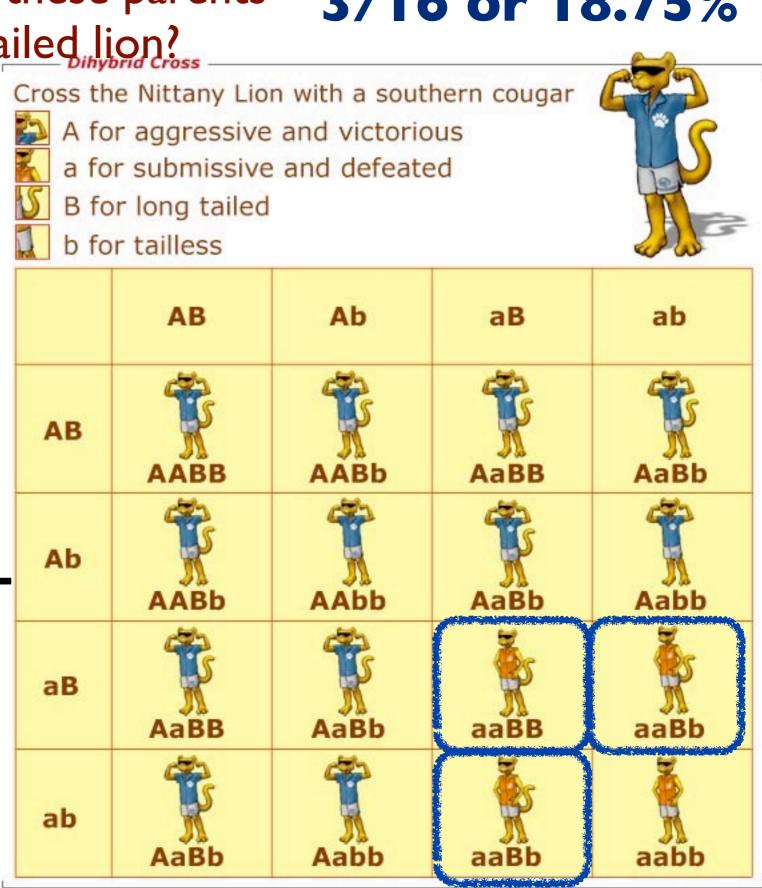


What is the probability that these parents 3/16 or 18.75% produce a submissive long tailed lion?

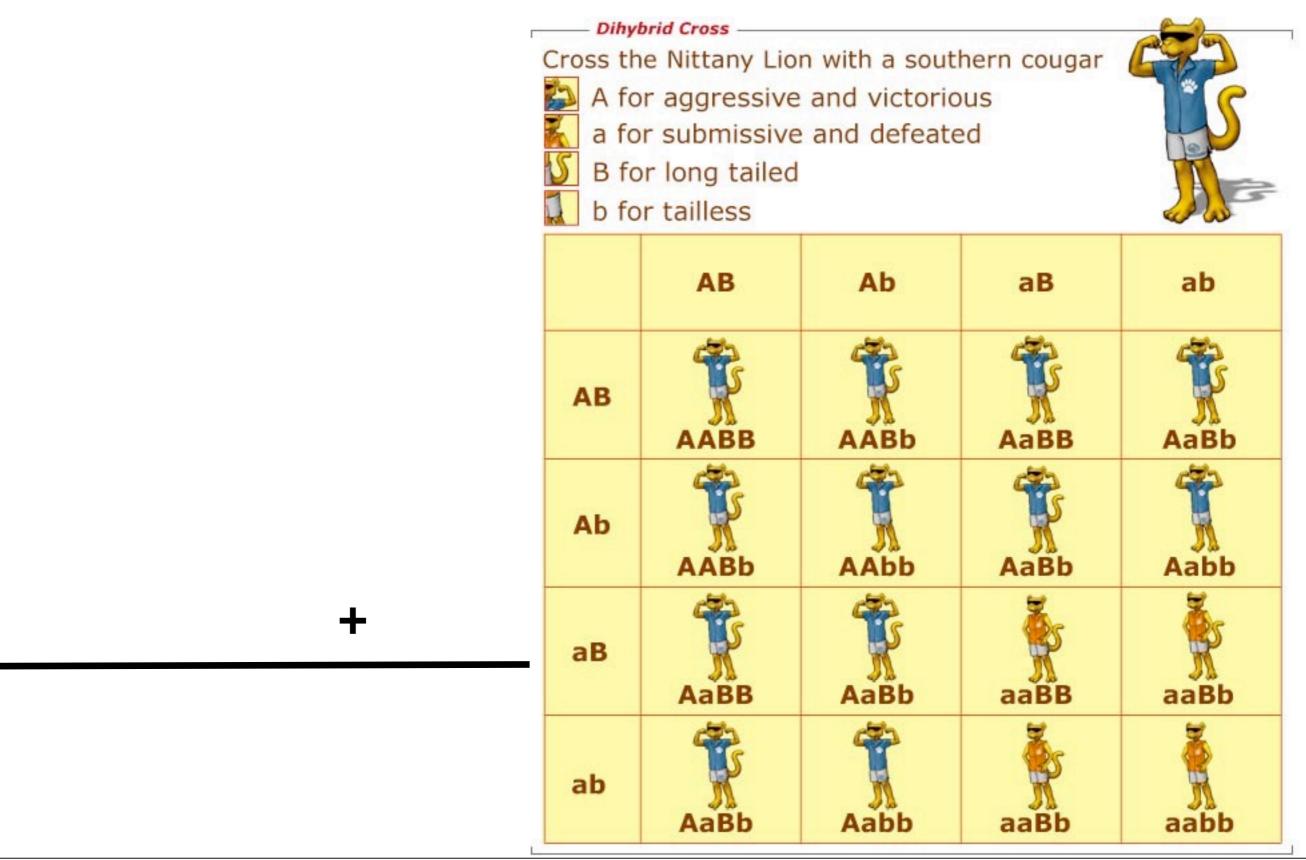
Start by writing out the genotypes you "desire" aaBB

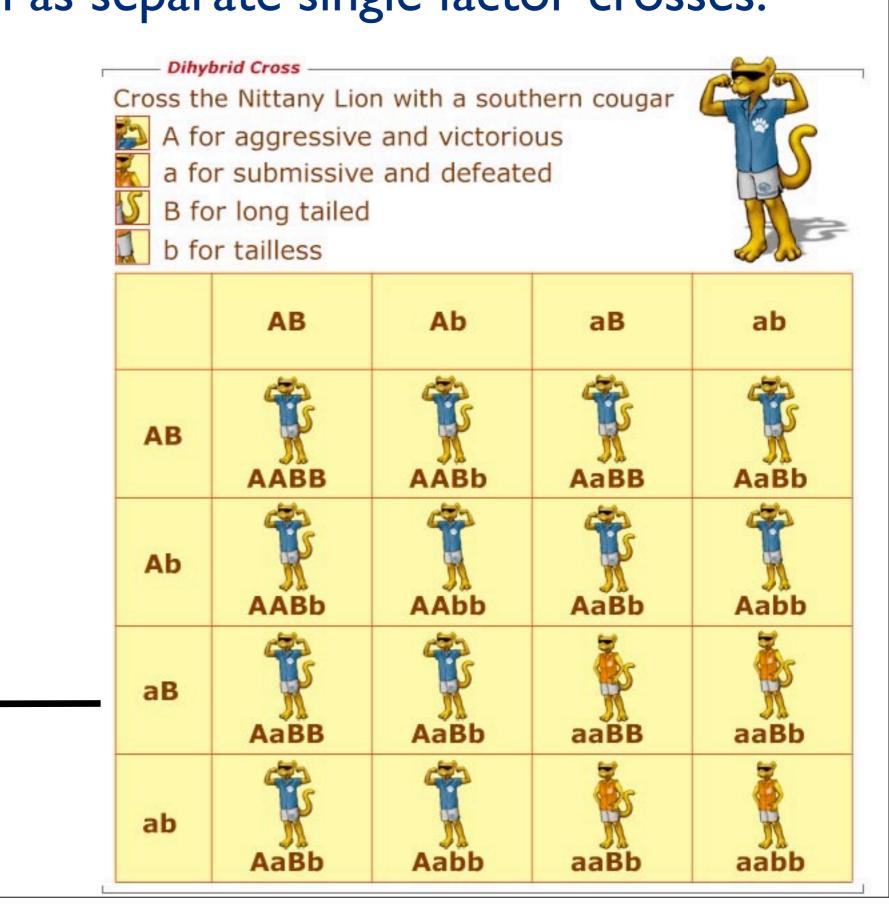
(1/2)(1/2)(1/2)(1/2)=1/16

```
(1/2)(1/2)(1/2)(1/2)(2)=2/16
```

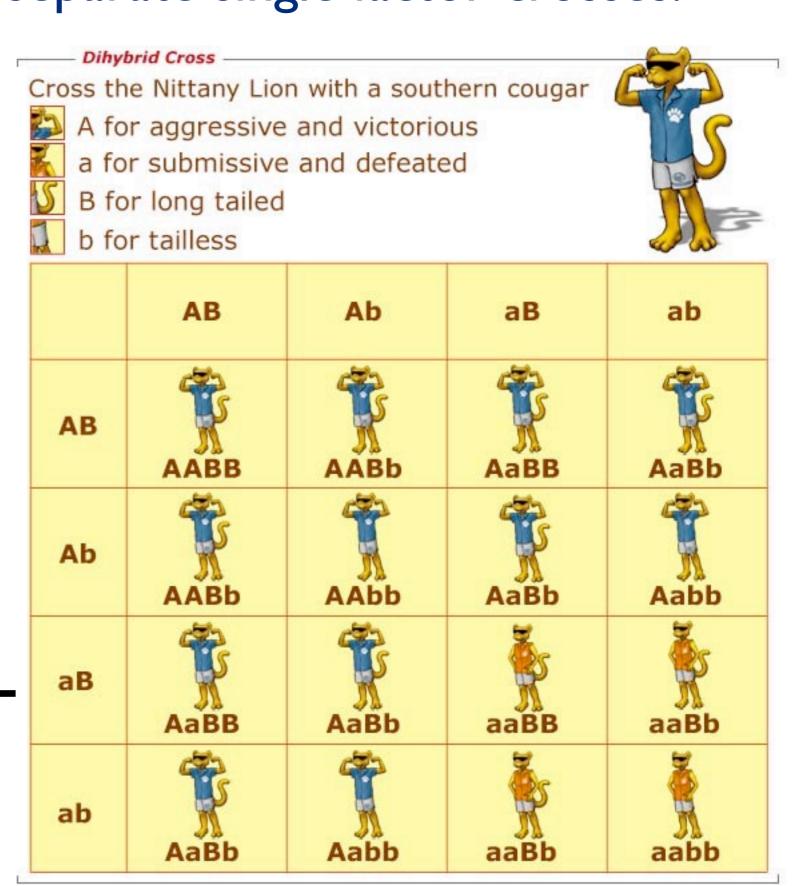


Same problem solved differently



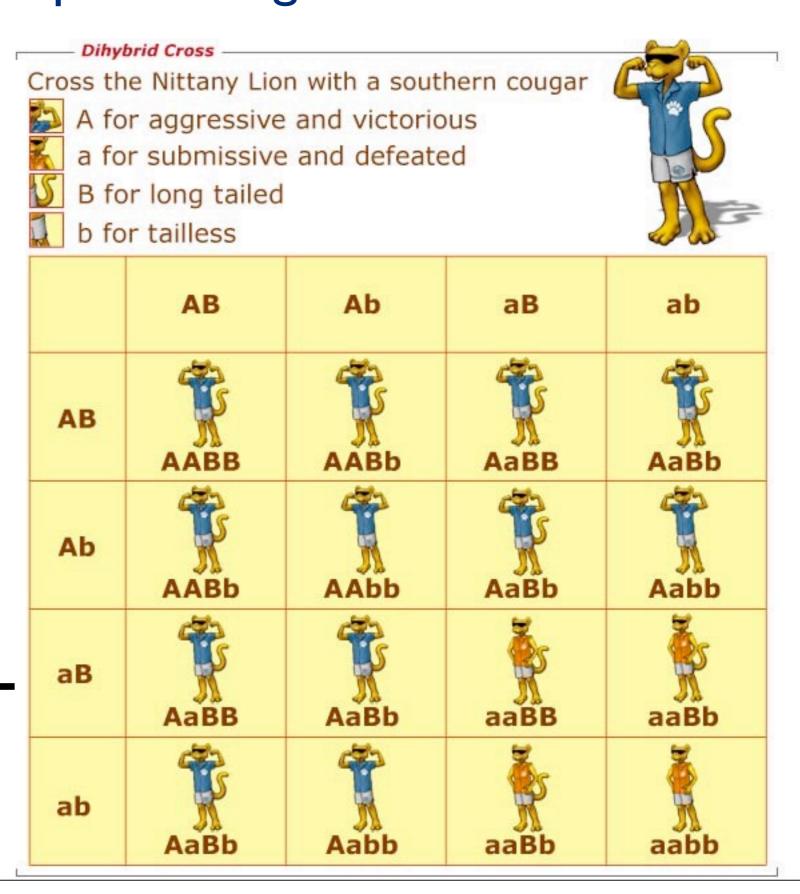


Aa x Aa



Aa x Aa

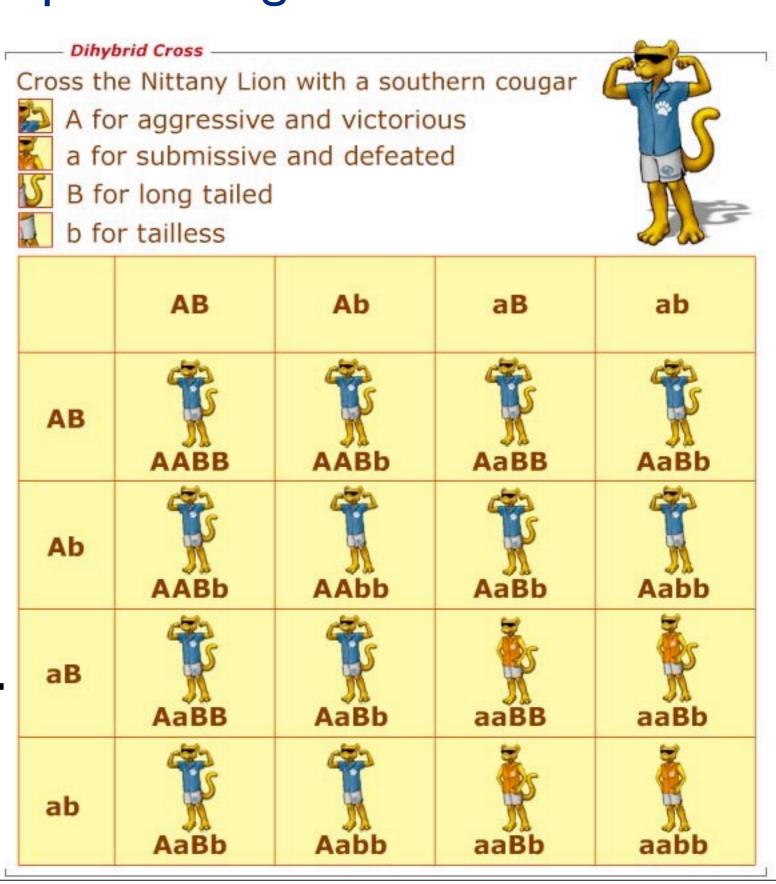
chance of aa = (1/2)(1/2)=1/4



Aa x Aa

chance of aa = (1/2)(1/2)=1/4

Bb x Bb

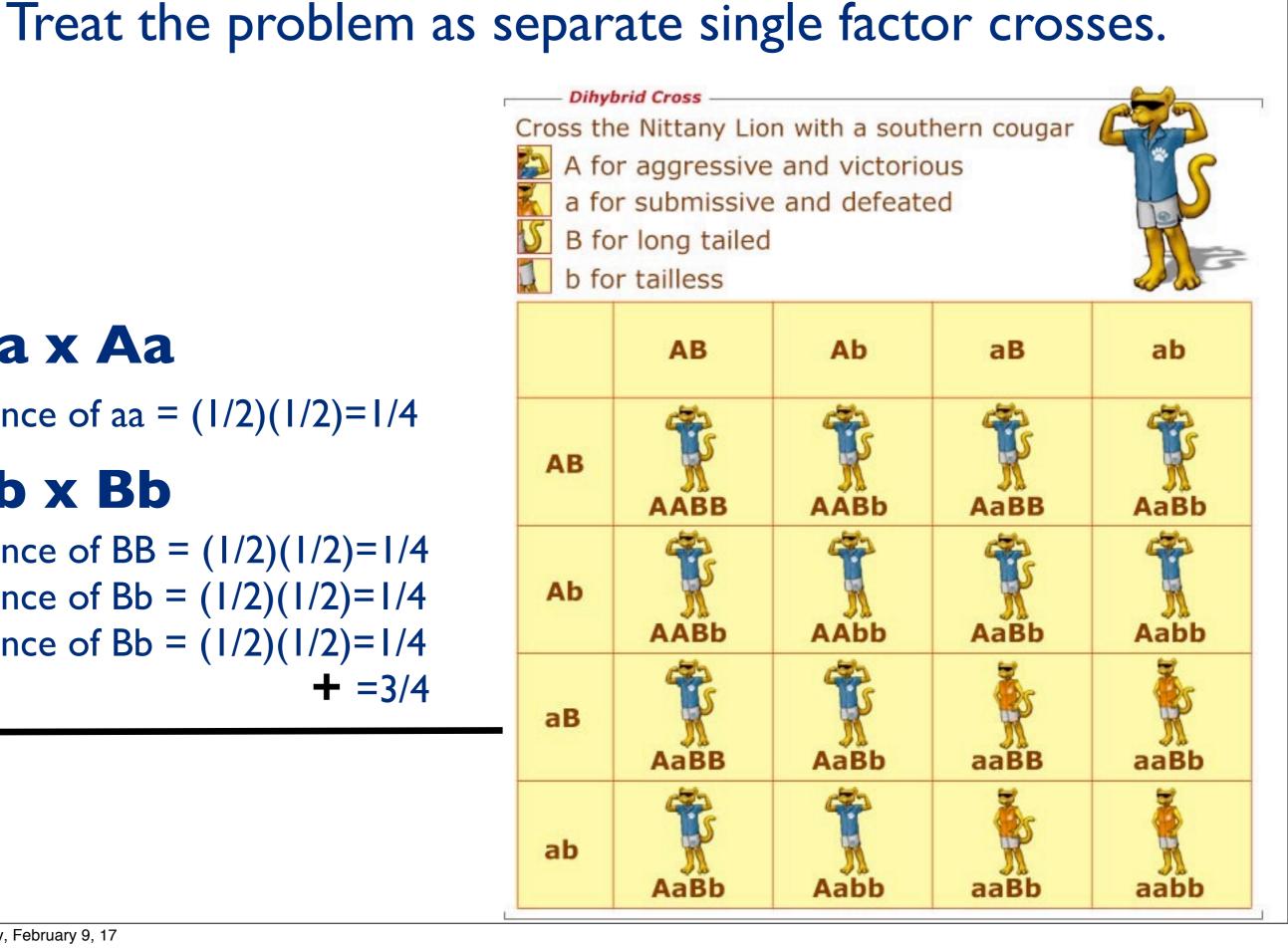


Aa x Aa

chance of aa = (1/2)(1/2)=1/4

Bb x Bb

chance of BB = (1/2)(1/2)=1/4chance of Bb = (1/2)(1/2)=1/4chance of Bb = (1/2)(1/2)=1/4+ = 3/4



Same problem solved differently

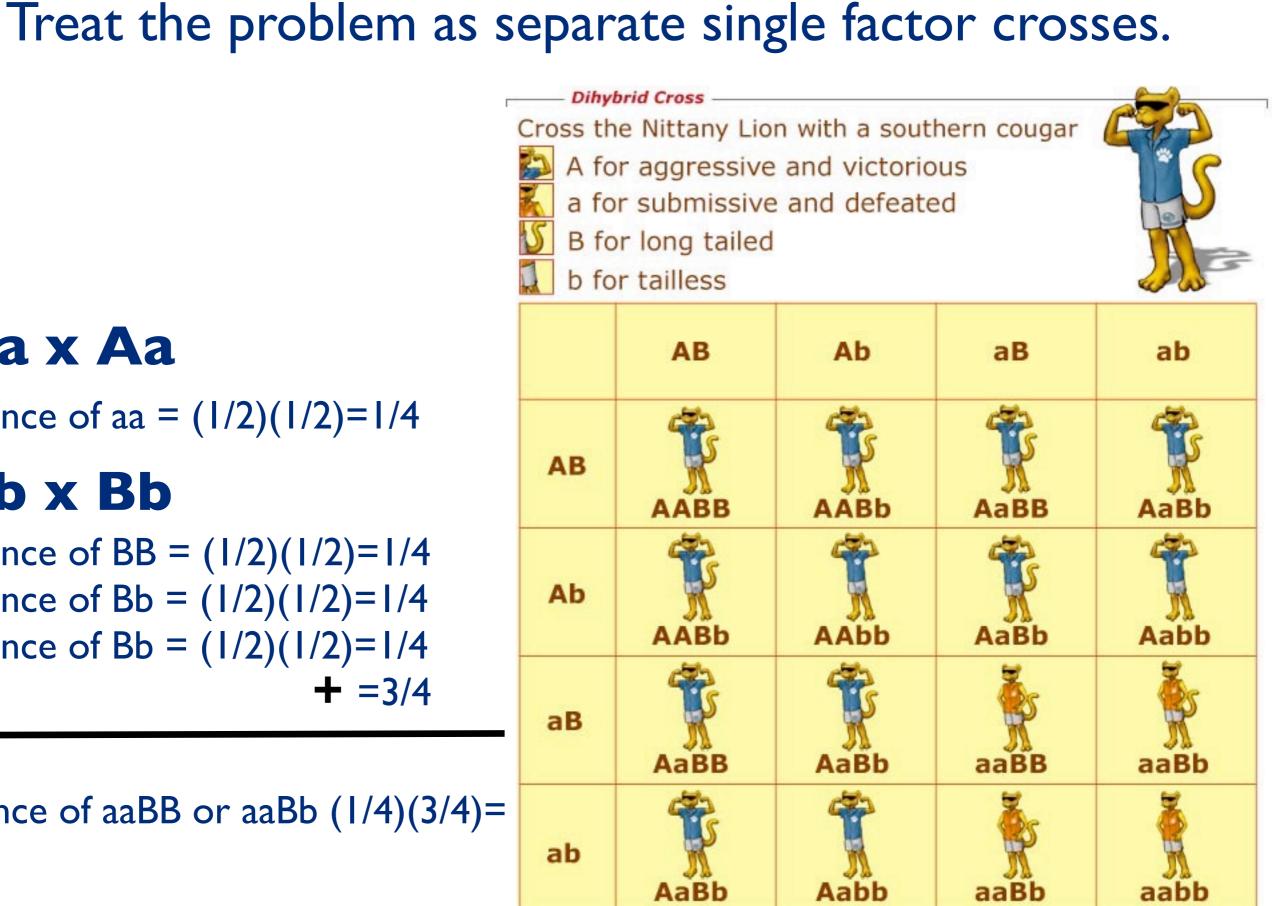
Aa x Aa

chance of aa = (1/2)(1/2)=1/4

Bb x Bb

chance of BB = (1/2)(1/2)=1/4chance of Bb = (1/2)(1/2)=1/4chance of Bb = (1/2)(1/2)=1/4+ = 3/4

chance of aaBB or aaBb (1/4)(3/4)=



Same problem solved differently

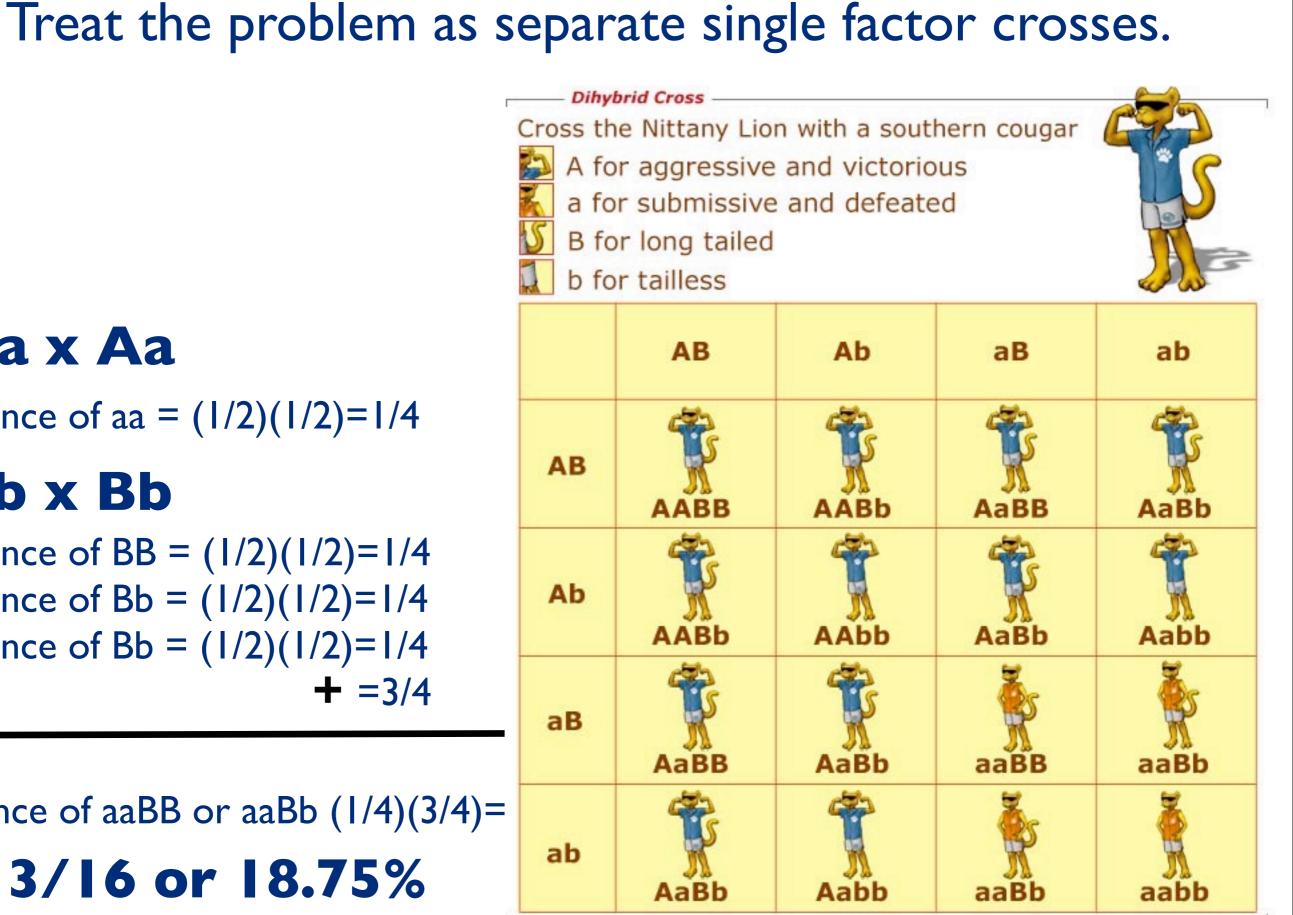
Aa x Aa

chance of aa = (1/2)(1/2)=1/4

Bb x Bb

chance of BB = (1/2)(1/2)=1/4chance of Bb = (1/2)(1/2)=1/4chance of Bb = (1/2)(1/2)=1/4+=3/4

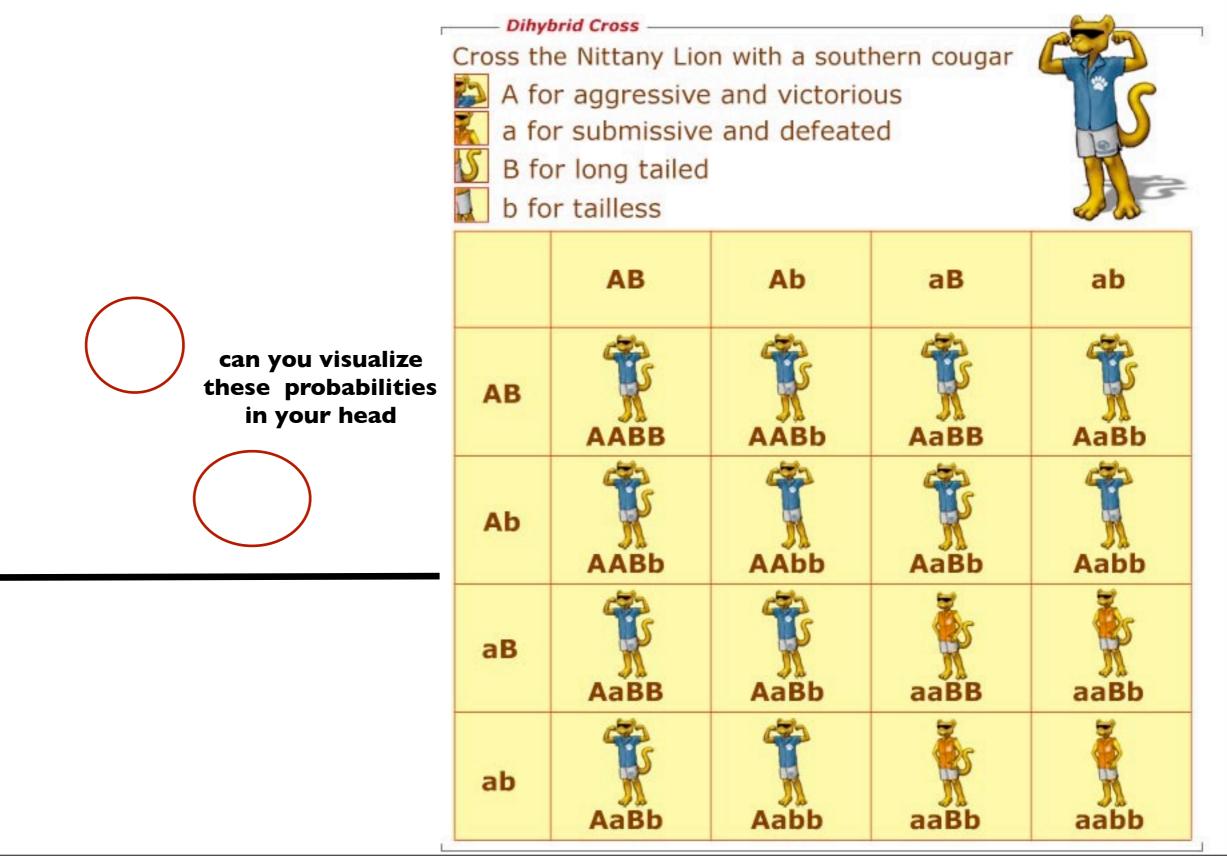
chance of aaBB or aaBb (1/4)(3/4)=3/16 or 18.75%

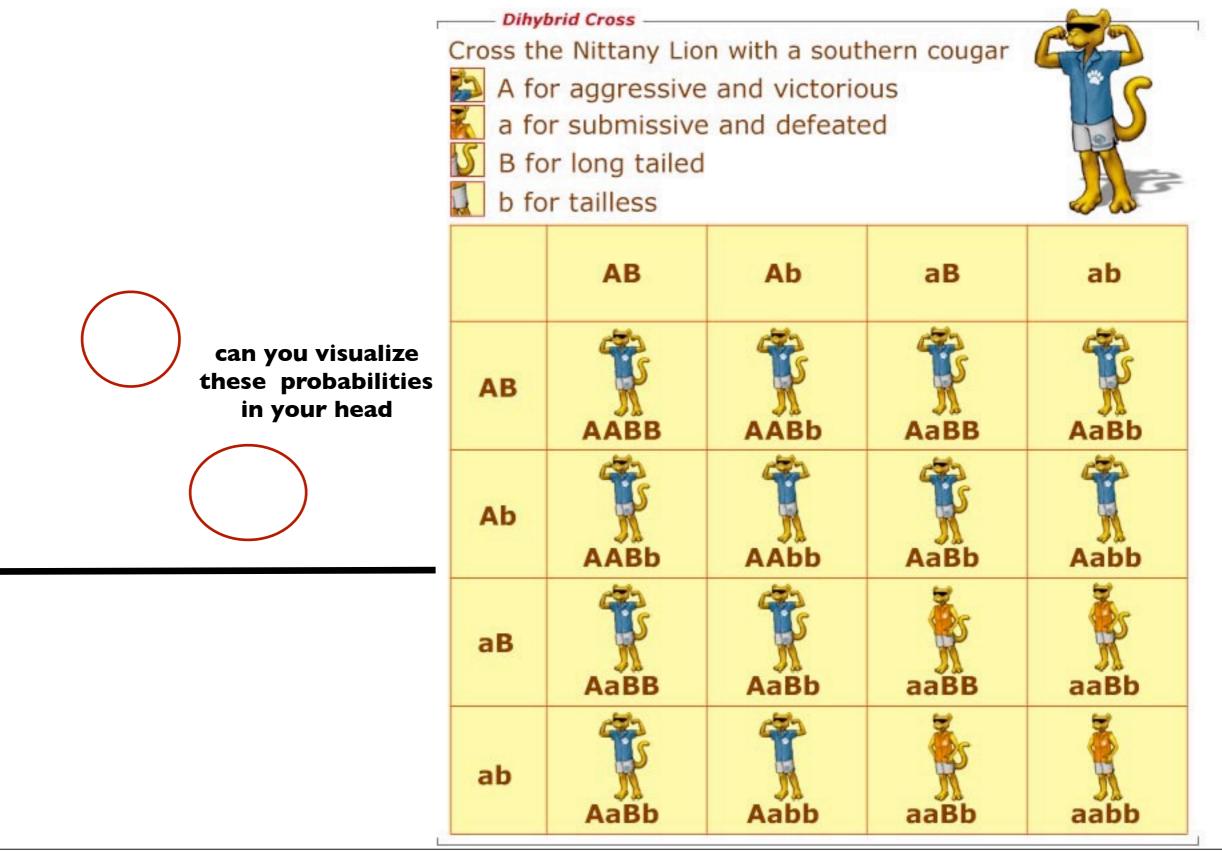


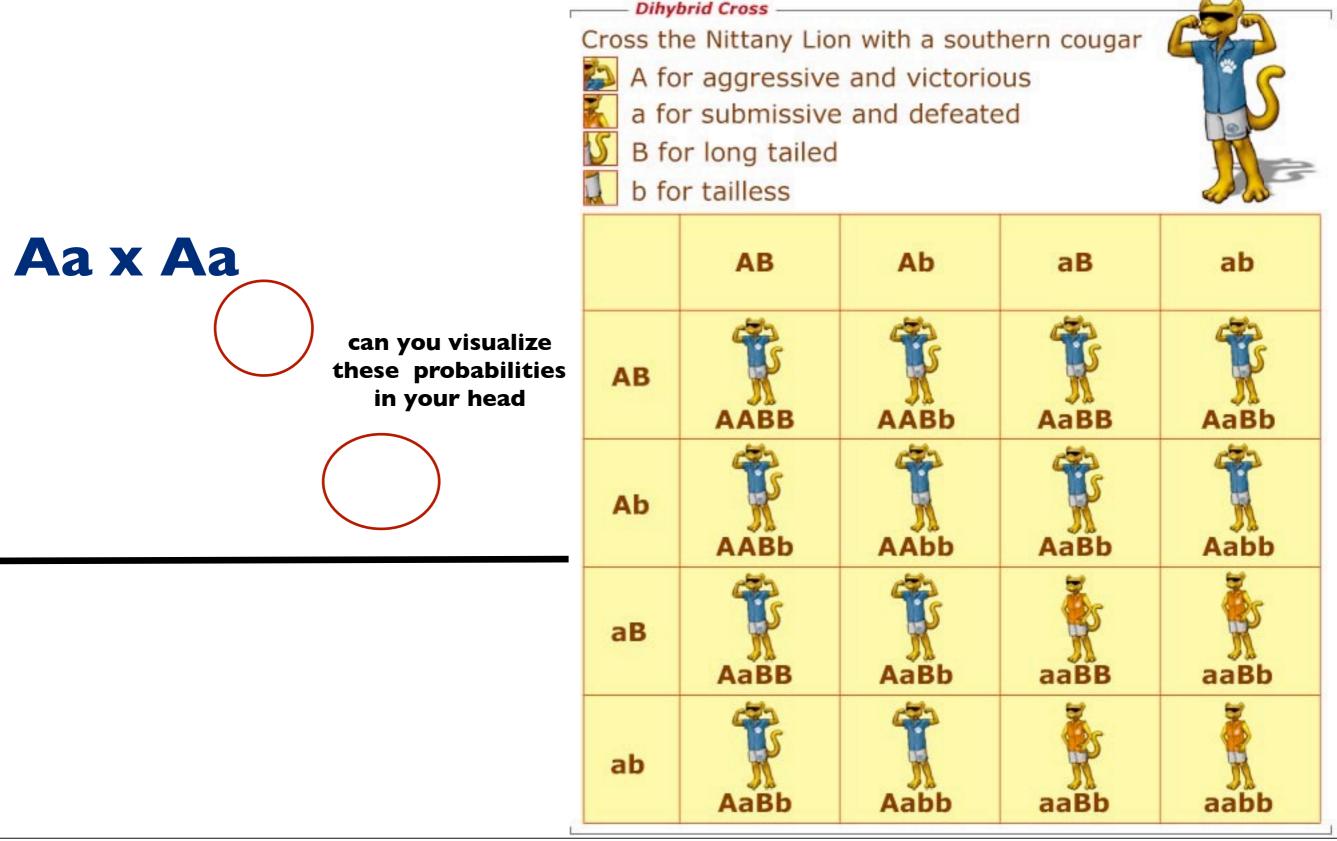
Same problem solved differently

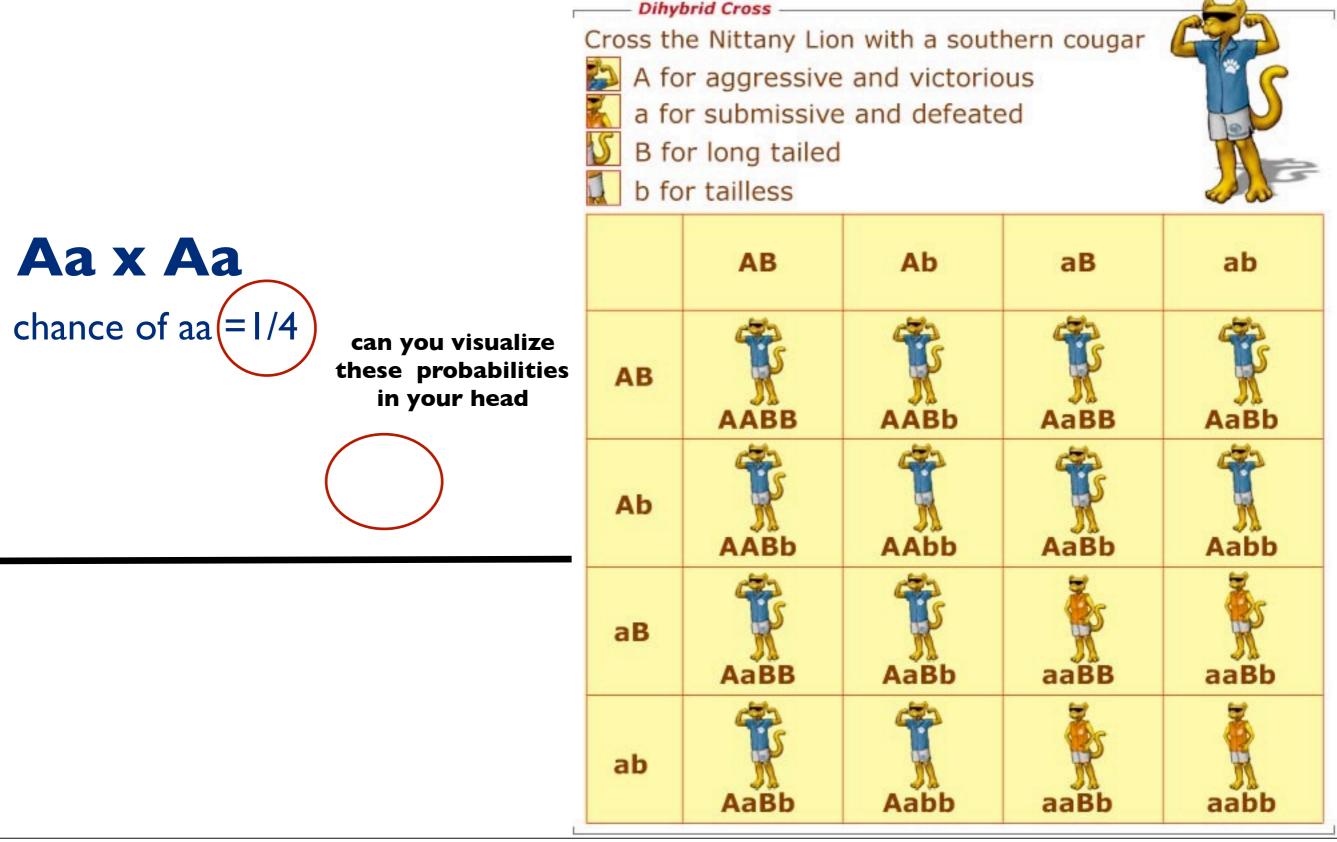
Thursday, February 9, 17

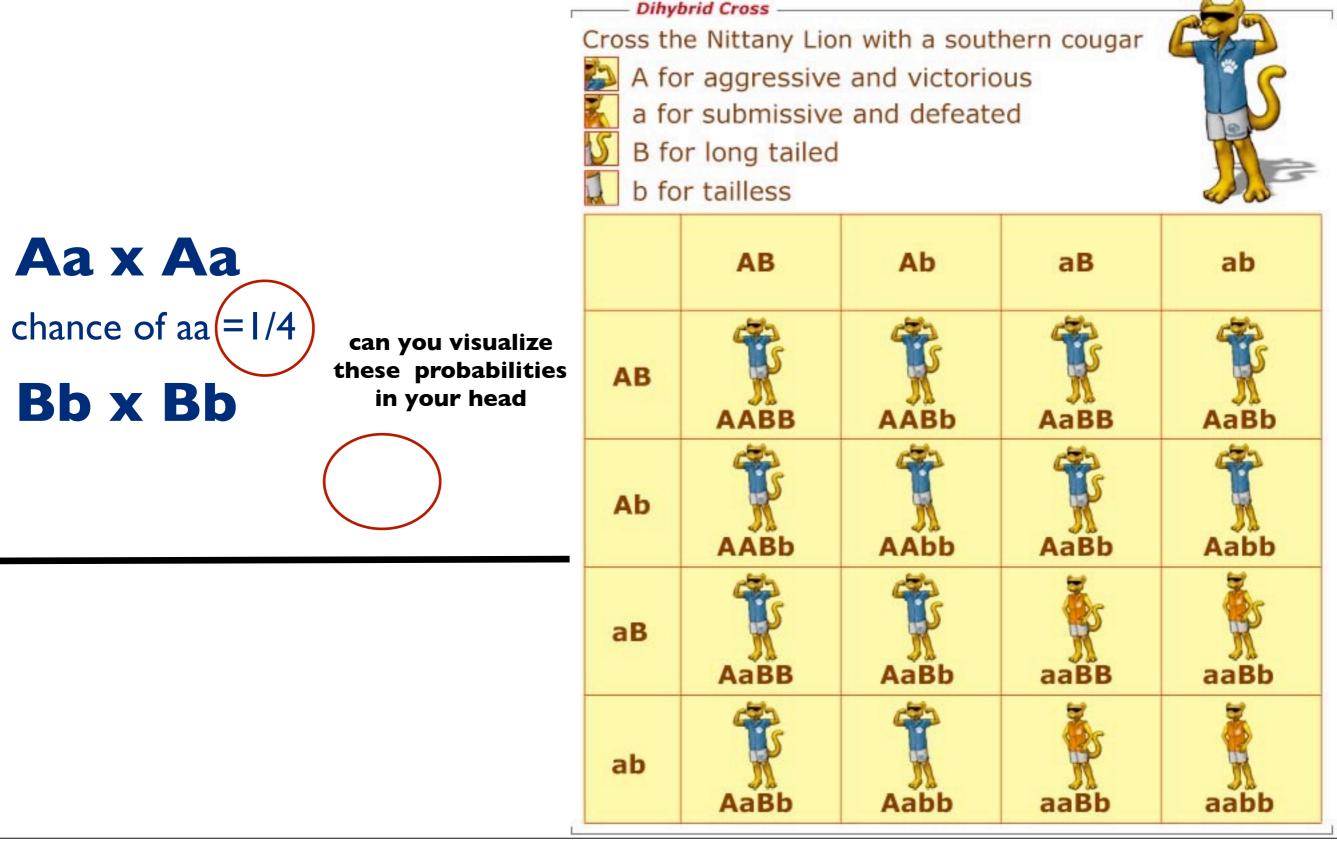
Same problem solved differently... yet again

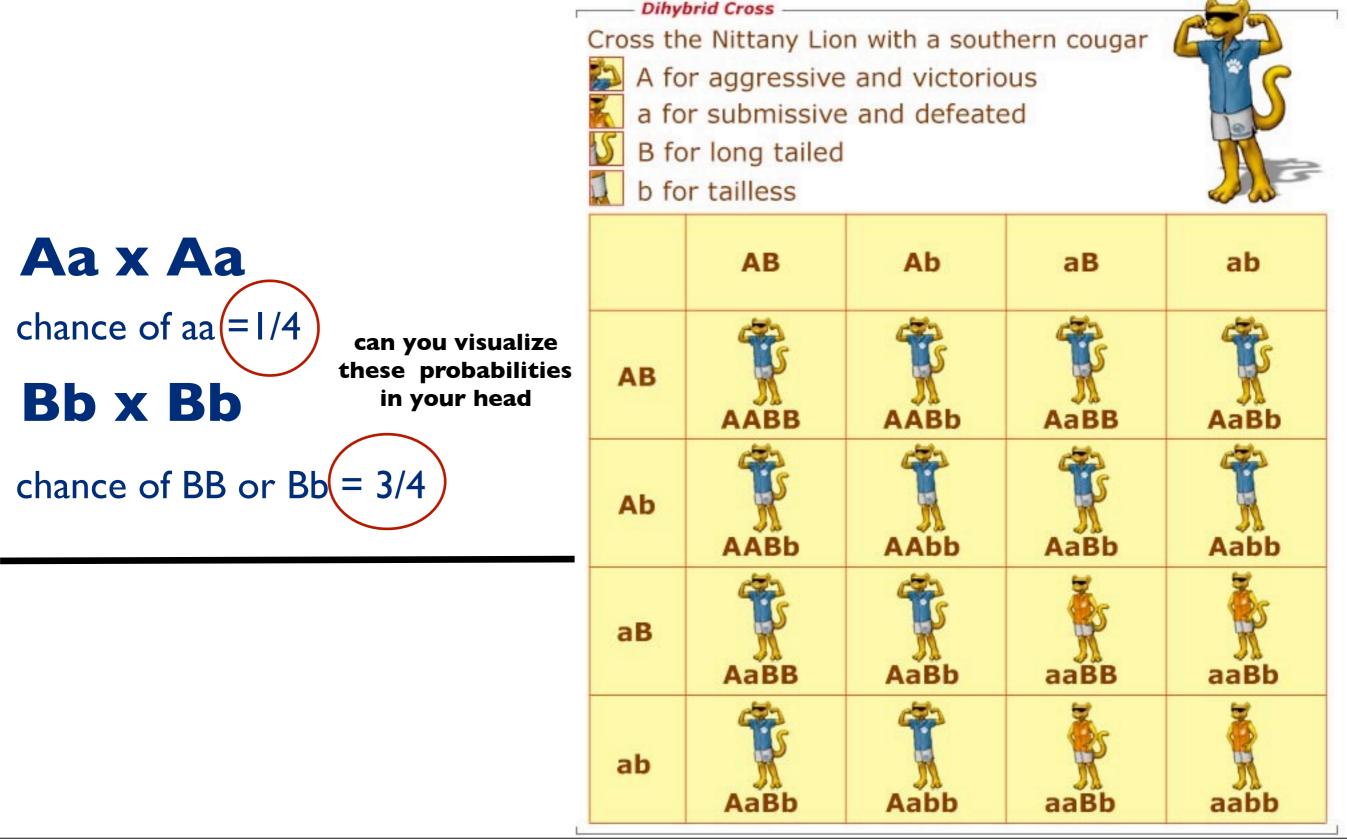


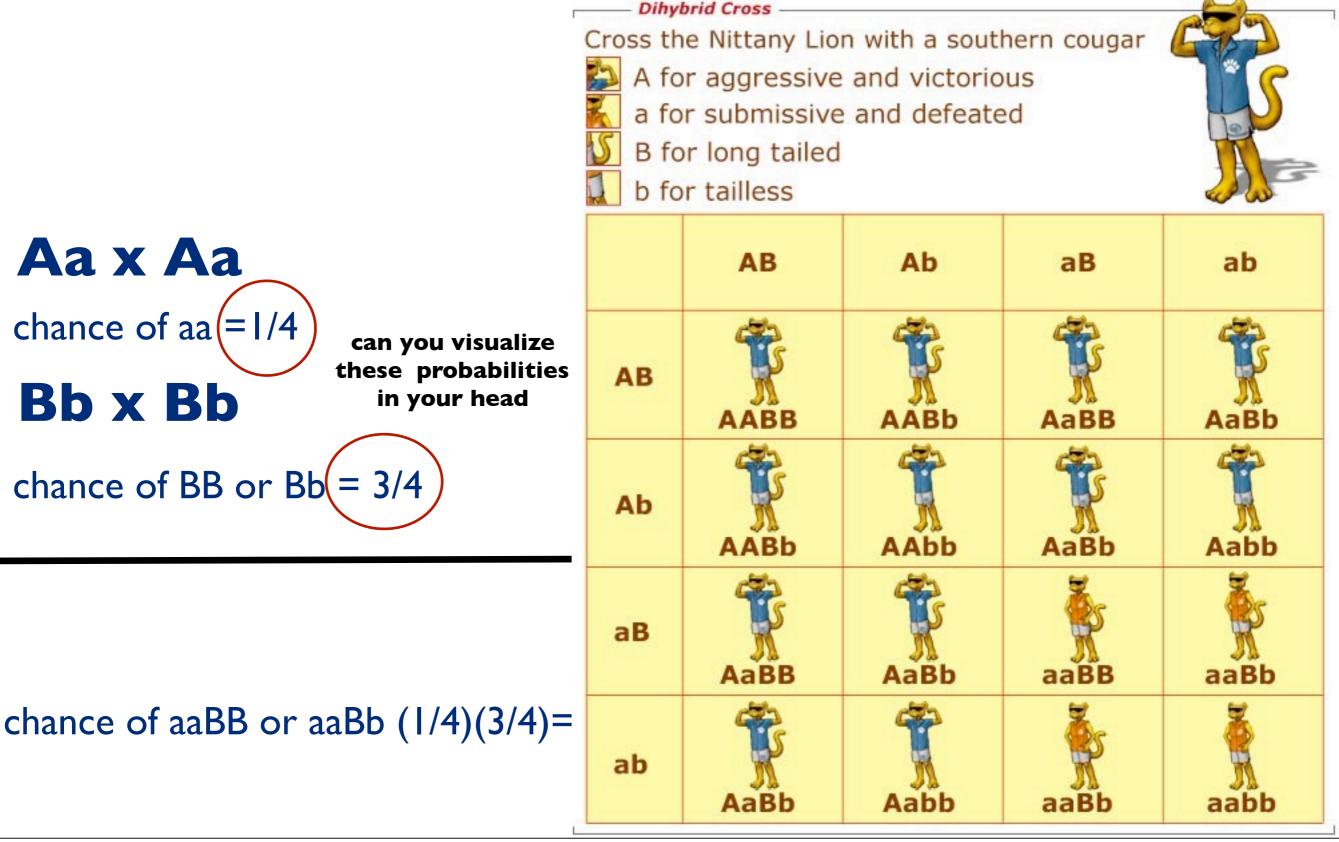


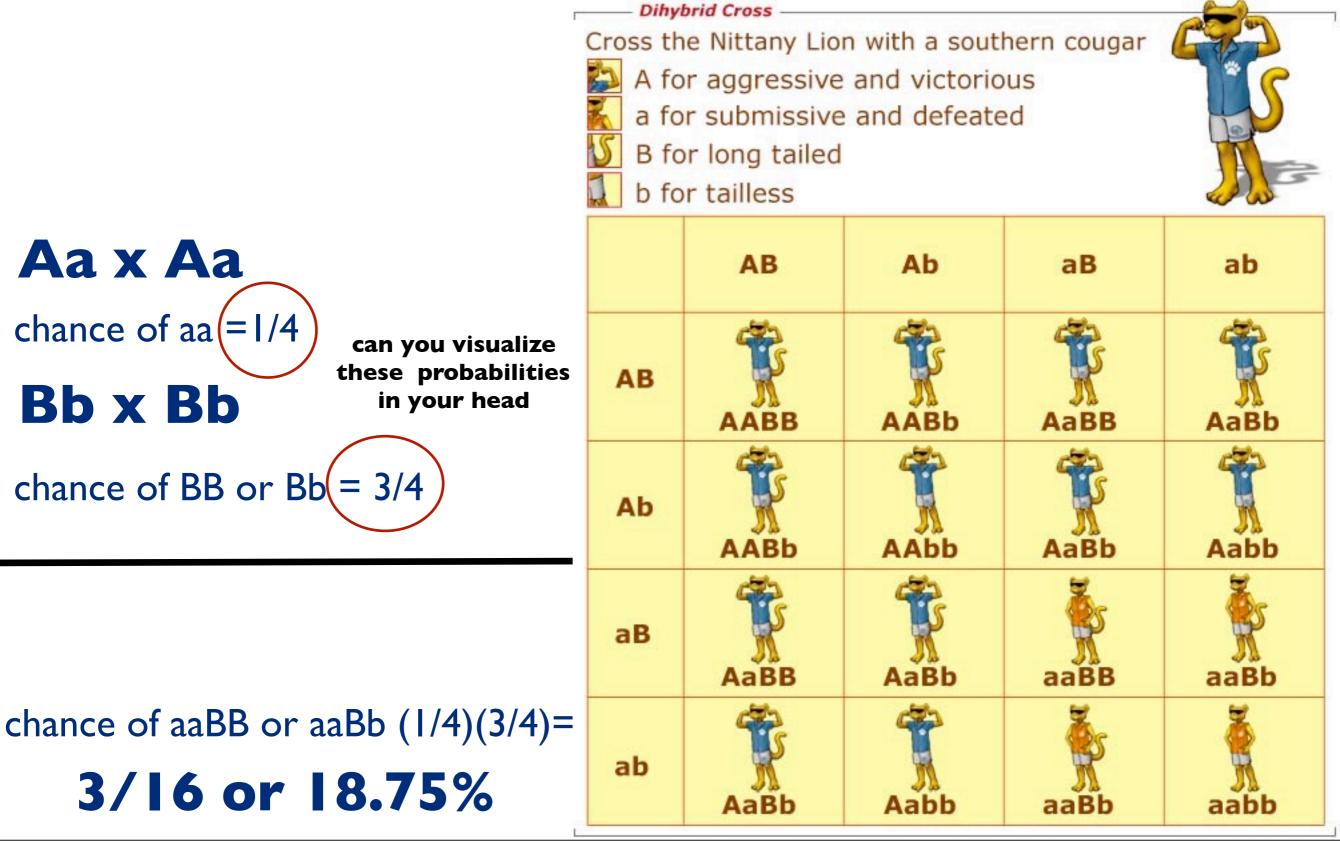








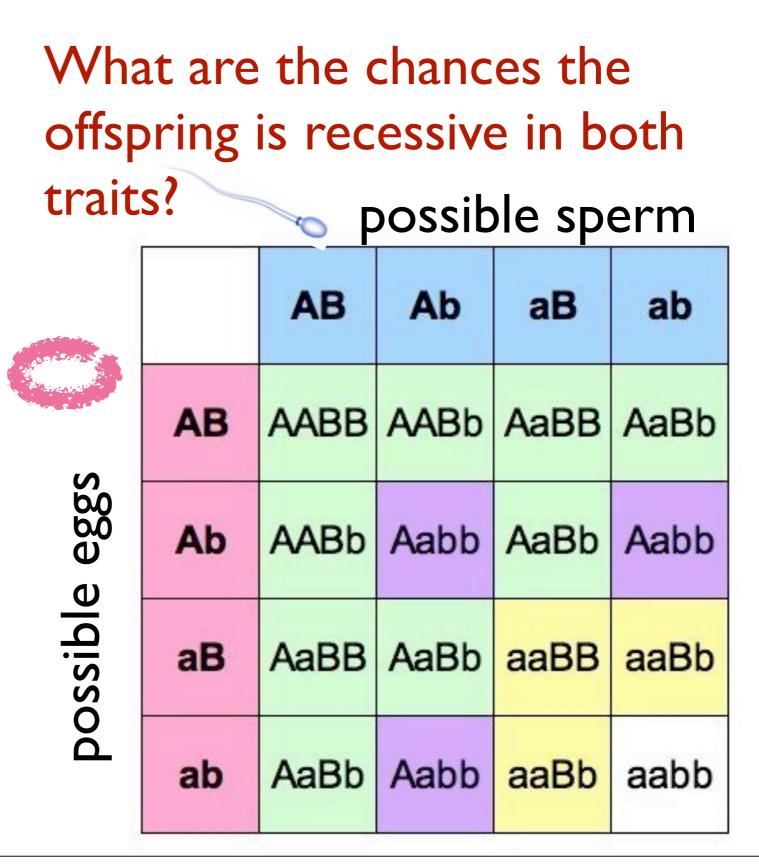




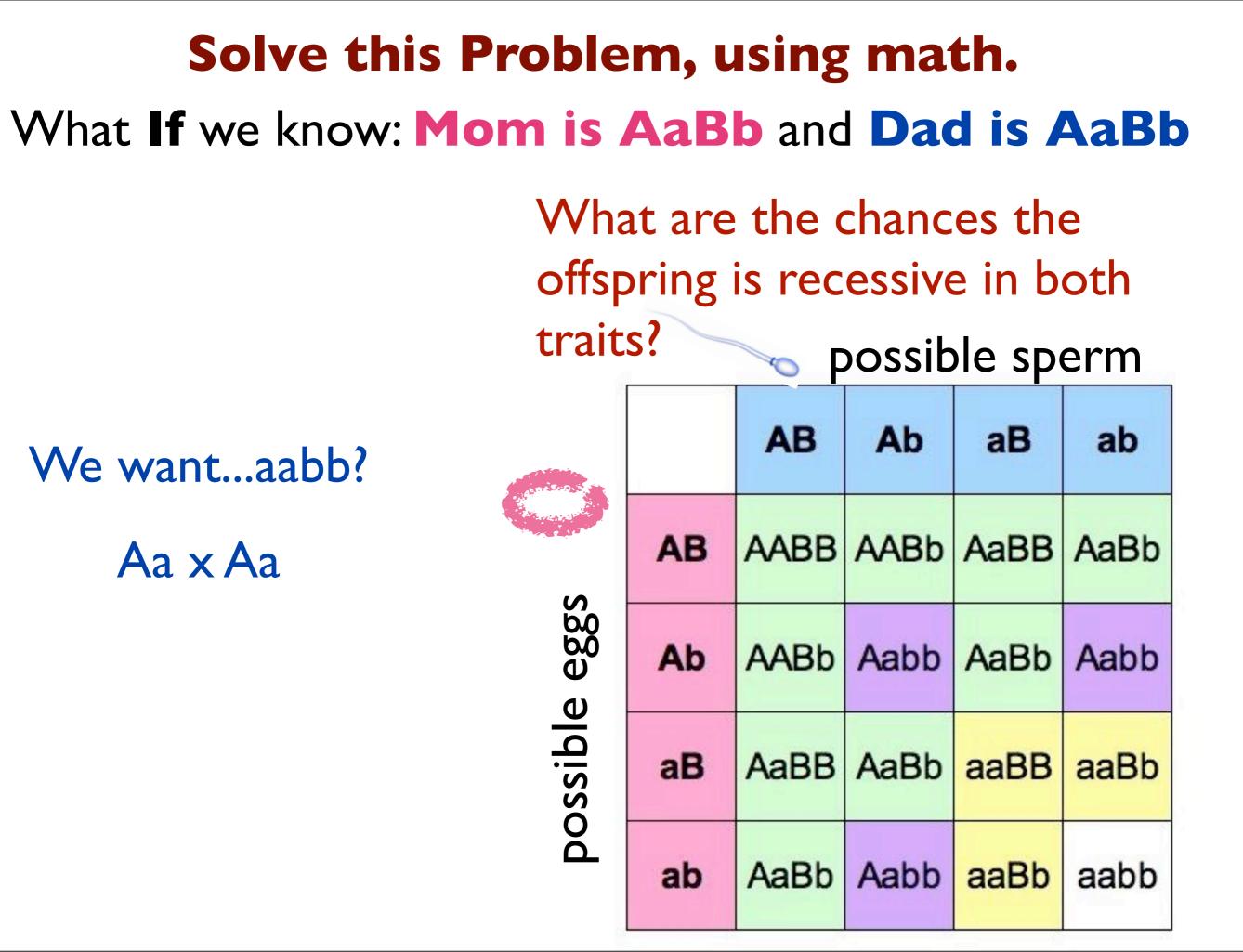
Thursday, February 9, 17

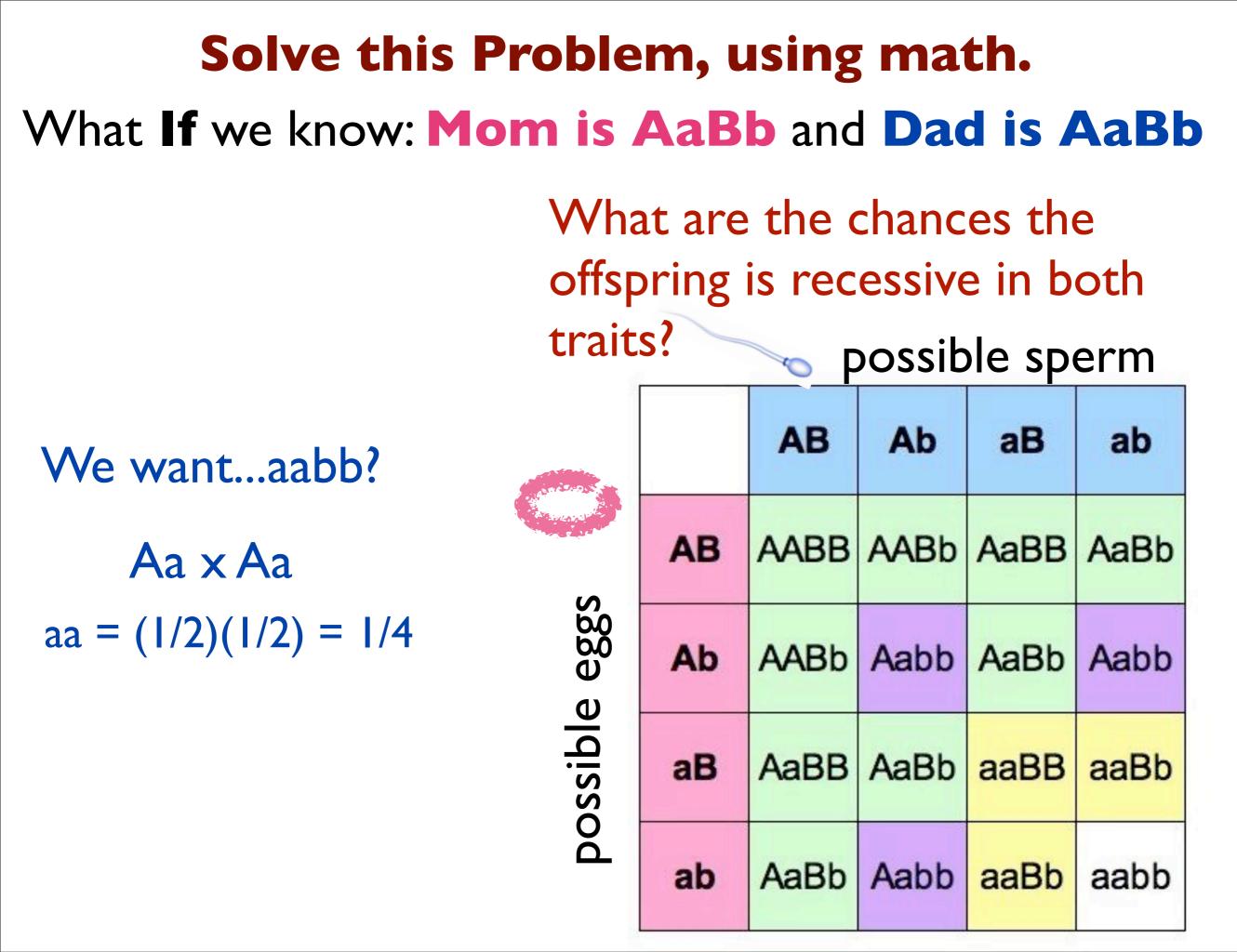
Solve this Problem, using math.

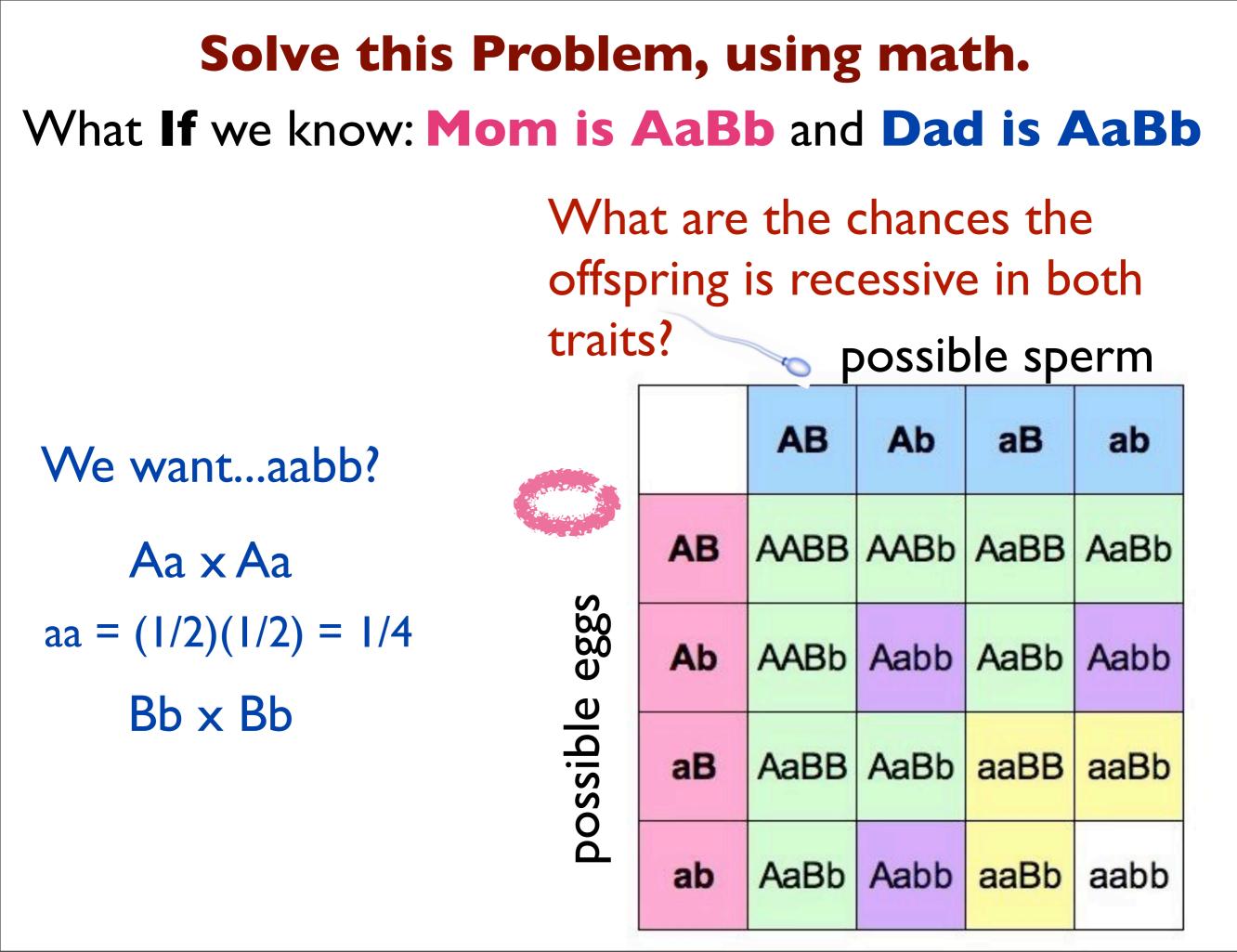
What **If** we know: **Mom is AaBb** and **Dad is AaBb**

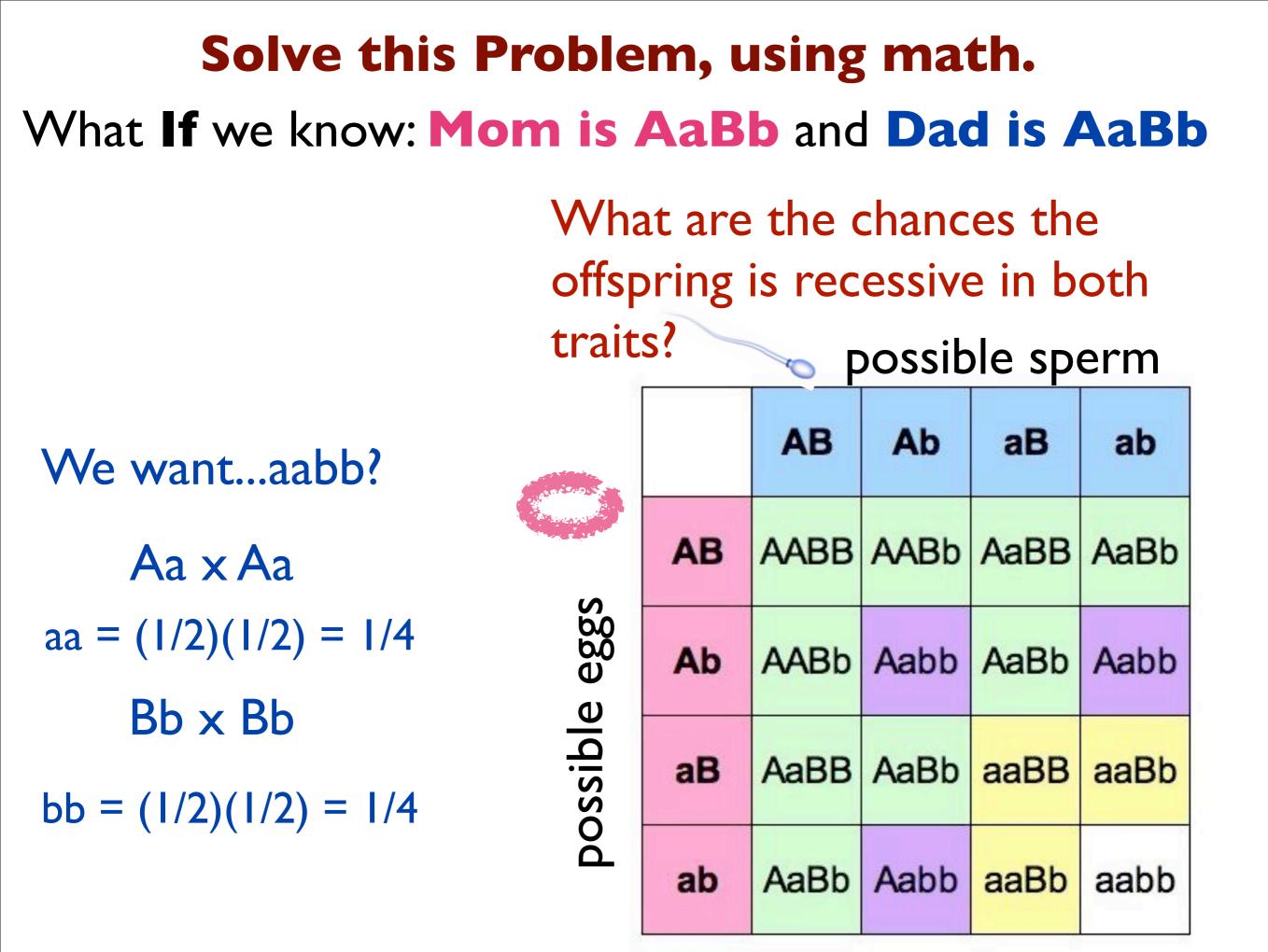


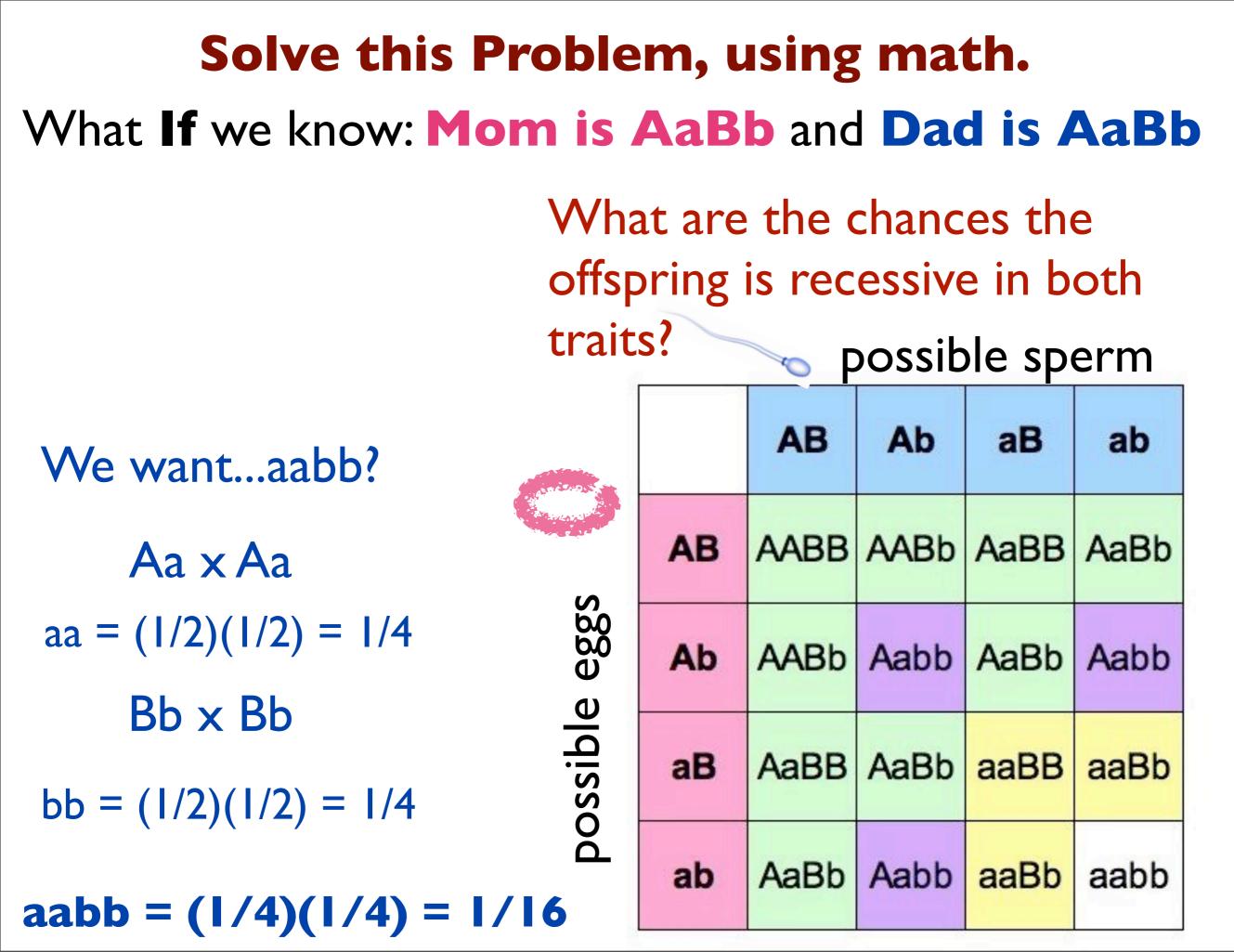
Solve this Problem, using math. What **If** we know: **Mom is AaBb** and **Dad is AaBb** What are the chances the offspring is recessive in both traits? possible sperm AB Ab aB ab We want...aabb? AB AABB AABb AaBB AaBb eggs AABb Aabb AaBb Ab Aabb possible aB AaBB AaBb aaBB aaBb Aabb ab AaBb aaBb aabb





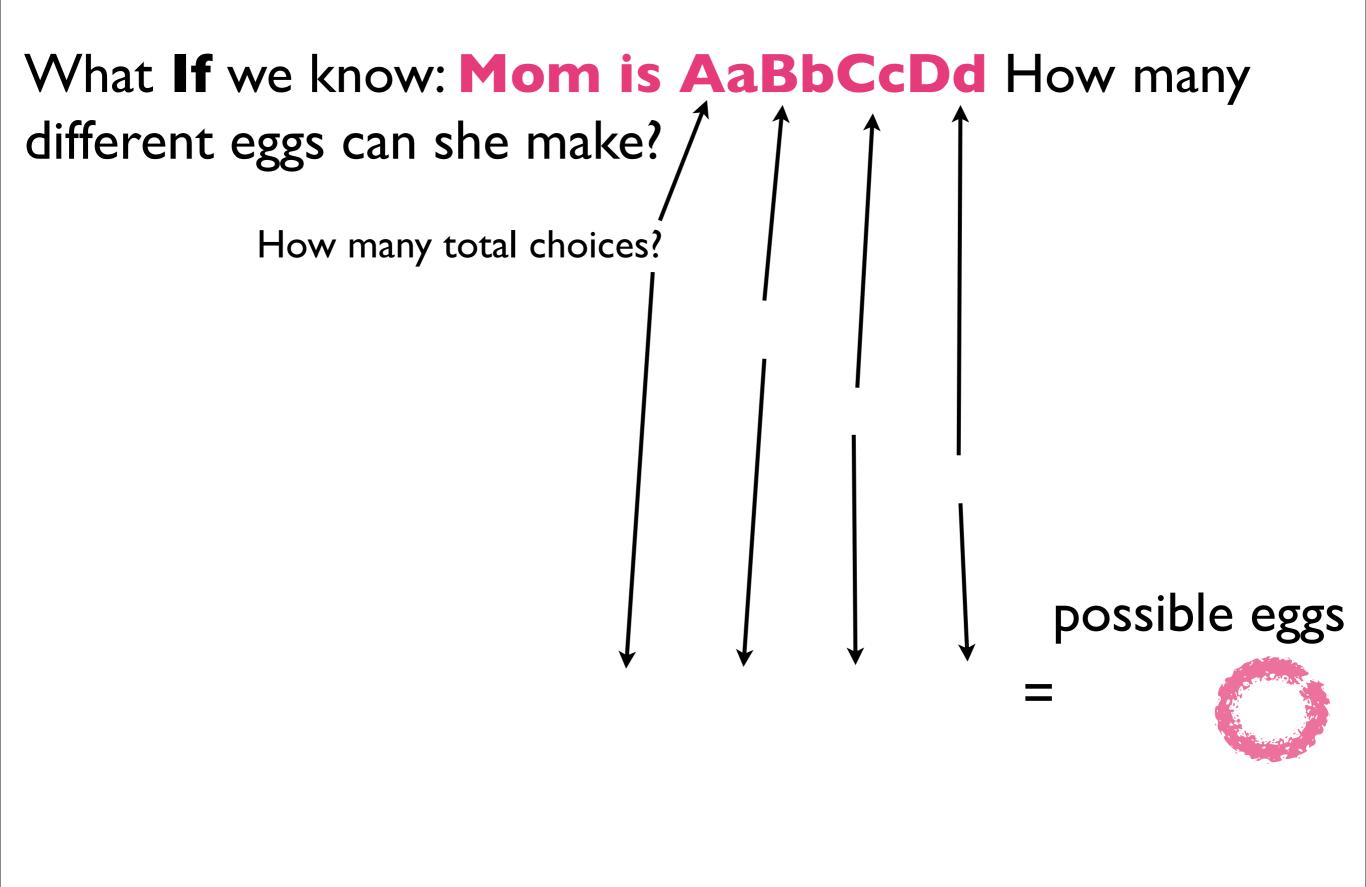


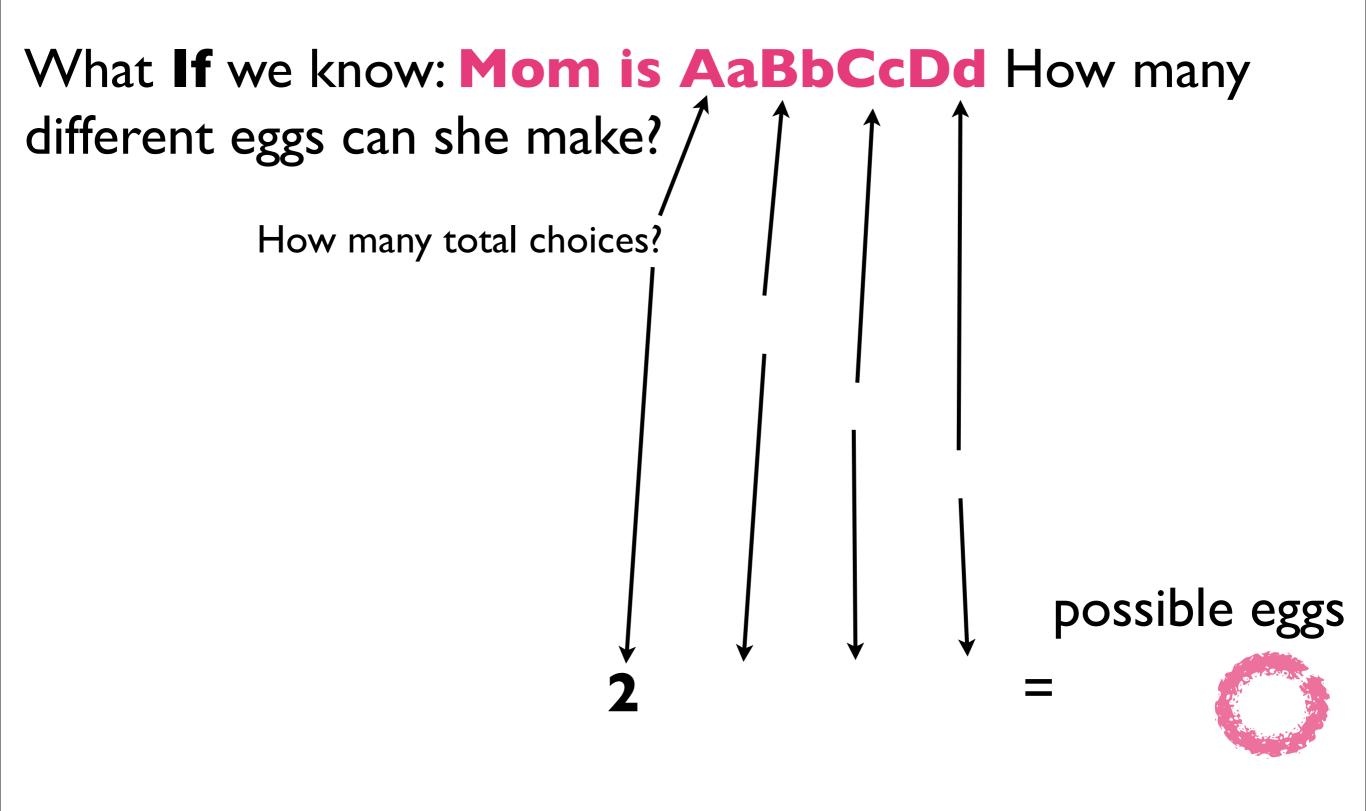


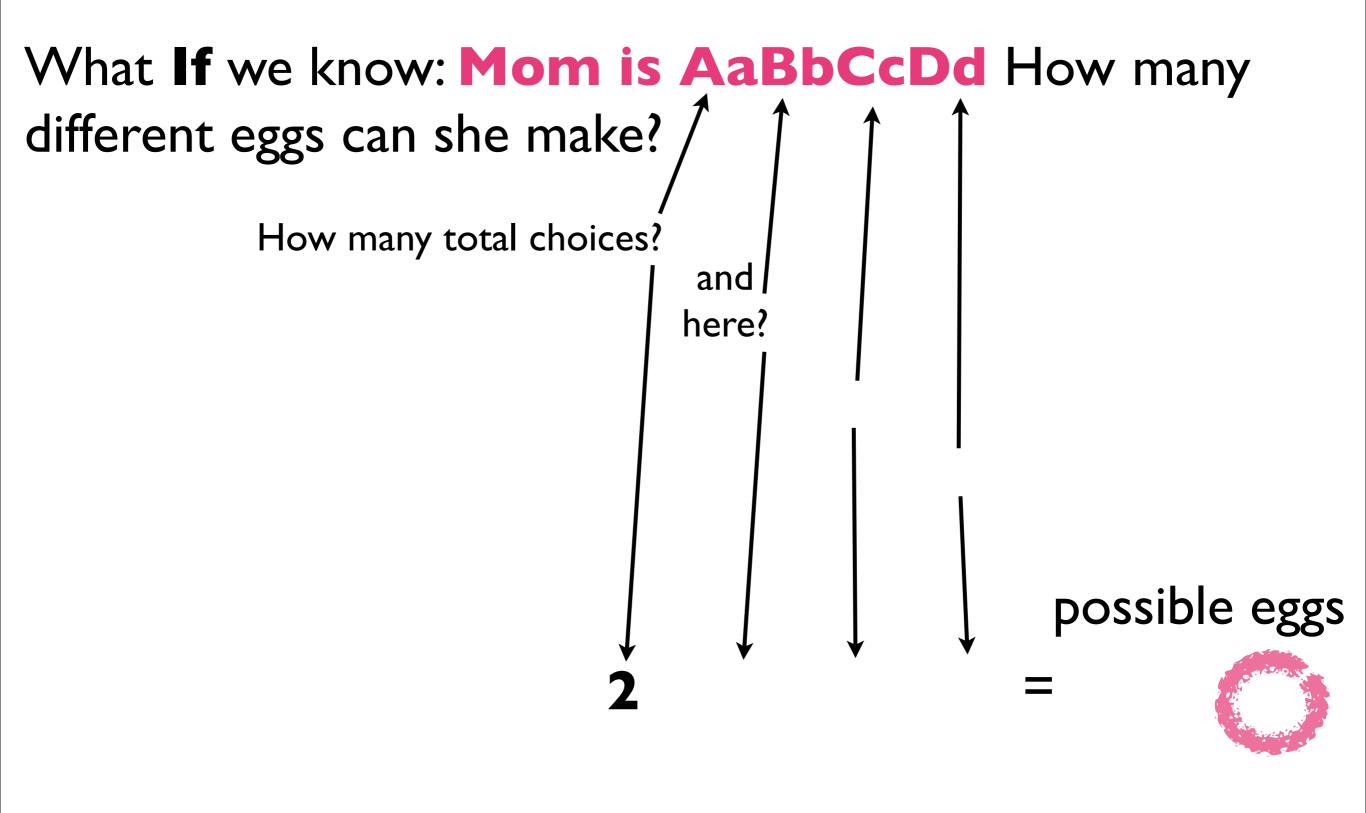


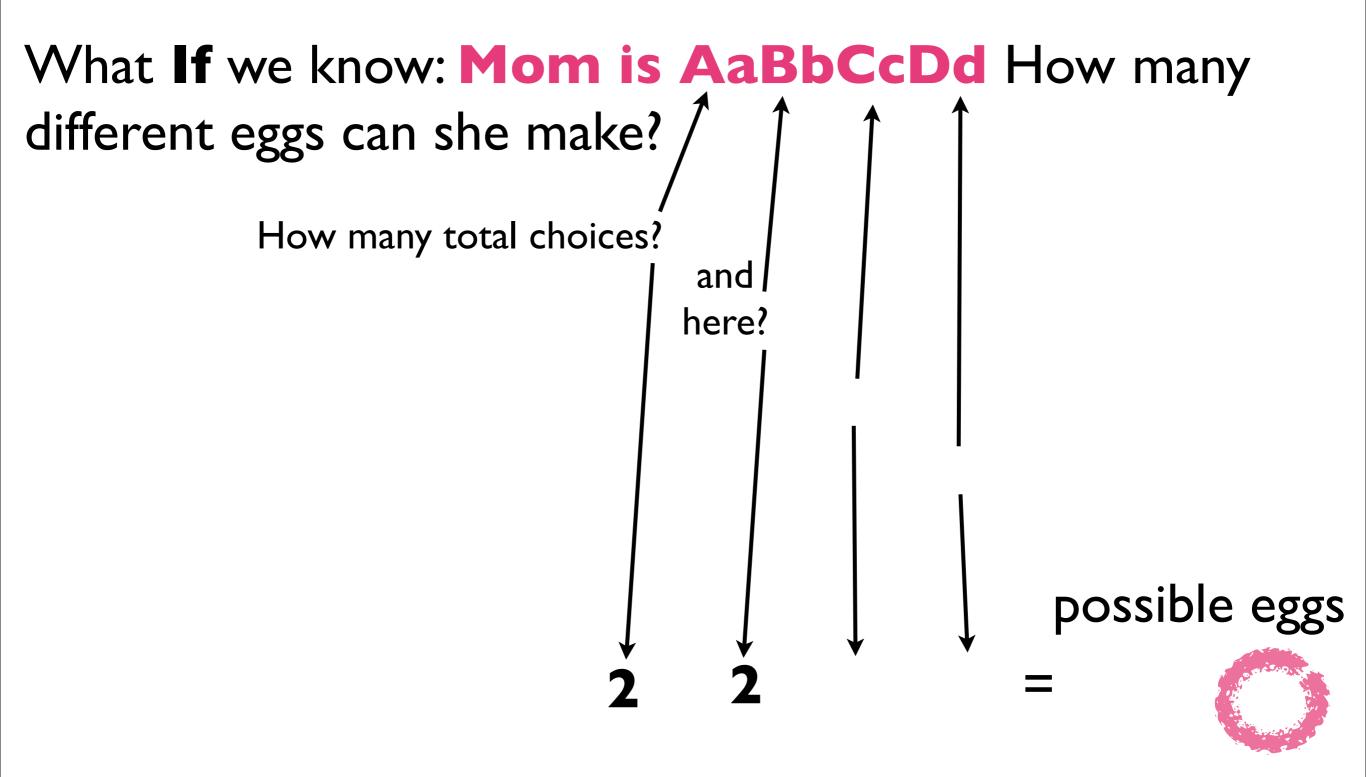
What **If** we know: **Mom is AaBbCcDd** How many different eggs can she make? $\int \int \int \int \int d$

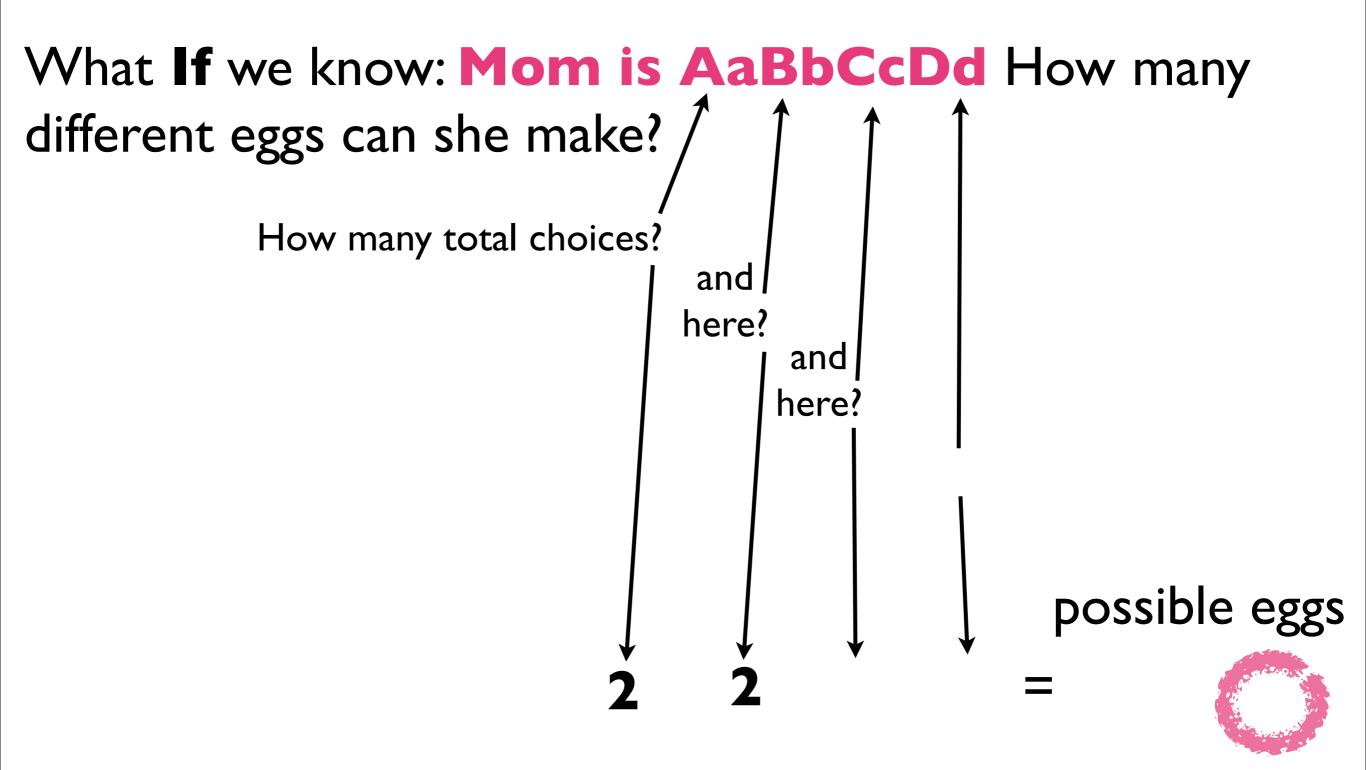
possible eggs

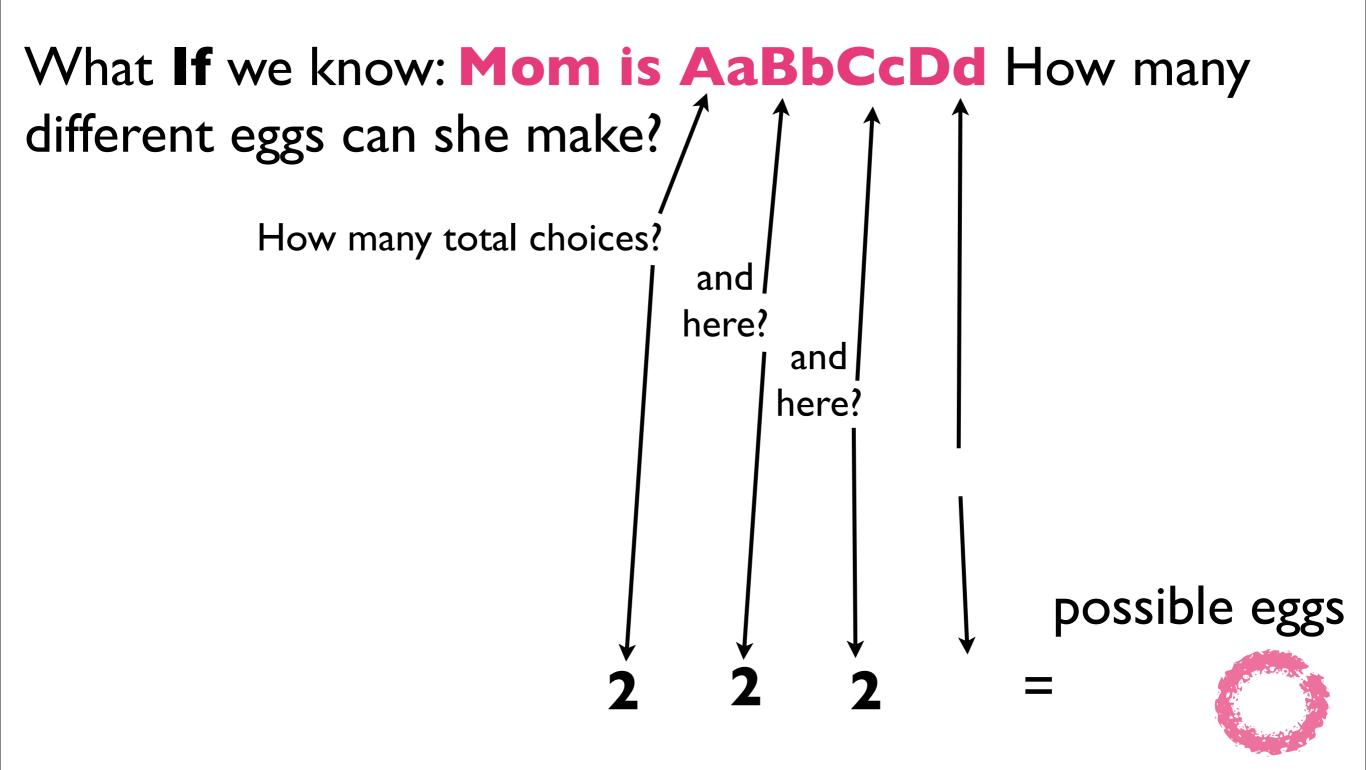


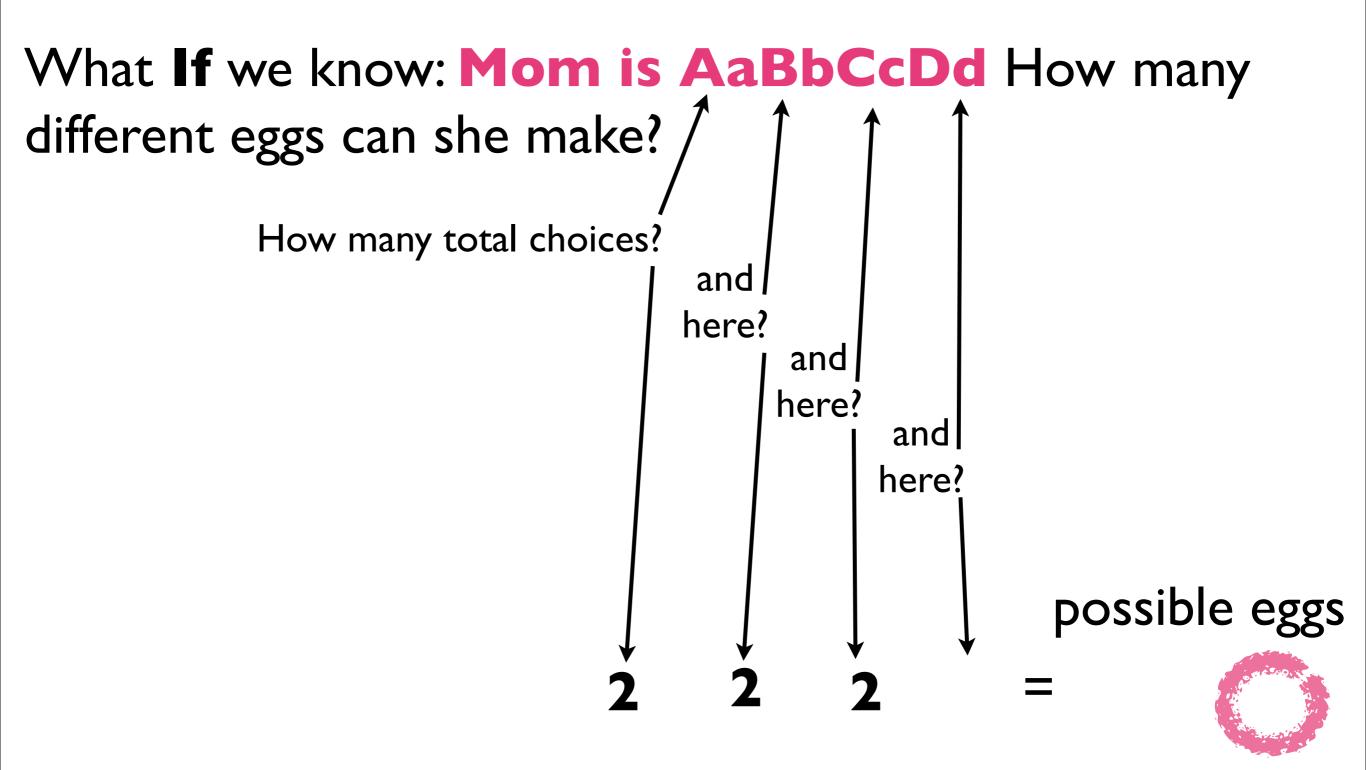


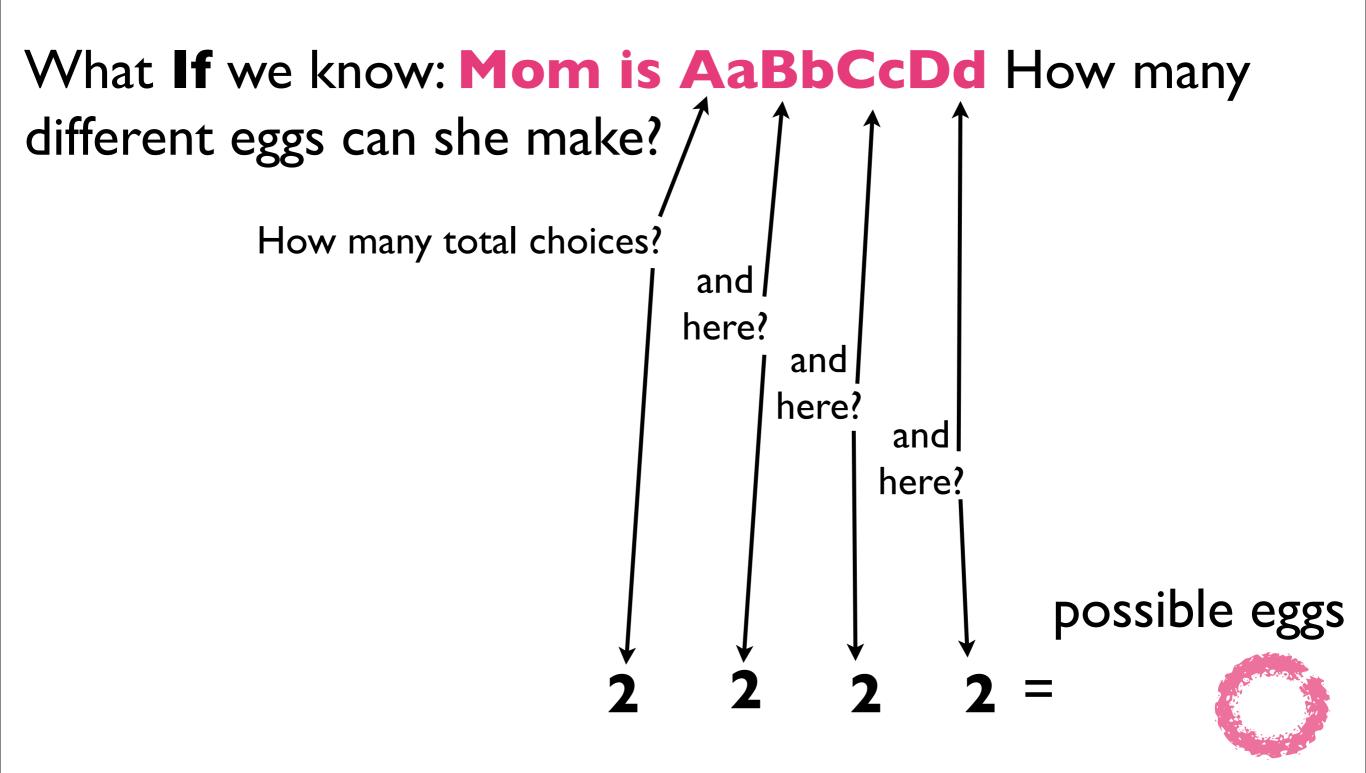


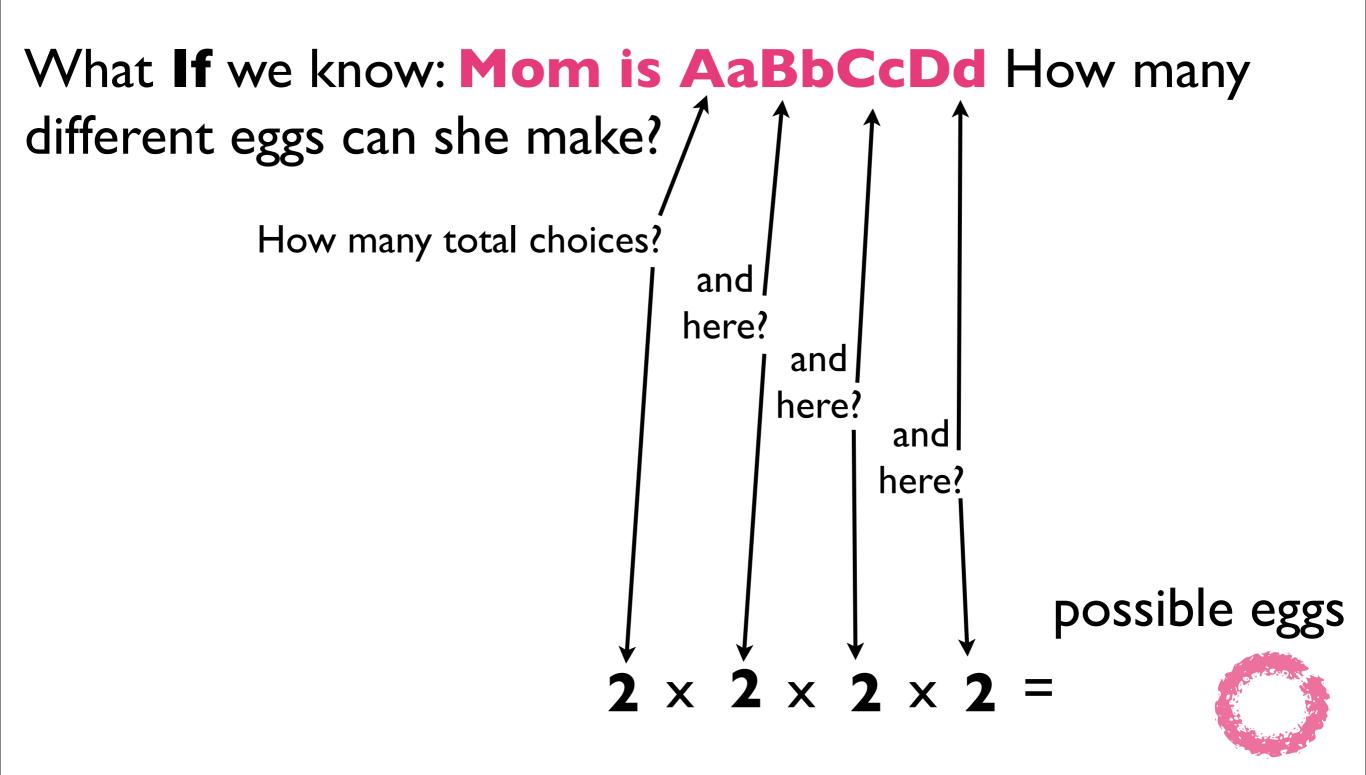


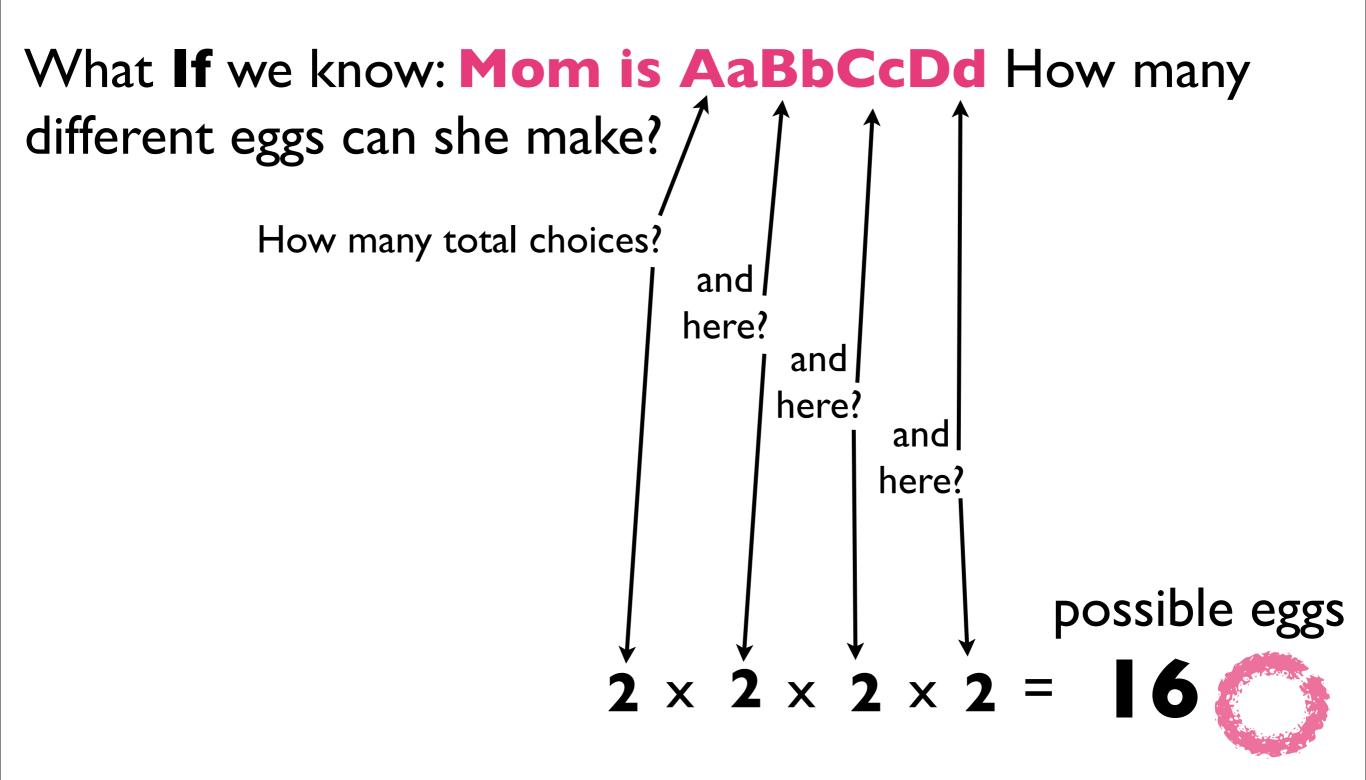


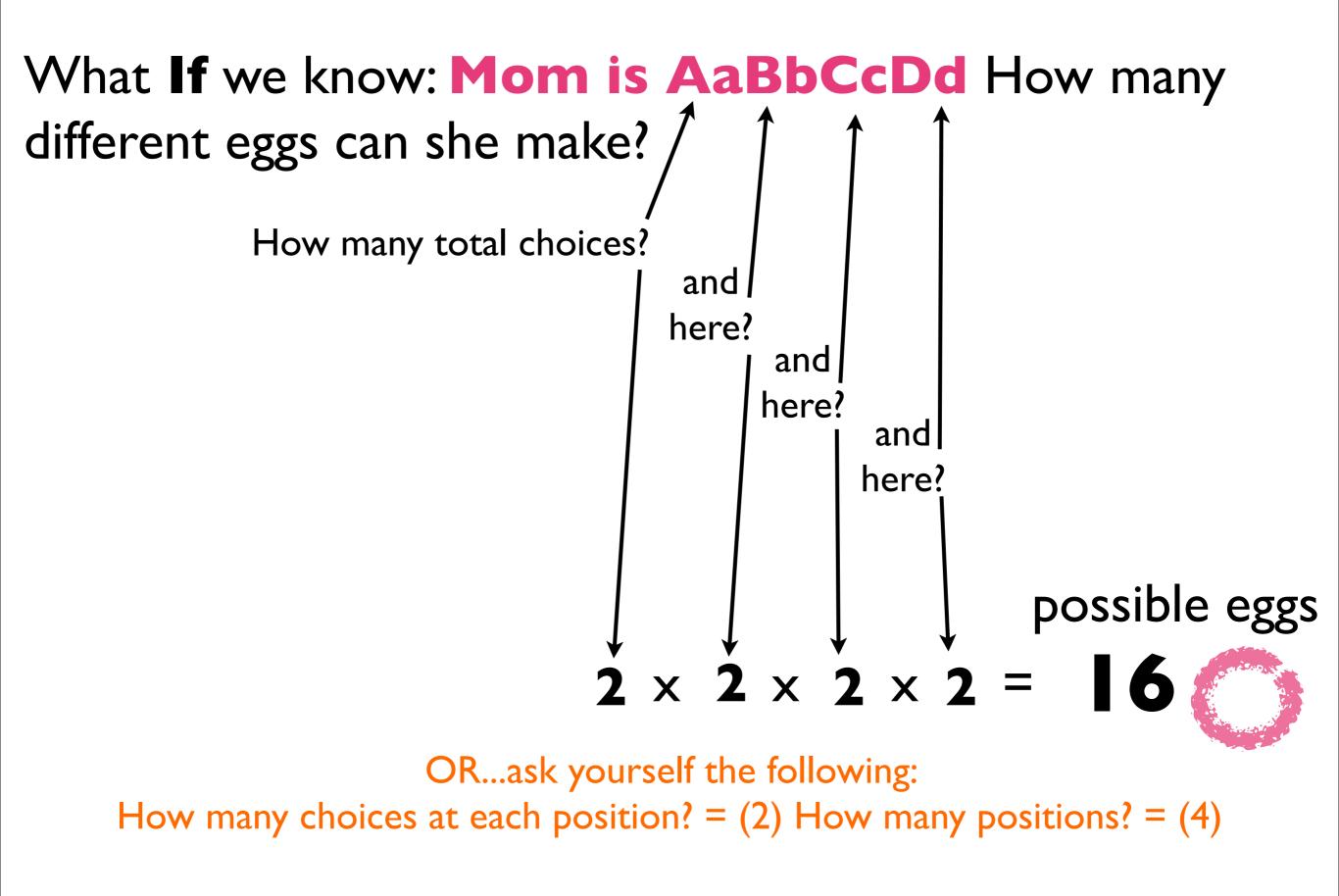


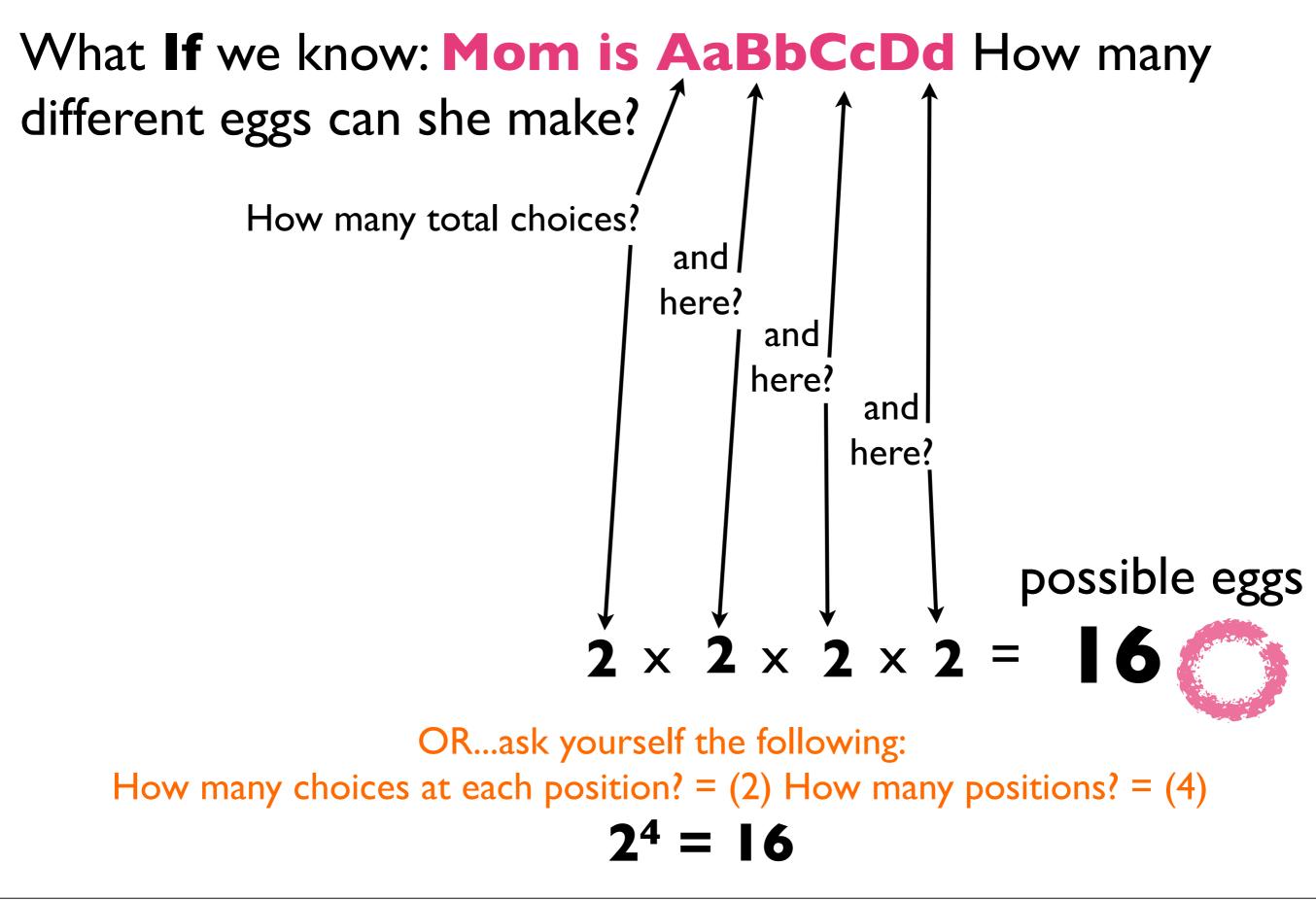




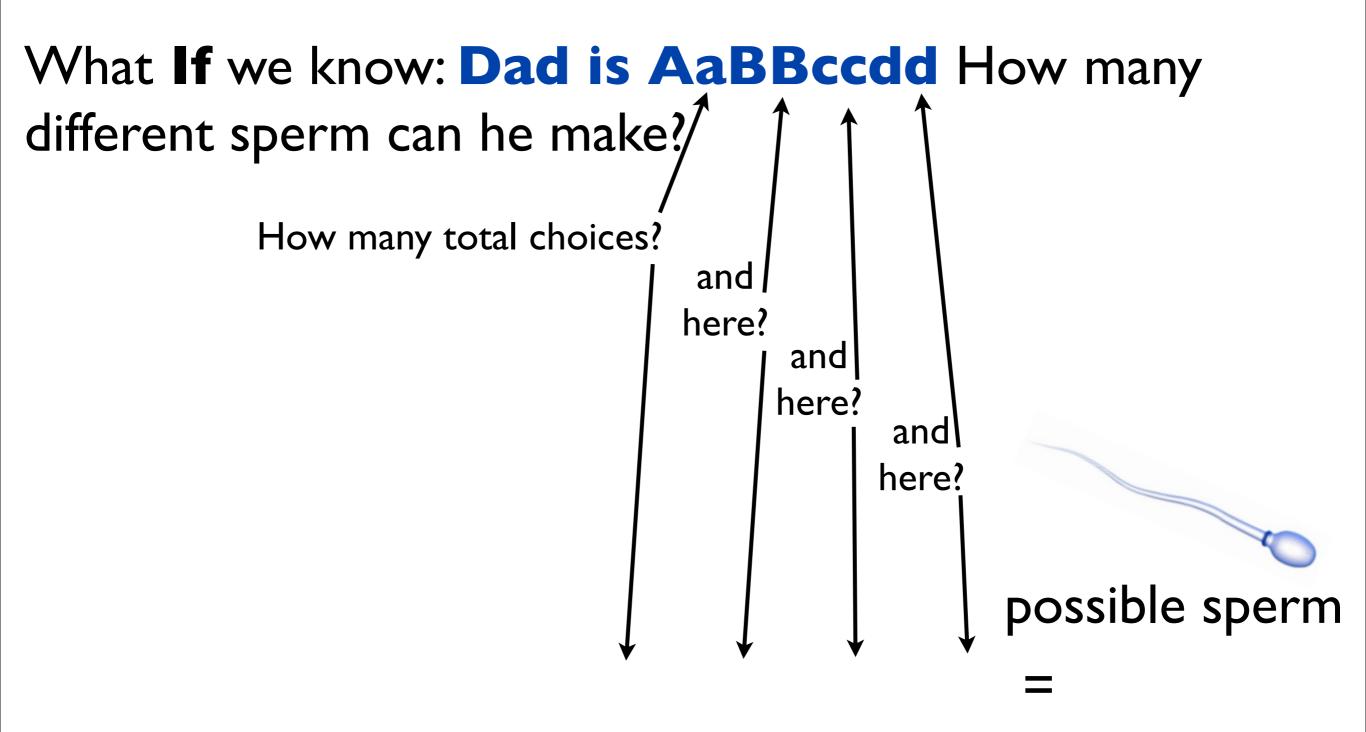




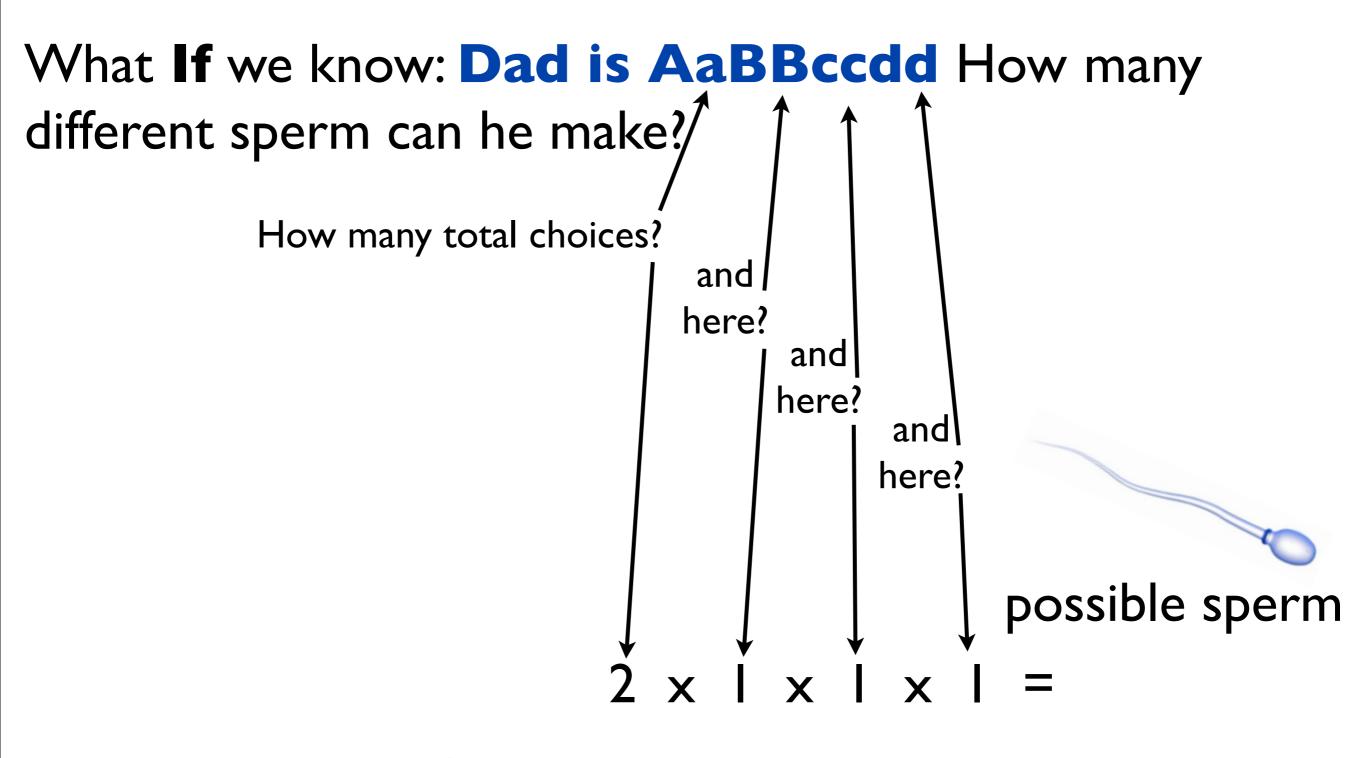




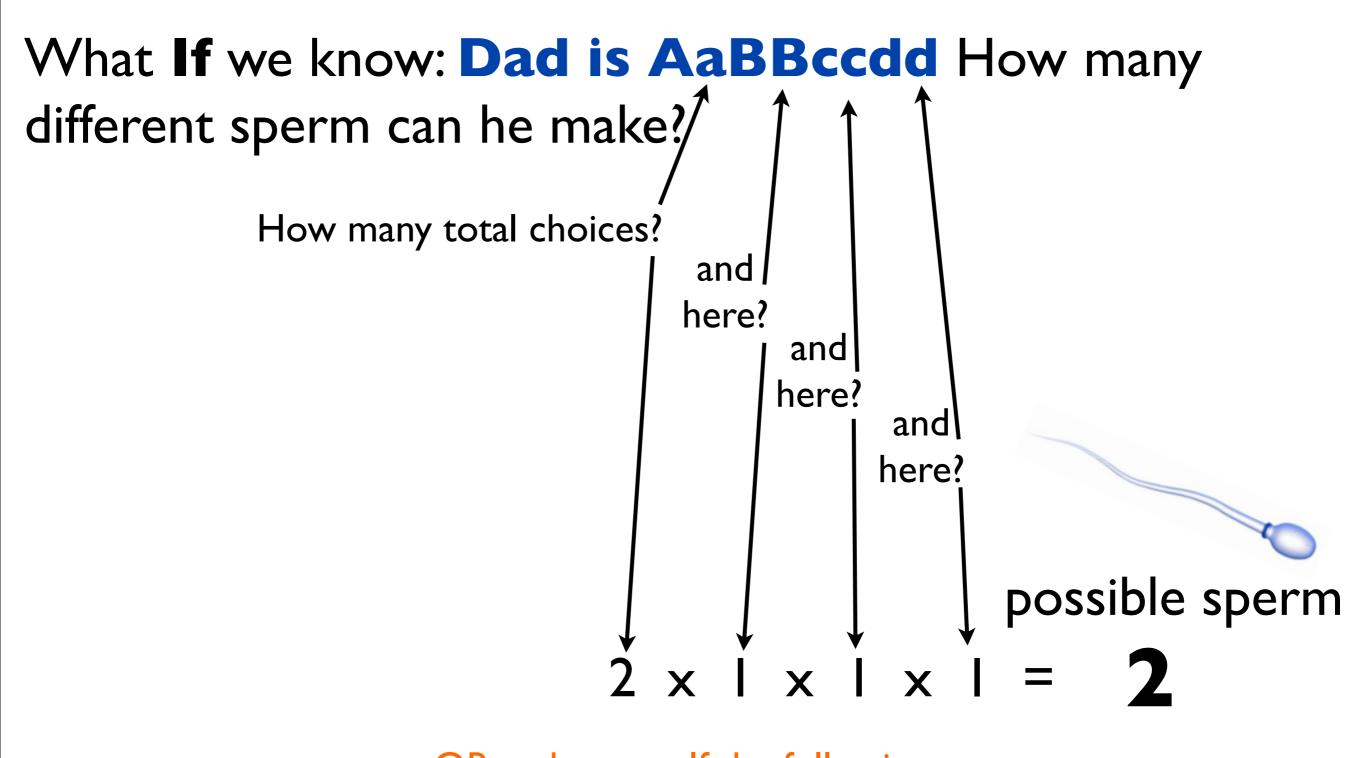
Thursday, February 9, 17



OR...ask yourself the following: How many choices at each position? = (2) How many positions? = (1)



OR...ask yourself the following: How many choices at each position? = (2) How many positions? = (1)

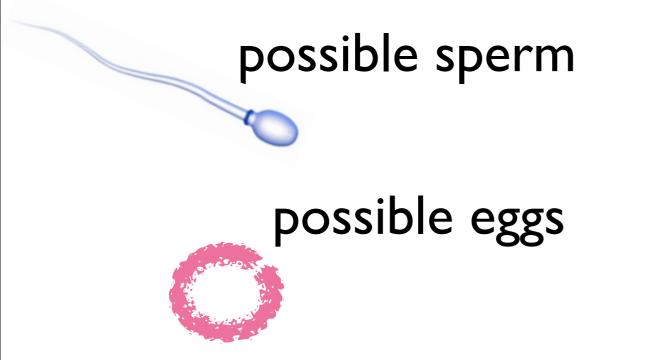


OR...ask yourself the following: How many choices at each position? = (2) How many positions? = (1)

What **If** we know: **Dad is AaBBccdd** How many different sperm can he make?/ How many total choices? and here? and here? and here? possible sperm 2 Х Χ OR...ask yourself the following: How many choices at each position? = (2) How many positions? = (1)2' = 2

Draw a empty punnet square for this cross? How many Boxes?

How many different offspring can this couple make? Phenotypically? Genotypically



Draw a empty punnet square for this cross? How many Boxes?

How many different offspring can this couple make? Phenotypically? Genotypically

possible sperm $2^2 = 4$ possible eggs

Draw a empty punnet square for this cross? How many Boxes?

How many different offspring can this couple make? Phenotypically? Genotypically

possible sperm $2^2 = 4$ possible eggs $2^2 = 4$

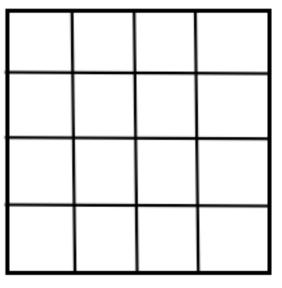
Draw a empty punnet square for this cross? How many Boxes?

How many different offspring can this couple make? Phenotypically? Genotypically

possible sperm

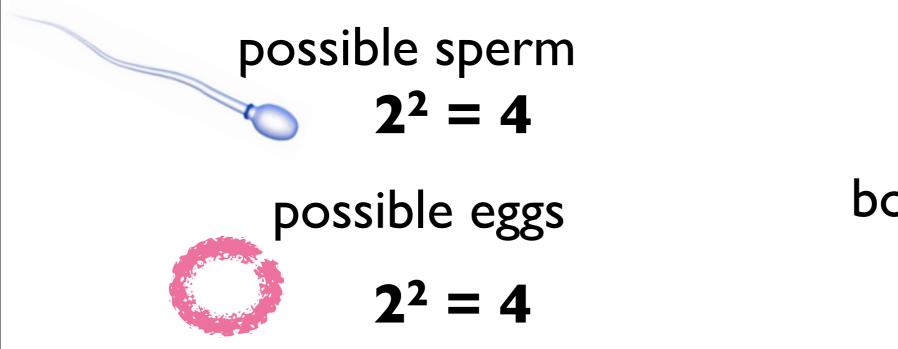
$$2^2 = 4$$

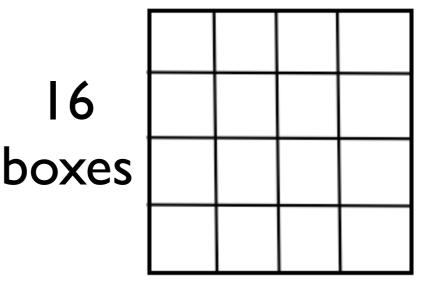
possible eggs
 $2^2 = 4$



Draw a empty punnet square for this cross? How many Boxes?

How many different offspring can this couple make? Phenotypically? Genotypically





Draw a empty punnet square for this cross? How many Boxes?

How many different offspring can this couple make? Phenotypically? Genotypically



(2)(2)(1)(1)(1)(3)(1) = 12 different genotypes (2)(1)(1)(1)(1)(2)(1) = 4 different phenotypes

Essential knowledge 3.A.3: The chromosomal basis of inheritance provides an understanding of the pattern of passage (transmission) of genes from parent to offspring.

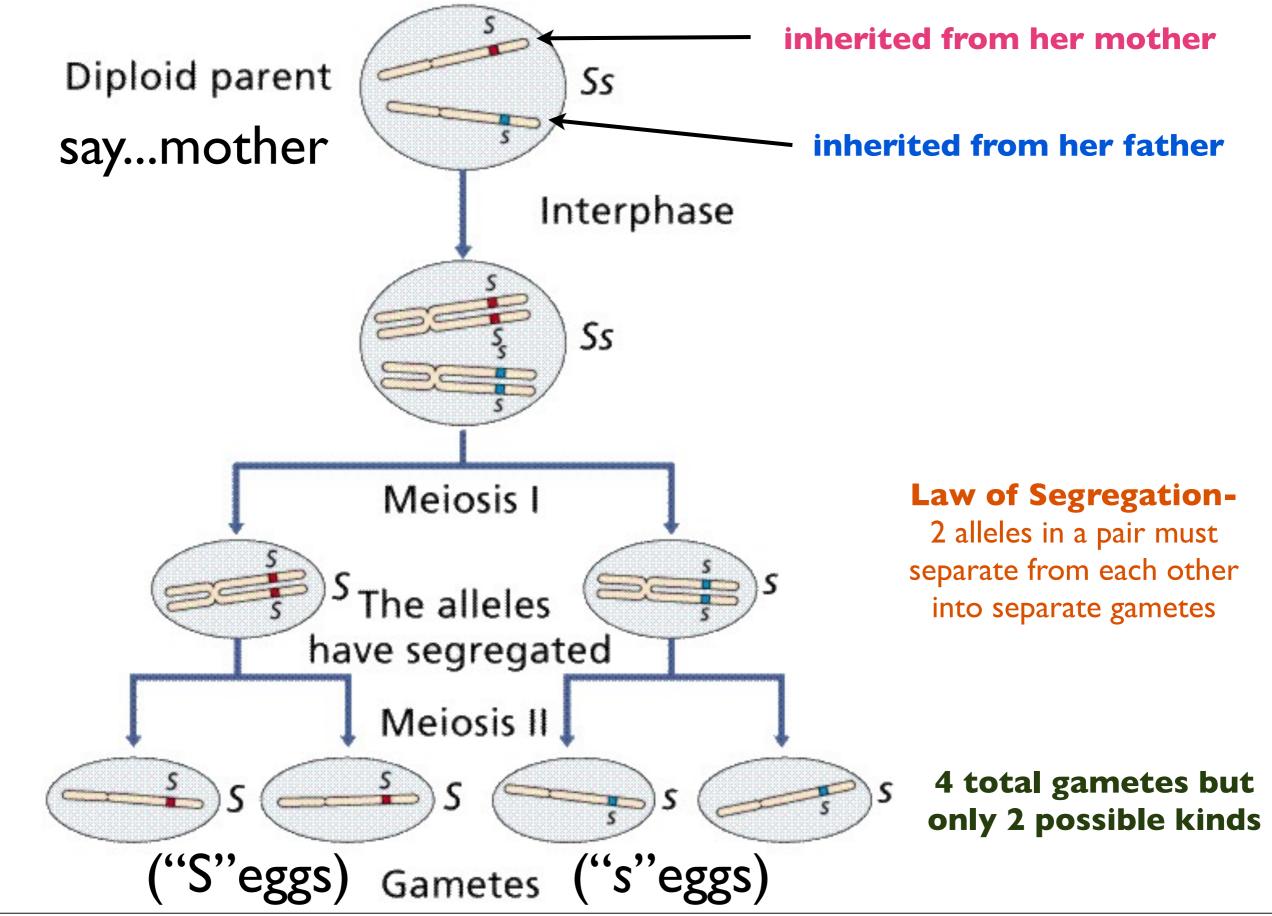
b. Segregation and independent assortment of chromosomes result in genetic variation.

Evidence of student learning is a demonstrated understanding of each of the following:

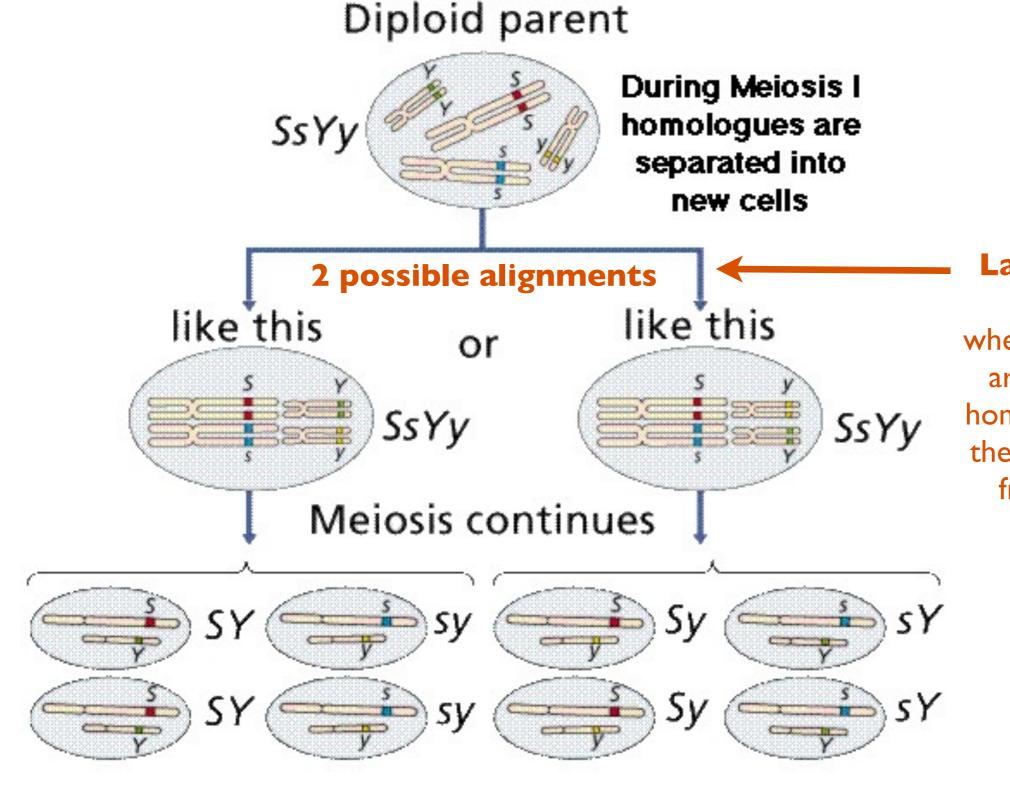
1. Segregation and independent assortment can be applied to genes that are on different chromosomes.

2. Genes that are adjacent and close to each other on the same chromosome tend to move as a unit; the probability that they will segregate as a unit is a function of the distance between them.

Follow the chromosome, follow the traits!



We Can Follow Two Traits Simultaneously

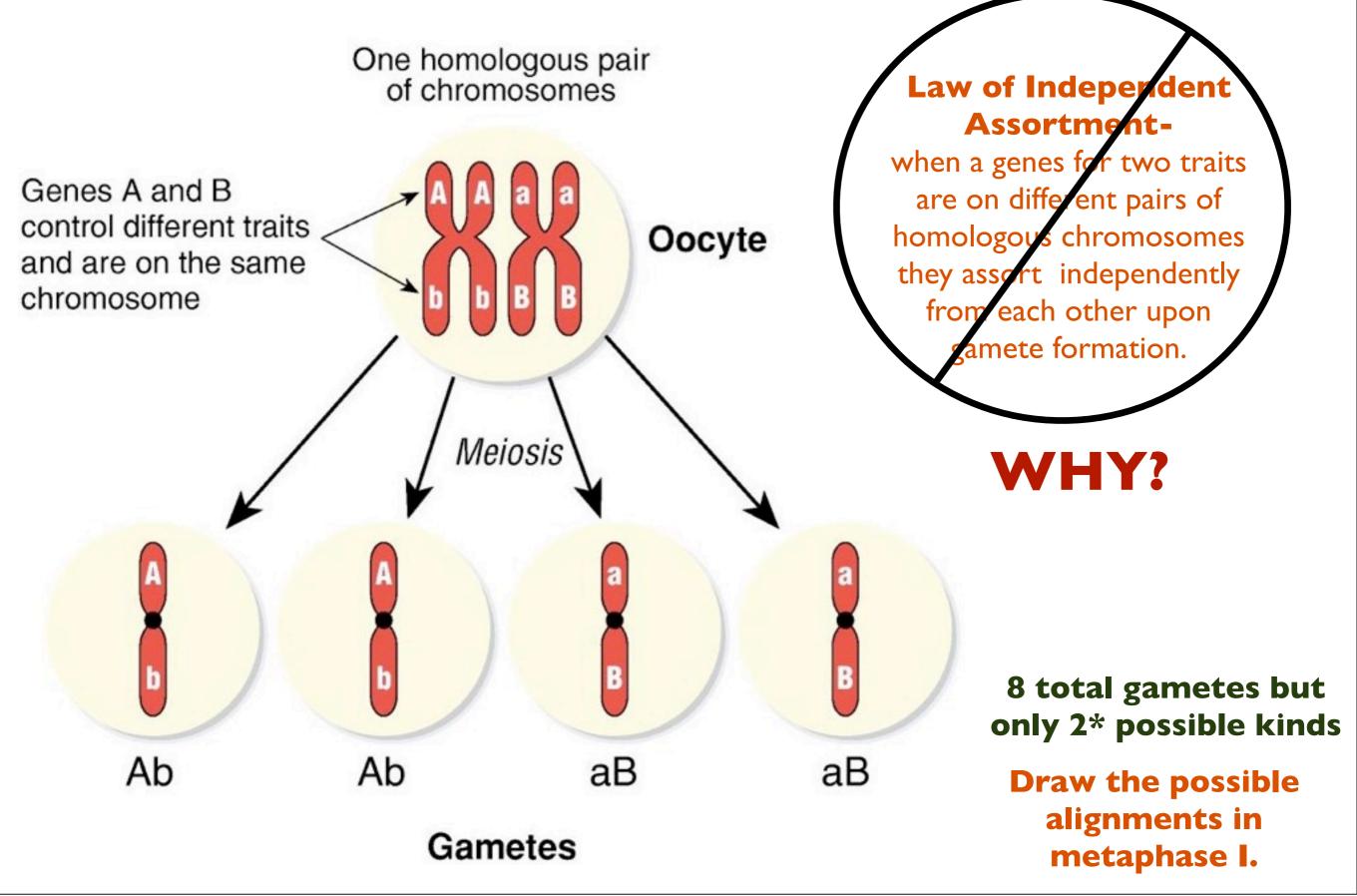


Law of Independent Assortment-

when a genes for two traits are on different pairs of homologous chromosomes they assort independently from each other upon gamete formation.

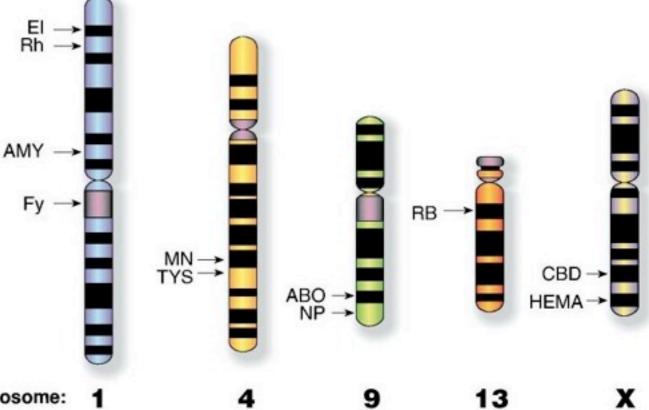
S assorts with Y or y s assorts with Y or y 8 total gametes but only 4 possible kinds

We Can Follow Two Traits Simultaneously



Autosomal Gene Linked Traits

- **IF** there are far more genes then chromosomes,
- and chromosomes carry genes,
- then each chromosome must carry multiple genes. Chromosome:
 - And they do!



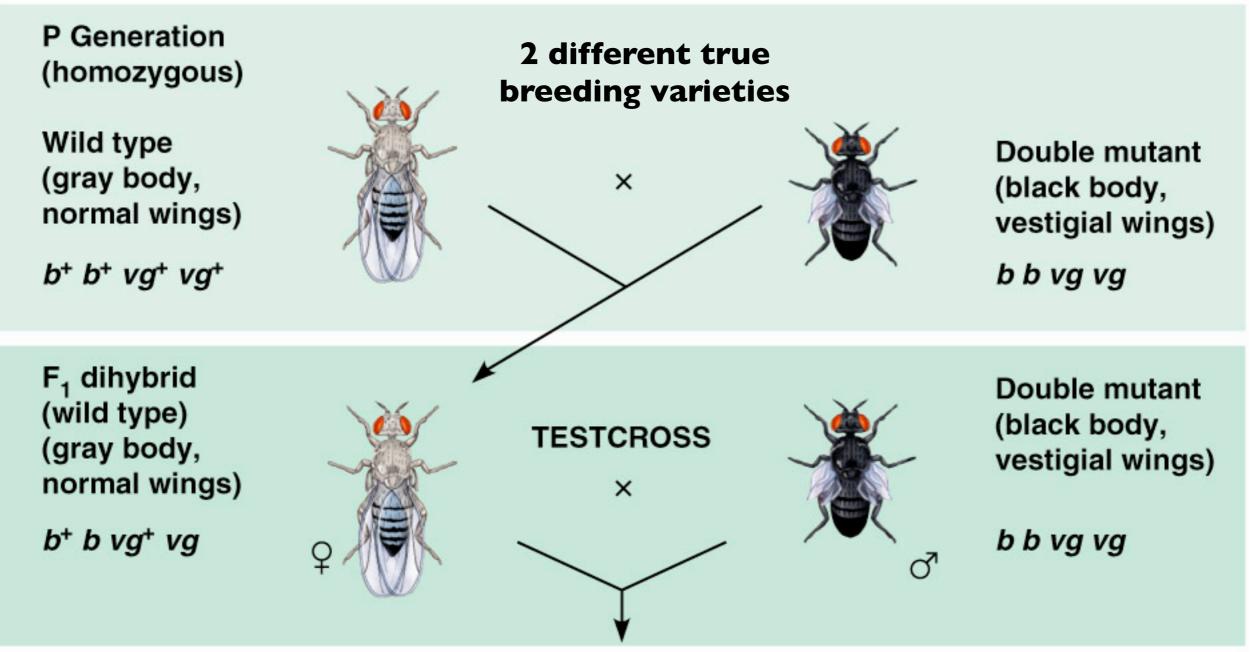
GENE SYMBOLS

ABO ABO blood type Production of amylase enzyme AMY CBD One form of colour blindness EI Shape of red blood cells Fy Duffy blood type HEMA Production of a blood clotting factor NP Structure of nails and kneecaps Rh Rhesus blood type Retinoblastoma (a cancer of the eye) RB MN MN blood type TYS Skin structure

Autosomal Gene Linked Traits

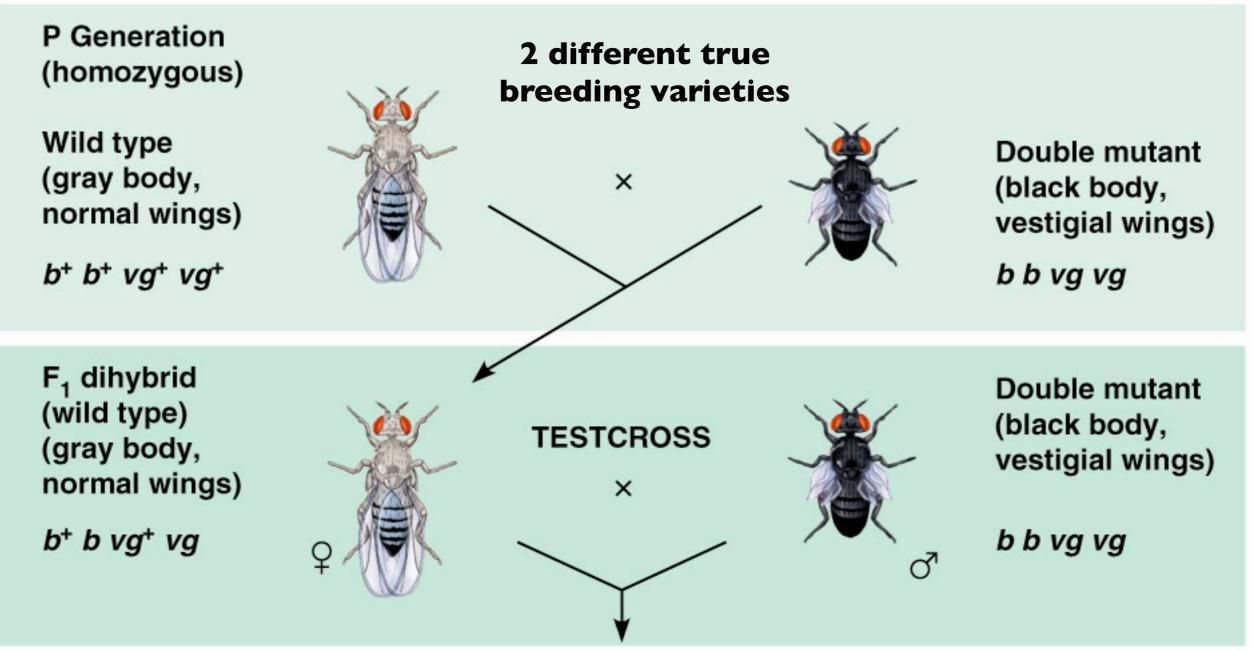
- Genes on the same autosomal (non sex chromosome) chromosome are said to be linked.
- Because linked genes travel together the results of breeding experiments deviate from those expected from Mendel's Law of Independent Assortment.
- Thomas Hunt Morgan, explored the idea of gene linkage by running a series of breeding experiments with fruit flies.

Morgan's Gene Linkage Work

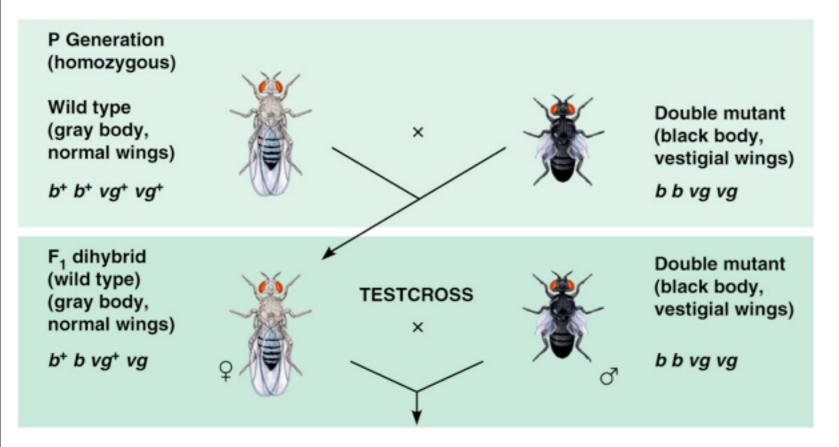


What do you think Morgan was expecting in the F2 generation?

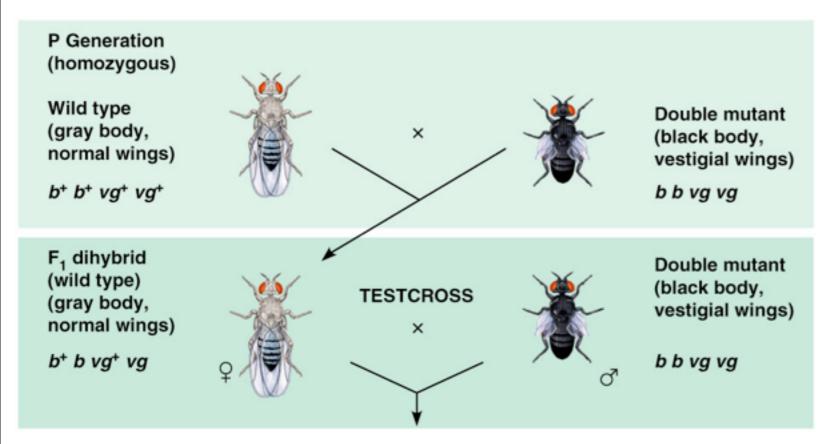
Morgan's Gene Linkage Work



What do you think Morgan was expecting in the F2 generation?



What do you think Morgan was expecting in the F2 generation?

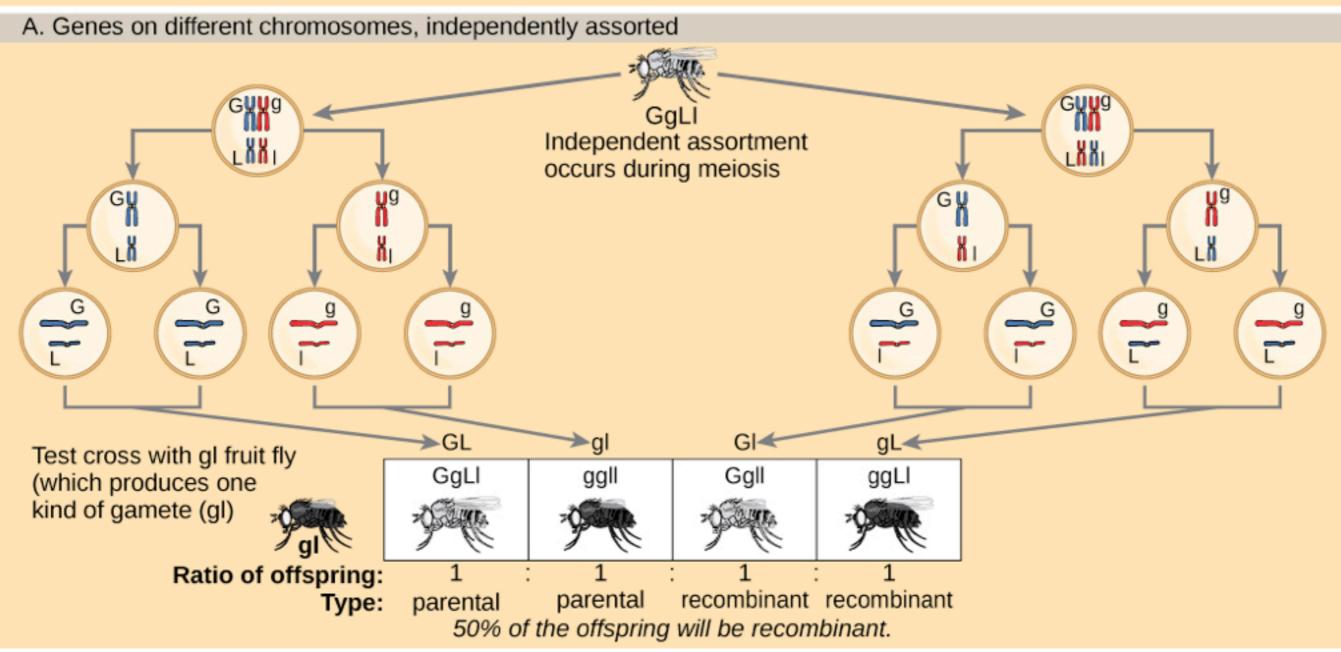


What do you think Morgan was expecting in the F2 generation?

He assumed that the genes were on separate chromosomes and thus they would assort independently from each other resulting in the predicted 1:1:1:1 ratio

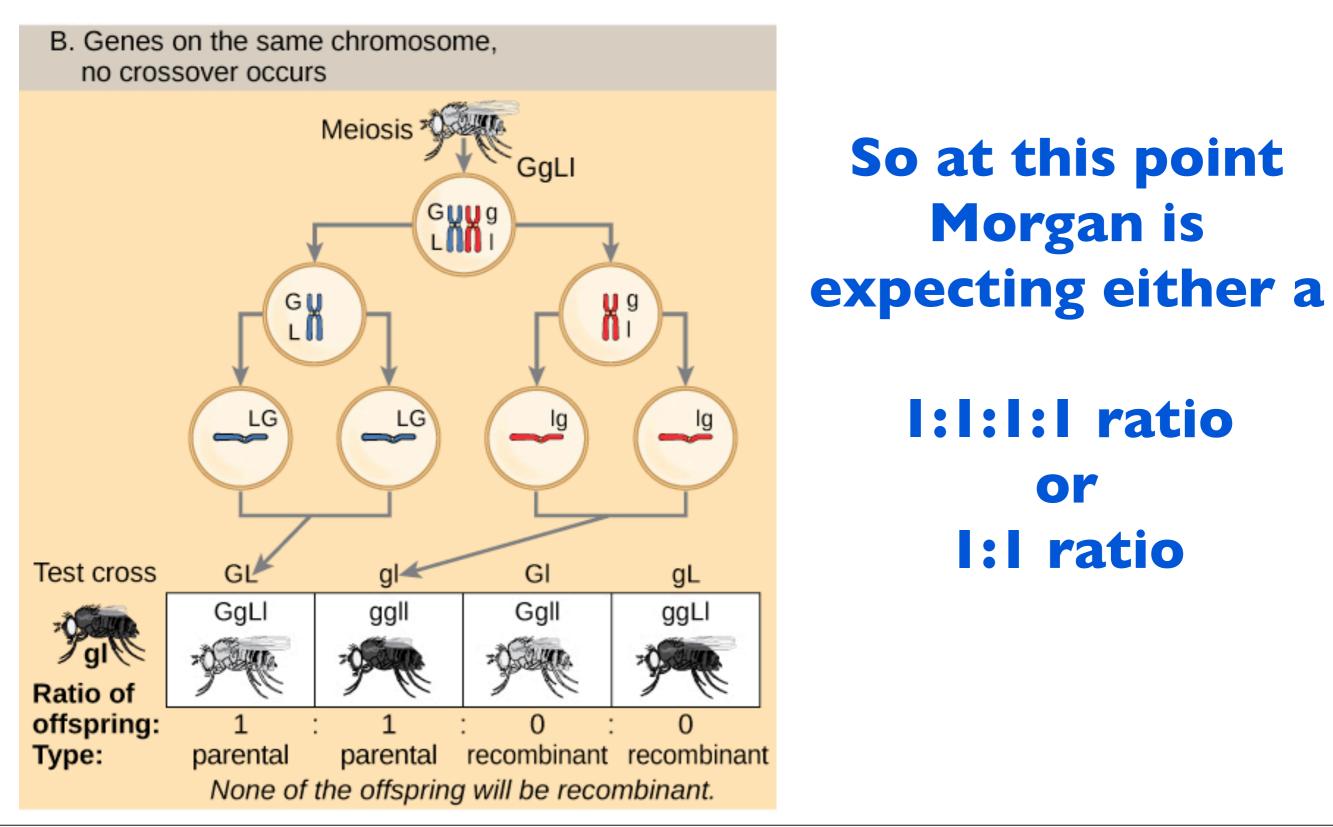
Inheritance Pattern of Linked and Unlinked Genes

Three hypothetical inheritance patterns for a test cross between a heterozygote and a homozygous recessive individual, based on gene placement, are shown in A through C. The actual experimental results published by Thomas Hunt Morgan in 1912 are shown in D.

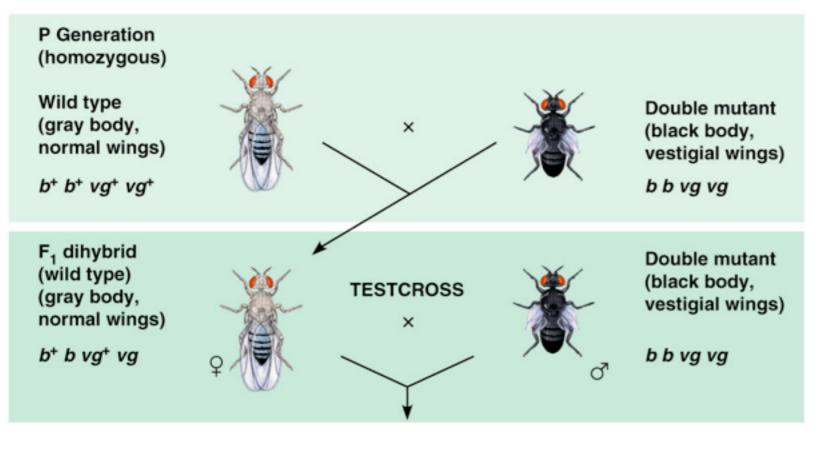


Thursday, February 9, 17

He also considered the alternative... that the genes were on the same chromosome and thus they would not assort independently from each other resulting in the predicted 1:1 ratio



He also considered the alternative... that the genes were on the same chromosome and thus they would not assort independently from each other resulting in the predicted 1:1 ratio



So at this point Morgan is expecting either

> I:I:I:I ratio or I:I ratio

So... which did he get?

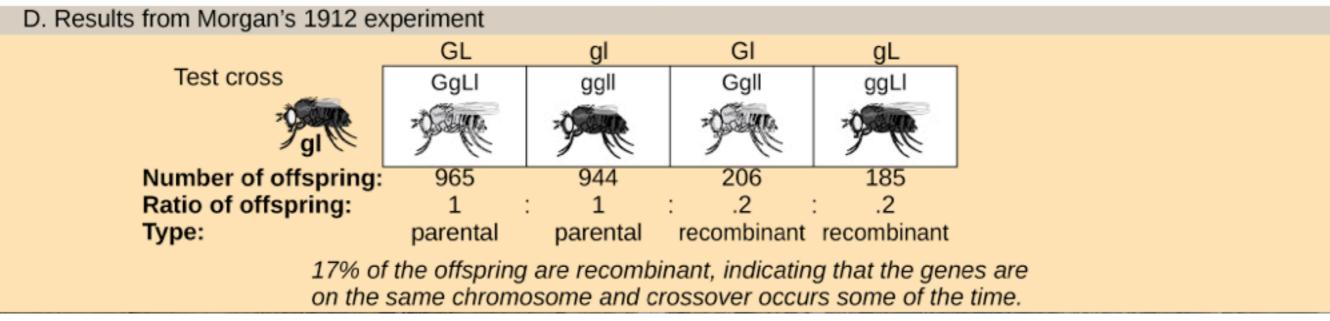
BUT, How would he explain the unexpected 17% that he called recombinants?

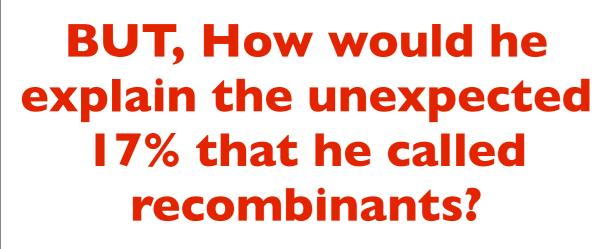
Thursday, February 9, 17

So... which did he get? NEITHER

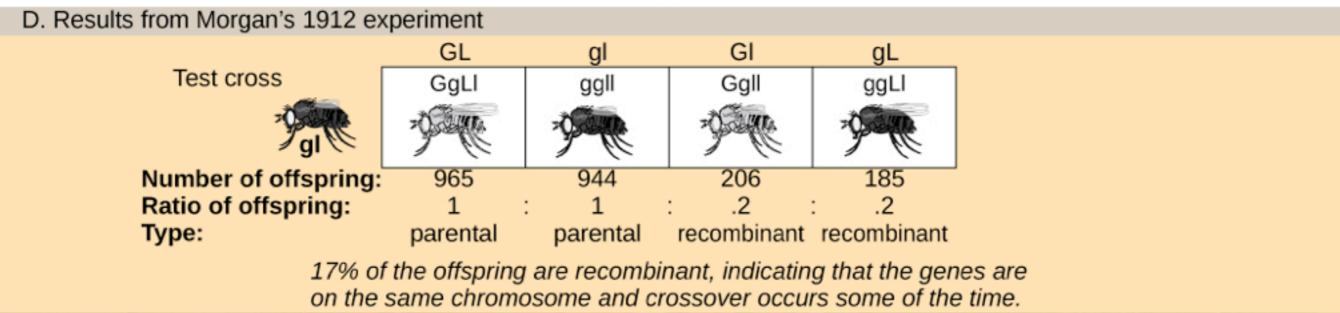
BUT, How would he explain the unexpected 17% that he called recombinants?

So... which did he get? NEITHER





So... which did he get? NEITHER



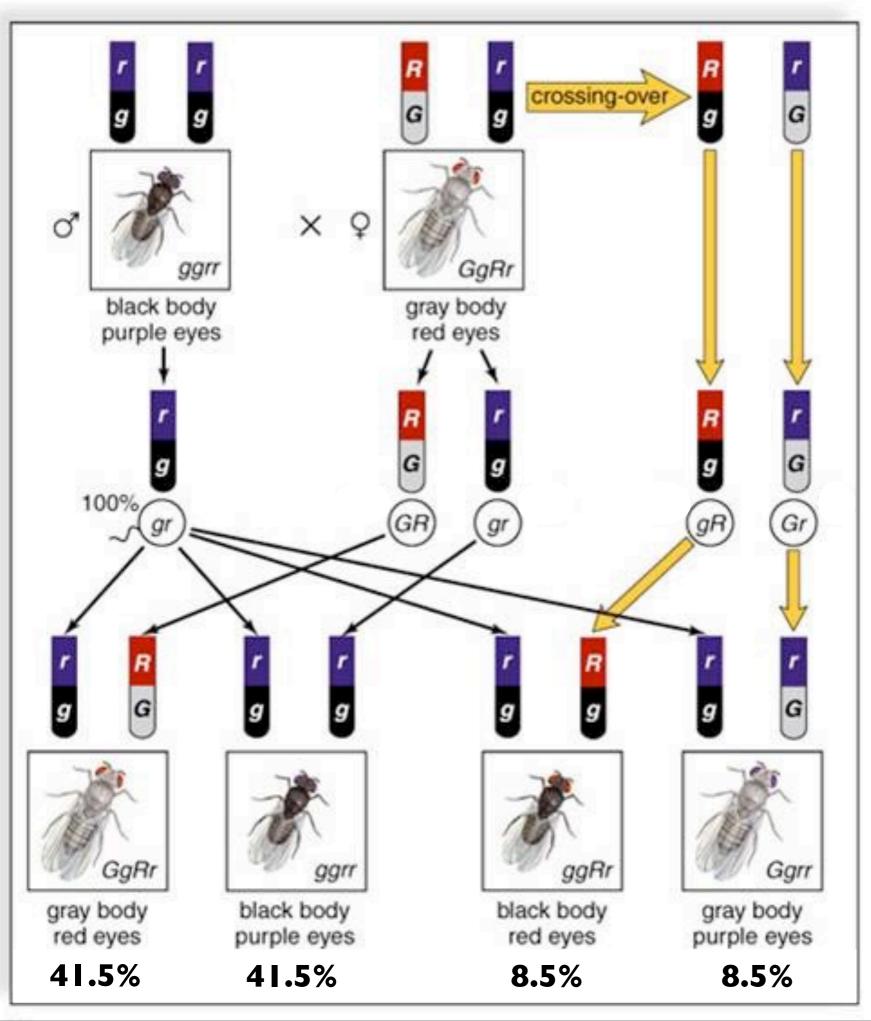
Morgan correctly concluded that the genes were linked! First, most offspring resembled the parents a result expected if genes were linked

Second, the results more resembled a 1:1 ratio also expected if genes were linked.

BUT, How would he explain the unexpected 17% that he called recombinants?

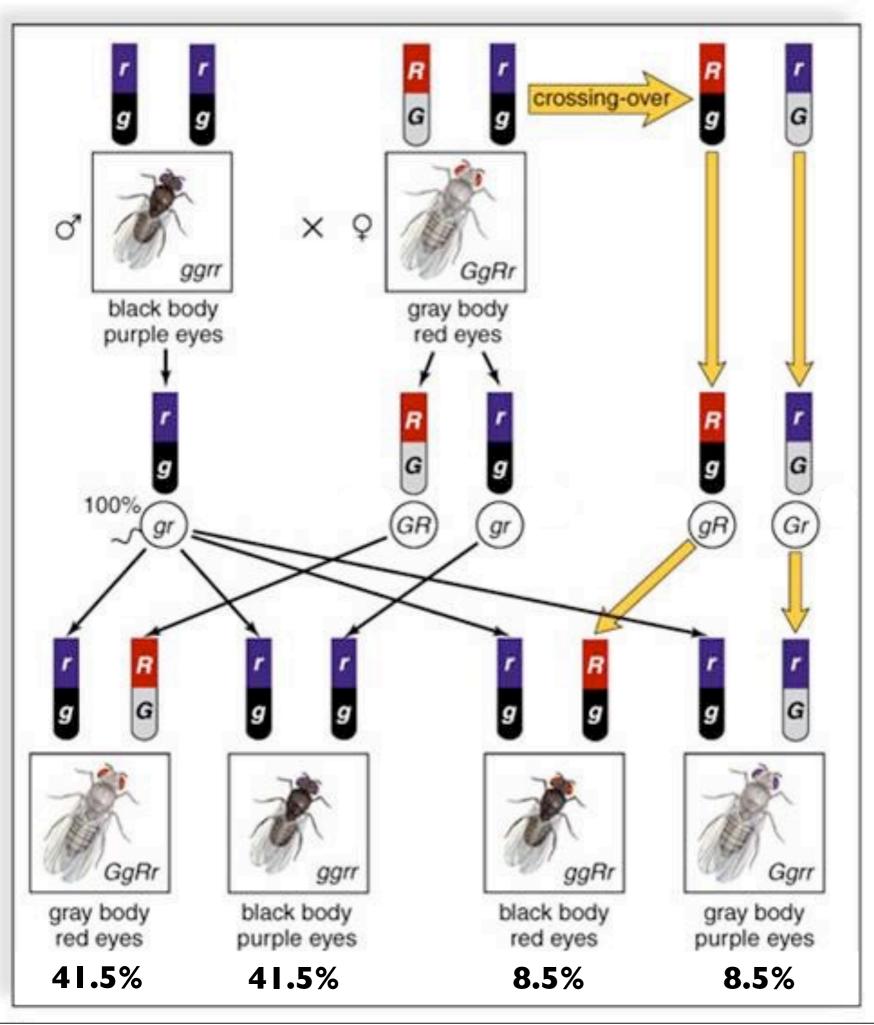
Remember Crossing Over?

- Morgan proposed that some process must occasionally break the physical connection between specific alleles of genes on the same chromosome.
- Subsequent experiments have confirmed and demonstrated this process which is today known as crossing over.
- Crossing would account for the recombination of genes and consequently the 17% recombinant phenotypes in Morgan's experiment.
 - here is what it would look like...



This explanation and these results lead to further questions...

Thursday, February 9, 17



If this experiment is repeated will you always get 17% recombinants? Why 17%? Why not some other %?

Will other linked genes give the same % of recombinants?

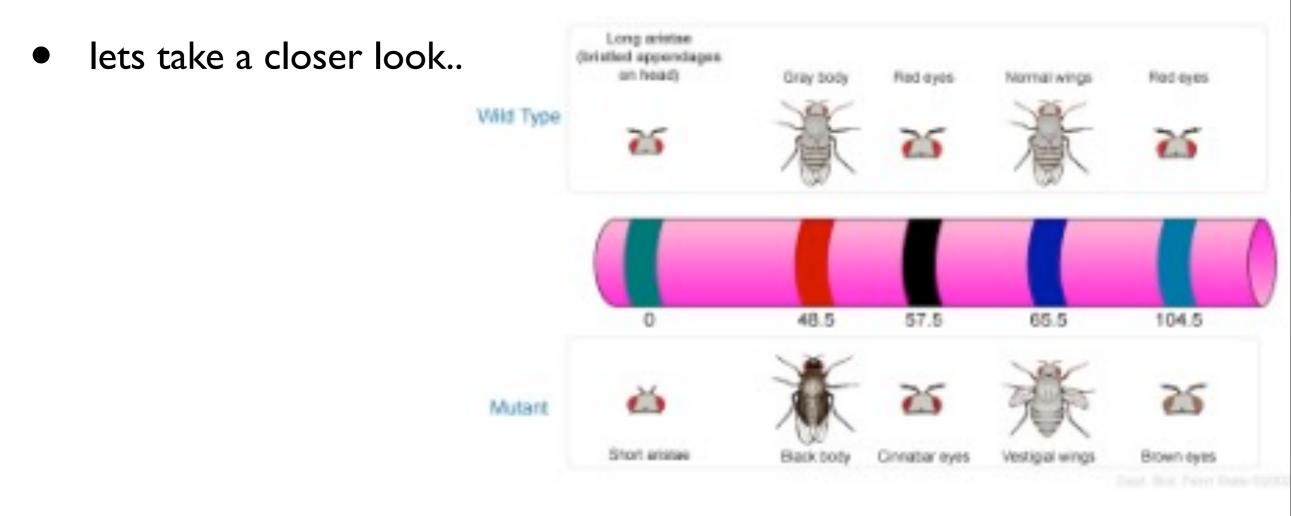
Thursday, February 9, 17

Recombinant Frequency

- The answers to these questions came later from one of Morgan's students none the less.
- Alfred H Sturtevant assumed that crossing over was random and hypothesized that recombination frequency (the 17%) was dependent upon the distance between the two linked genes.
- He reasoned and predicted that the farther apart two genes are, the higher the probability that a crossover will occur between them and therefore the higher the recombination frequency.
 - here is what it would look like...

Gene Mapping

- Using recombinant data from other similar crosses, Sturtevant began assigning relative positions to genes on the same chromosome, he began *mapping genes*.
- A genetic map based upon recombination frequencies is called a linkage map.
- The distance between genes was measured in **map units** which Sturtevant defined as a unit equivalent of 1% recombination frequency.



APPLICATION

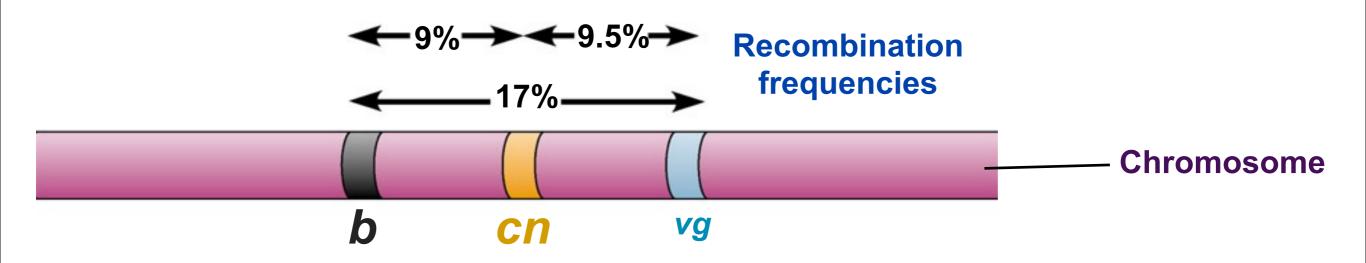
A linkage map shows the relative locations of genes along a chromosome.

TECHNIQUE

A linkage map is based on the assumption that the probability of a crossover between two genetic loci is proportional to the distance separating the loci. The recombination frequencies used to construct a linkage map for a particular chromosome are obtained from experimental crosses, such as the cross depicted in Figure 15.6. The distances between genes are expressed as map units (centimorgans), with one map unit equivalent to a 1% recombination frequency. Genes are arranged on the chromosome in the order that best fits the data.

RESULTS

In this example, the observed recombination frequencies between three *Drosophila* gene pairs (*b*–*cn* 9%, *cn*–*vg* 9.5%, and *b*–*vg* 17%) best fit a linear order in which *cn* is positioned about halfway between the other two genes:



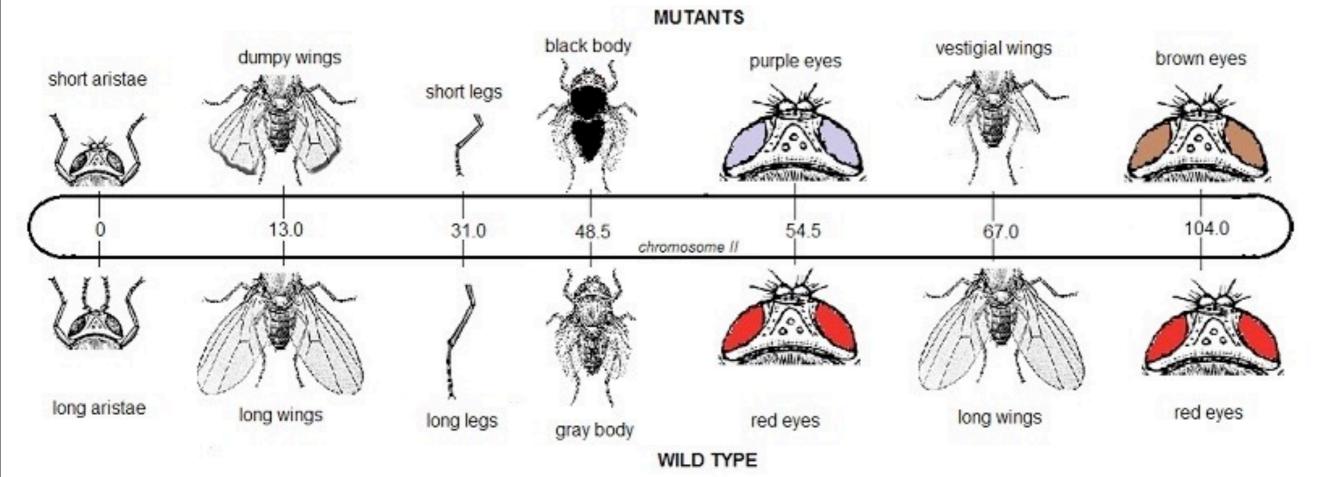
The *b*-*vg* recombination frequency is slightly less than the sum of the b-cn and cn-*vg* frequencies because double crossovers are fairly likely to occur between *b* and *vg* in matings tracking these two genes. A second crossover would "cancel out" the first and thus reduce the observed *b*-*vg* recombination frequency.

Gene Mapping

- In practice gene mapping can be a bit more complicated.
- Recall Morgan's prediction for "unlinked genes".
 - He predicted a I:I:I:I ratio (25% of each phenotype)
 - The prediction also stated that 50% of the offspring would have a different phenotype than the parents, in other words 50% would be recombinants.

Gene Mapping

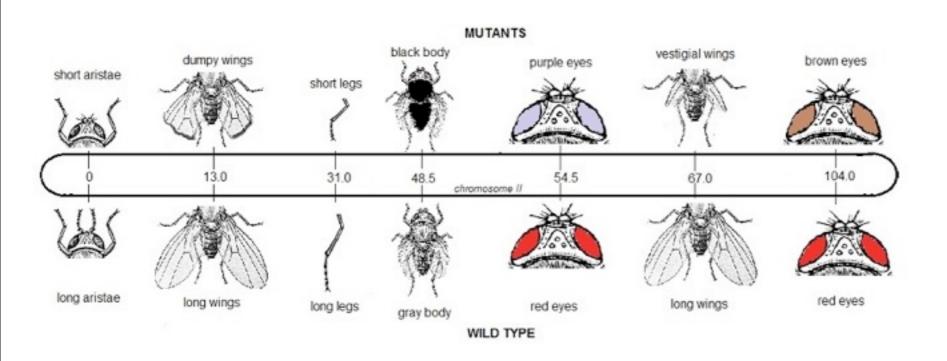
- A problem arises because two genes far apart are virtually guaranteed to cross over resulting in 50% recombination frequency.
- SO... When we get 50% recombinants we can not tell if they are assorting independently on different chromosomes OR whether they are far apart on the same chromosome.



Notice **body color** and **eye color** are more 50 map units away from each other thus your breeding experiment looked at these two traits you would 50% recombinants and could not tell if they are assorting independently on different chromosomes OR whether they are far apart on the same chromosome. However, if you cross **eye color** and **wing length** then you would get 37% recombinants, evidence that the genes controlling these traits are on the same chromosome, they are LINKED.

Now cross **body color** and **wing length**, you would get 18.5%

recombinants, evidence that the genes controlling these traits are on the same chromosome, they are LINKED.



IF the genes for eye color and wing length are linked AND the genes for wing length and body color are linked THEN the genes controlling eye color and body color are on the same chromosome, they are LINKED. Essential knowledge 3.A.3: The chromosomal basis of inheritance provides an understanding of the pattern of passage (transmission) of genes from parent to offspring.

b. Segregation and independent assortment of chromosomes result in genetic variation.

Evidence of student learning is a demonstrated understanding of each of the following:

3. The pattern of inheritance (monohybrid, dihybrid, sex-linked, and genes linked on the same homologous chromosome) can often be predicted from data that gives the parent genotype/ phenotype and/or the offspring phenotypes/genotypes.

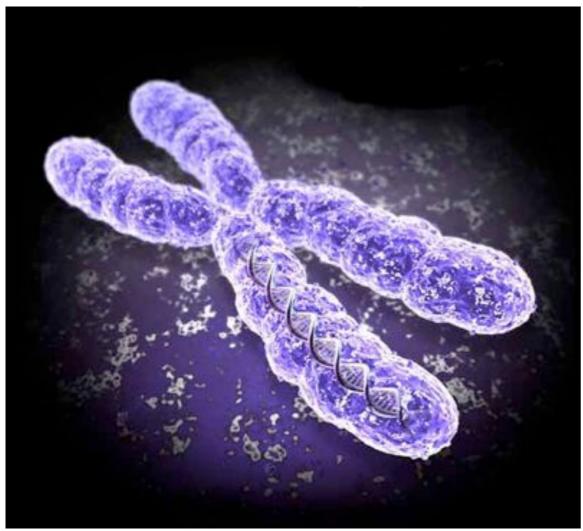
PREFACE

- I would argue that mankind has a fundamental understanding and even an innate interest heredity.
- For centuries humans have observed a dichotomy that exists in sexual reproduction of offspring...
 - Each offspring is both unique and yet at the same time exhibits identical traits found in its parents.
- For years many explained heredity by the "blending" hypothesis, an idea that each parent donated genetic material that would blend like two color paints.
- However, everyday observations and breeding results contradict the predictions if this

- The alternative to the blending hypothesis is the particulate hypothesis, where parents pass on discrete units of hereditary information that retain their identity in the offspring.
 - like dealing cards from deck of cards rather than mixing two colors of paint
- The conformation of this idea and consequently the foundation of all genetic understanding began in an abbey garden by a monk studying the common pea plant.
- Ironically, this monk, (Gregor Mendel) laid down the foundation of genetic principles before anyone knew about DNA, genes, chromosomes and meiosis.

Mendelian Genetics

Main Idea: Mendel discovered the basic principles of heredity through carefully planned experiments, meticulous data collection / analysis and a little luck.



Historical Mendel

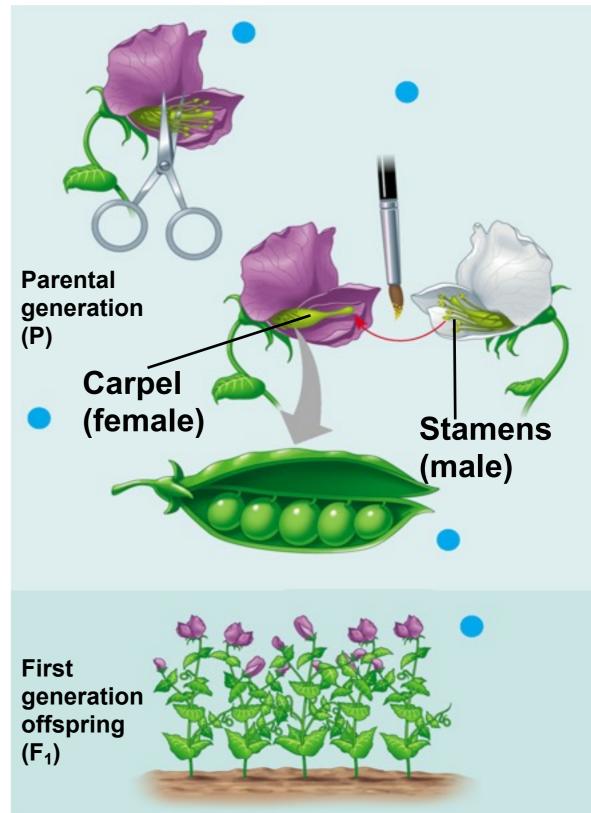
- Born in Austria
- Grew up on parent's farm
- Had agricultural training
- Overcame financial hardship/illness
- Excelled in high school
- Attended Olmutz Philosophical Institute
- Failed exam to become a teacher
- Entered Augustinian monastery at age 21
- Left monastery at 29 to study physics and chemistry at University of Vienna
- Two particular professors had a profound influence on Mendel



- One professor, a physicists, emphasized learning science through experimentation and mathematics
- The other professor, a botanist, sparked Mendel's curiosity in plant heredity
- Returns to the monastery, Teaches at a local school
- Other teaching monks also interested in breeding plants
- In 1857 at the age of 35 Mendel begins breeding pea plants

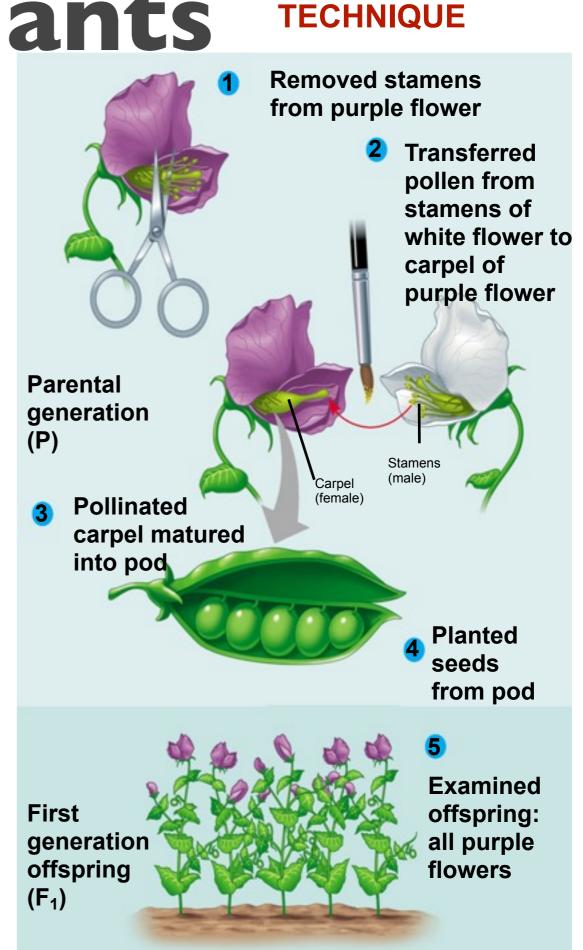
Choosing Pea Plants

- Pea plants are a wise choice for genetic studies:
 - I. many varieties
 - 2. short generation time
 - 3. numerous offspring
 - 4. easy to control mating
 - 5. easy to count new seeds



APPLICATION By crossing (mating) two truebreeding varieties of an organism, scientists can study patterns of inheritance. In this example, Mendel crossed pea plants that varied in flower color.

RESULTS When pollen from a white flower fertilizes eggs of a purple flower, the first-generation hybrids all have purple flowers. The result is the same for the reciprocal cross, the transfer of pollen from purple flowers to white flowers.



Additional important information about Mendel's Crosses:

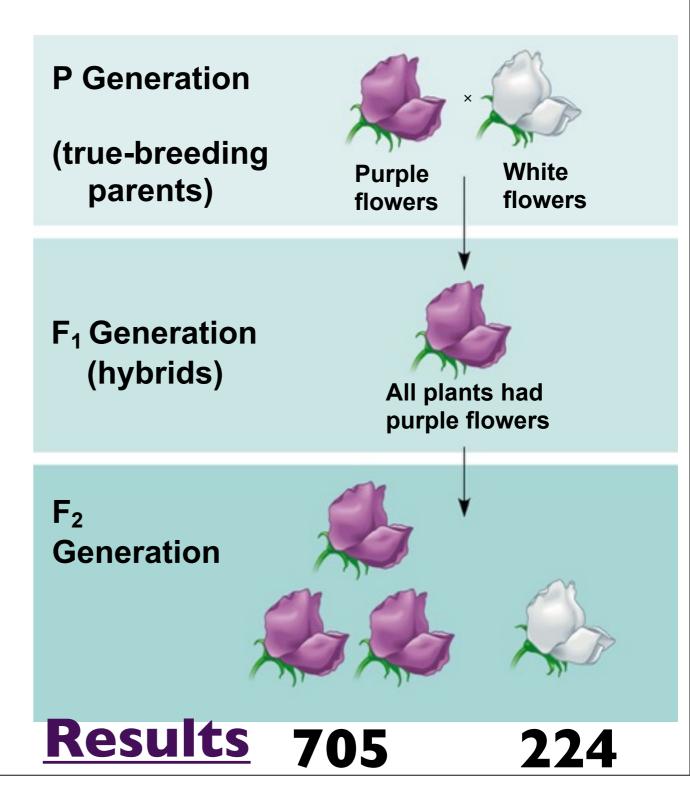
- I. He chose to track traits that occurred in only two distinct varieties, like flower color (purple or white)
 - turns out this decision was both fortuitous and lucky as you will learn later

- 2. He painstakingly produced plants that he called "true breeding" meaning if they self fertilized they would always produce the same trait as the parent plant, in other words purple flower plants always produced purple flower plants.
- 3. His typical experiments involved mating two different "true breeding" varieties in what he called "hybridization" and then analyzing the offspring in the further generations.
 - he made another fortuitous decision to track the trait into 2 or more generations, not just a single generation.

 Below is a classic Mendelian Cross that illustrates the points made on the last slide(s):

Experiment

True-breeding purple-flowered pea plants and white-flowered pea plants were crossed (symbolized by \times). The resulting F₁ hybrids were allowed to selfpollinate or were cross-pollinated with other F₁ hybrids. Flower color was then observed in the F₂ generation.



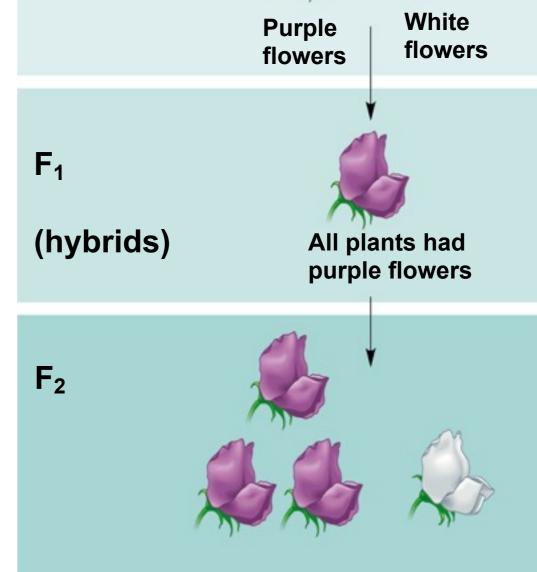
 This cross and many just like it, provided strong evidence against the blending hypothesis of inheritance.

If the blending hypothesis were correct then Mendel would expect the F1 hybrids to be purple & white OR lavender in color.

When white colored flowers reappear in the F2 generation, Mendel knew that the "heritable factor" had not been diluted or destroyed.

Mendel reasoned that the white trait was hidden or masked in someway, he called this trait the *recessive trait* and called the purple trait the *dominant trait*.

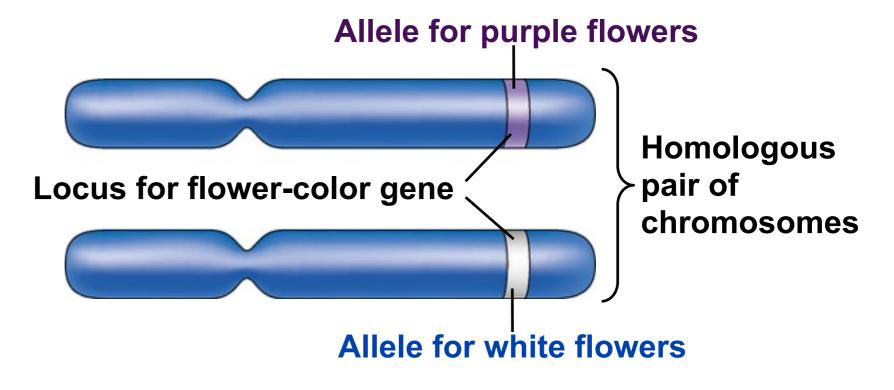




Character	Dominant Trait	×	Recessive Trait	F ₂ Generation Dominant:Recessive	Ratio	
Flower color	Purple	×	White	705:224	3.15:1	
Flower position	Axial	×	Terminal	651:207	3.14:1	
Seed color	Yellow	×	Green	6022:2001	3.01:1	
Seed shape	Round	×	Wrinkled	5474:1850	2.96:1	
Pod shape	Inflated	×	Constricted	882:299	2.95:1	
Pod color	Green	×	Yellow	428:152	2.82:1	
Stem length	Tall	×	Dwarf	787:277	2.84:1	

- Mendel developed a model to explain this pattern of inheritance in the FI and F2 generations.
 - **First:** Alternative versions of "heritable factors" account for variations in inherited characters.
 - NOTE to students Mendel never knew of genes but today we know his "heritable factors" are **genes** and the alternate forms of genes are called **alleles** as such I will use these terms exclusively from this point on knowing of course that Mendel did not use terms himself!

Today we can relate this idea to chromosomes and DNA



- Mendel developed a model to explain this pattern of inheritance in the FI and F2 generations.
 - Second: For each character, an organism inherits two copies of a gene, one from each parent.
 - The two alleles may be identical or they may be different!
 - Identical alleles are today referred to as a homozygous genotype, Mendel used the term "true breeding"
 - Different alleles are today referred to as a heterozygous genotype, Mendel used the term "hybrid"

- Mendel developed a model to explain this pattern of inheritance in the FI and F2 generations.
 - **Third:** If the two alleles at a locus differ, then one, the **dominant allele**, determines the organism's appearance; the other, the **recessive allele**, has no noticeable effect on the organism's appearance.
 - Capital letters often symbolize dominant alleles (A) while lower case letters often represent the recessive alleles (a).

```
Letters are used
a symbols to
represent the
alleles that we
can see with the
naked eye.
```

- Mendel developed a model to explain this pattern of inheritance in the FI and F2 generations.
 - Fourth: The Law of Segregation states that two alleles for a heritable character separate from each other during gamete formation and end up in different gametes.
 - Thus sperm and eggs only carry one allele/gene.
 - If an organism is true breeding then every gamete will carry the same allele however if the organism is a hybrid the 50% of the gametes will carry one allele while the other 50% carry the other allele.

Each true-breeding plant of the parental generation has identical alleles, *PP* or *pp*.

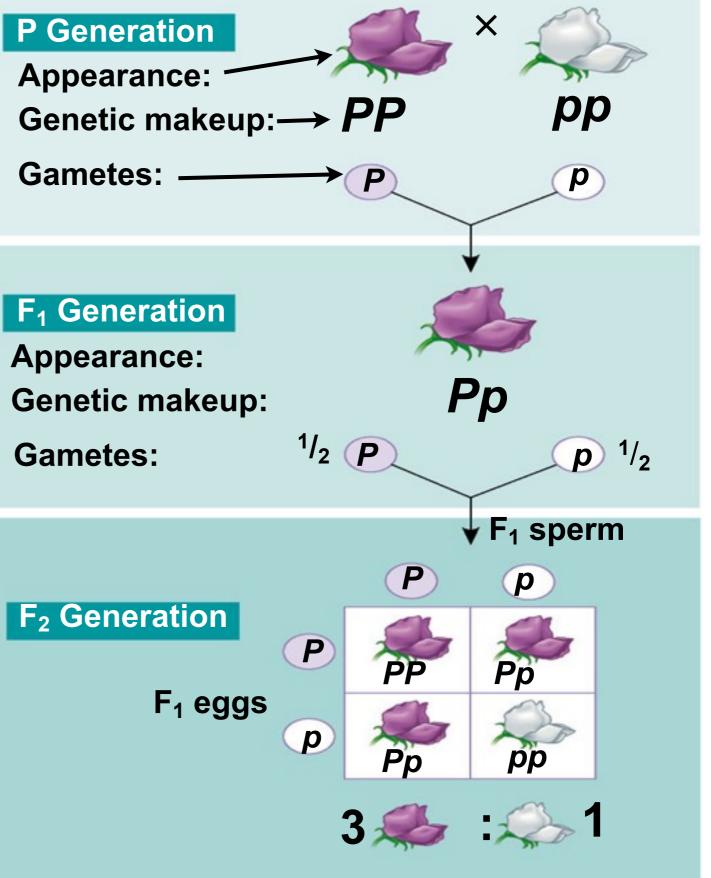
Gametes (circles) each contain only one allele for the flower-color gene. In this case, every gamete produced by one parent has the same allele.

Union of the parental gametes produces F_1 hybrids having a *Pp* combination. Because the purple-flower allele is dominant, all these hybrids have purple flowers.

When the hybrid plants produce gametes, the two alleles segregate, half the gametes receiving the *P* allele and the other half the *p* allele.

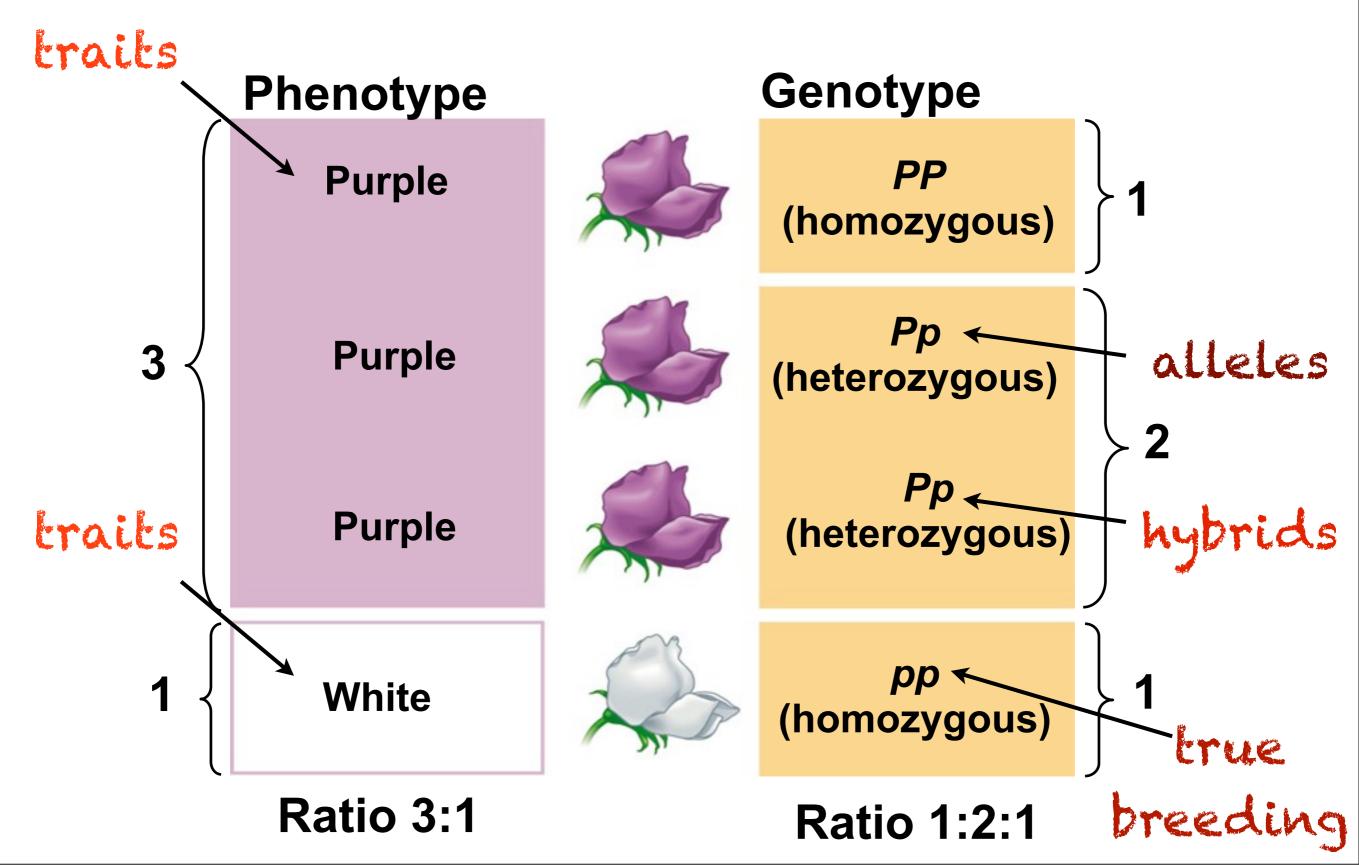
This box, a Punnett square, shows all possible combinations of alleles in offspring that result from an $F_1 \times F_1$ ($Pp \times Pp$) cross. Each square represents an equally probable product of fertilization. For example, the bottom left box shows the genetic combination resulting from a p egg fertilized by a P sperm.

Random combination of the gametes results in the 3:1 ratio that Mendel observed in the F_2 generation.



- Notice that Mendel's Model lends itself to specific expected results or predictions.
- How do we make conclusions in science?
 - We compare the actual results and the expected results, when the two results are concur then we have support for the hypothesis!
- Does the data and results support Mendel's model?
 - Yes, absolutely!

Genetic Vocabulary



The Punnet Square

- A tool used to predict (<u>future</u>) possible allele compositions of offspring from a cross between parents whose genetic make up is known.
- If we know: Mom is Aa and Dad is Aa
- And we remember that gametes carry only one allele.
- **Then** we can predict possible allele combinations in their offspring using a punnet square.

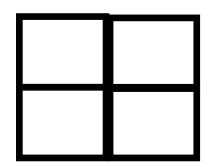
 A tool used to predict possible allele compositions of offspring from a cross between parents whose genetic make up is known.

If we know: Mom is Aa and Dad is Aa

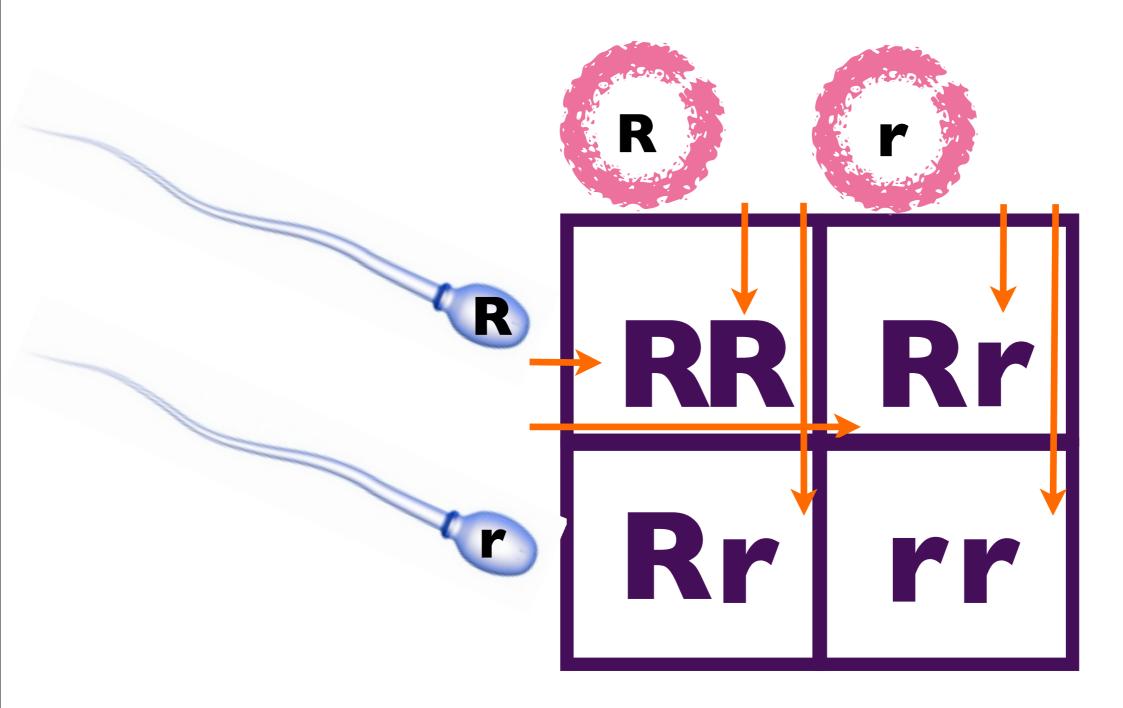
And we remember that gametes carry only one allele.



Then we can predict possible allele combinations in their offspring using a punnet square.



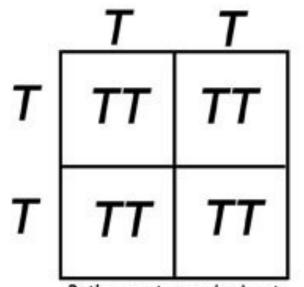
1.) What are the possible sperm?
2.) What are the possible eggs?
3.) What are the possible fertilizations?
4.) What are the possible offspring?



Punnett's Squares

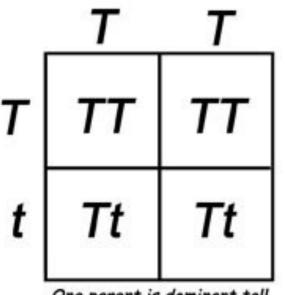
These show the 2 alleles of each parent plant crossed with each other and the resulting 4 possible offspring with T = tall, t = short. TT = dominant tall, tt = recessive short, Tt = mixed hybrid

> TT = dominant tall (genotype tall, phenotype tall) Tt = mixed hybrid (genotype hybrid, phenotype tall) tt = recessive short (genotype short, phenotype short)



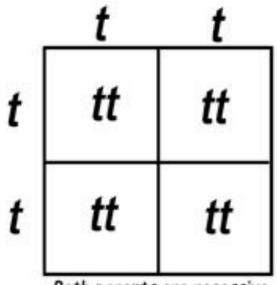
 $\begin{array}{c|c} T & t \\ \hline T & TT & Tt \\ t & Tt & tt \\ \end{array}$

Both parents are dominant tall so all offspring are tall.



One parent is dominant tall and one is mixed hybrid so all offspring are tall.

Both parents are mixed hybrids so offspring are a 3:1 ratio.



Both parents are recessive short so all offspring are short.

What are the parents in cross? What does the single "T or t" on the outside of the punnet square represent?

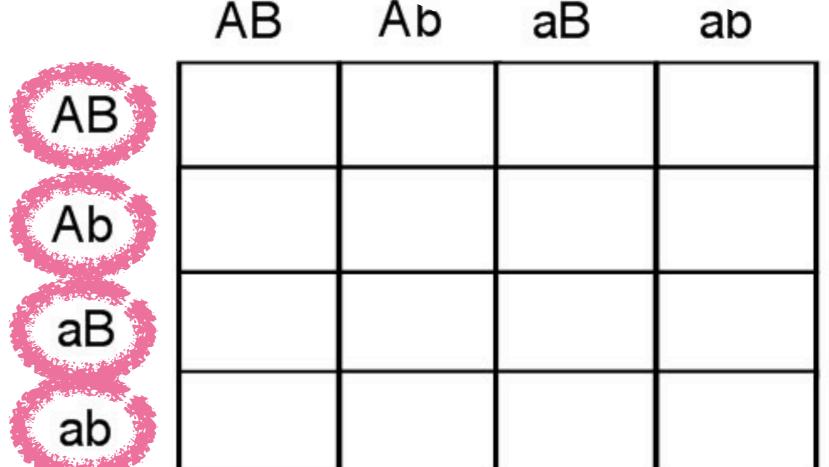
What are the genotypic ratios of each? Phenotypic?

Which punnet square shows a monohybrid cross?

The Punnet Square

- We can use the punnet square to track multiple alleles at the same time.
- What **If** we know: **Mom is AaBb** and **Dad is AaBb**

You must remember that every gamete must have one of each allele



The Punnet Square

- We can use the punnet square to track multiple alleles at the same time.
- What **If** we know: **Mom is AaBb** and **Dad is AaBb**

What is the phenotypic ratio?	possible sperm				
9:3:3:1		AB	Ab	aB	ab
Genotypic?					
4.7.7.7.7.1.1.1	AB	AABB	AABb	AaBB	AaBb
60 60 0	Ab	AABb	Aabb	AaBb	Aabb
possible	aB	AaBB	AaBb	aaBB	aaBb
ď	ab	AaBb	Aabb	aaBb	aabb

Mendel's Model Continues

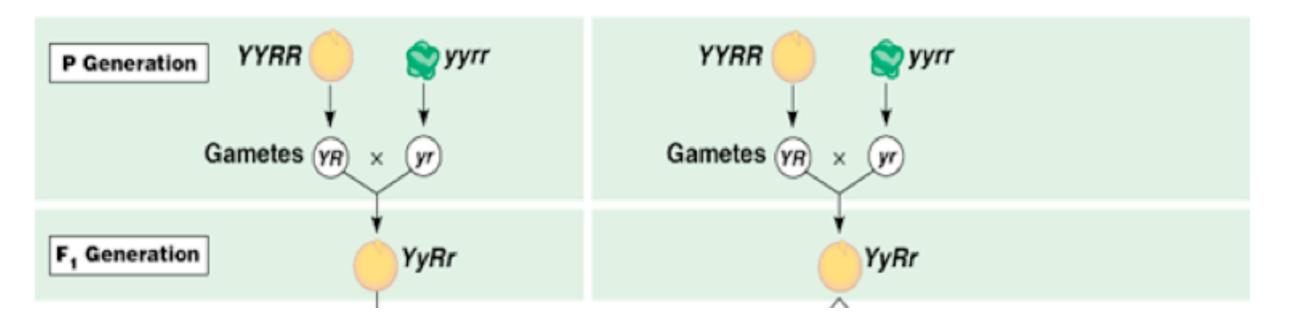
• Mendel also looked at two traits at one time.

- However asked himself the follow question, a question that you should have asked yourself on the last slide.
- Do the "a" alleles and the "b" alleles travel separately or as a package?
- In modern terms: Do the "a" alleles and the "b" alleles travel on the same chromosome or on different chromosomes?

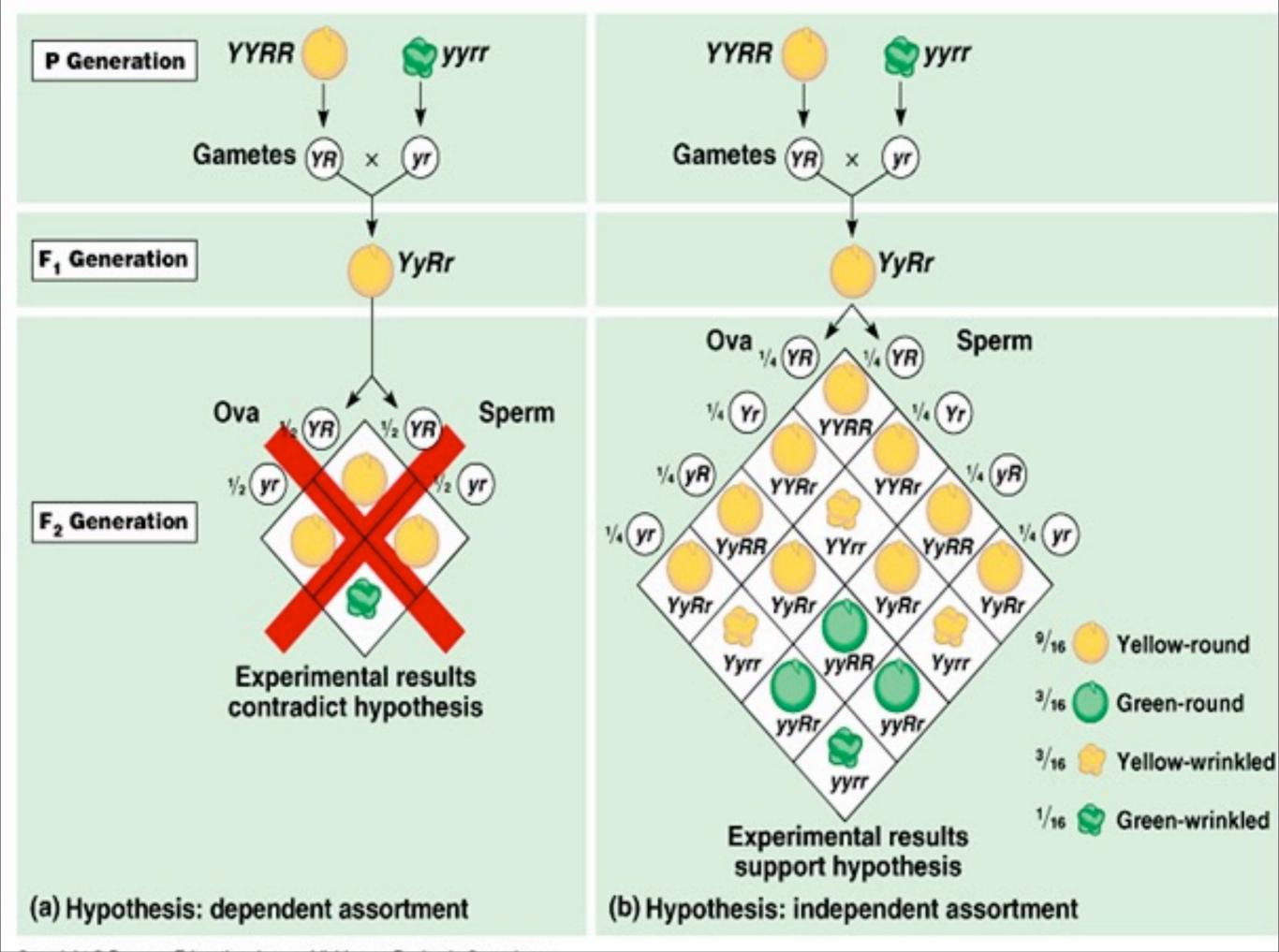
The answer to this question very much matters and as you will see it led Mendel to his 2nd Law of Inheritance!

Mendel's Model Continues

 Mendel crossed two plants both true breeding. The first plant was a true plant with Yellow, Smooth seeds and the other true breeding plant produced Green, wrinkled seeds.

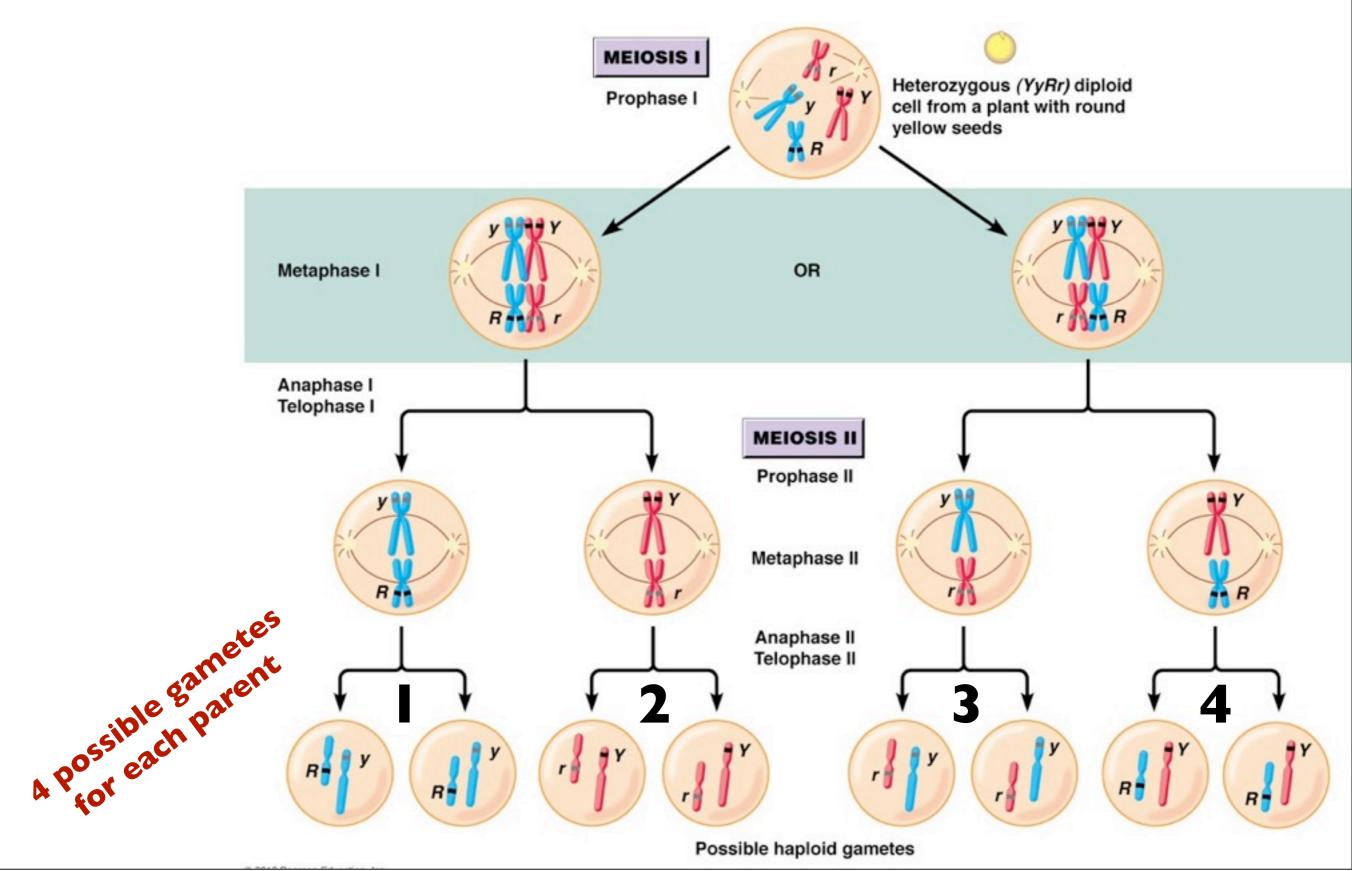


- Mendel realized and so should you that the F1 hybrids can only produce Yellow, smooth seeds.
- The F2 generation is were it gets interesting!

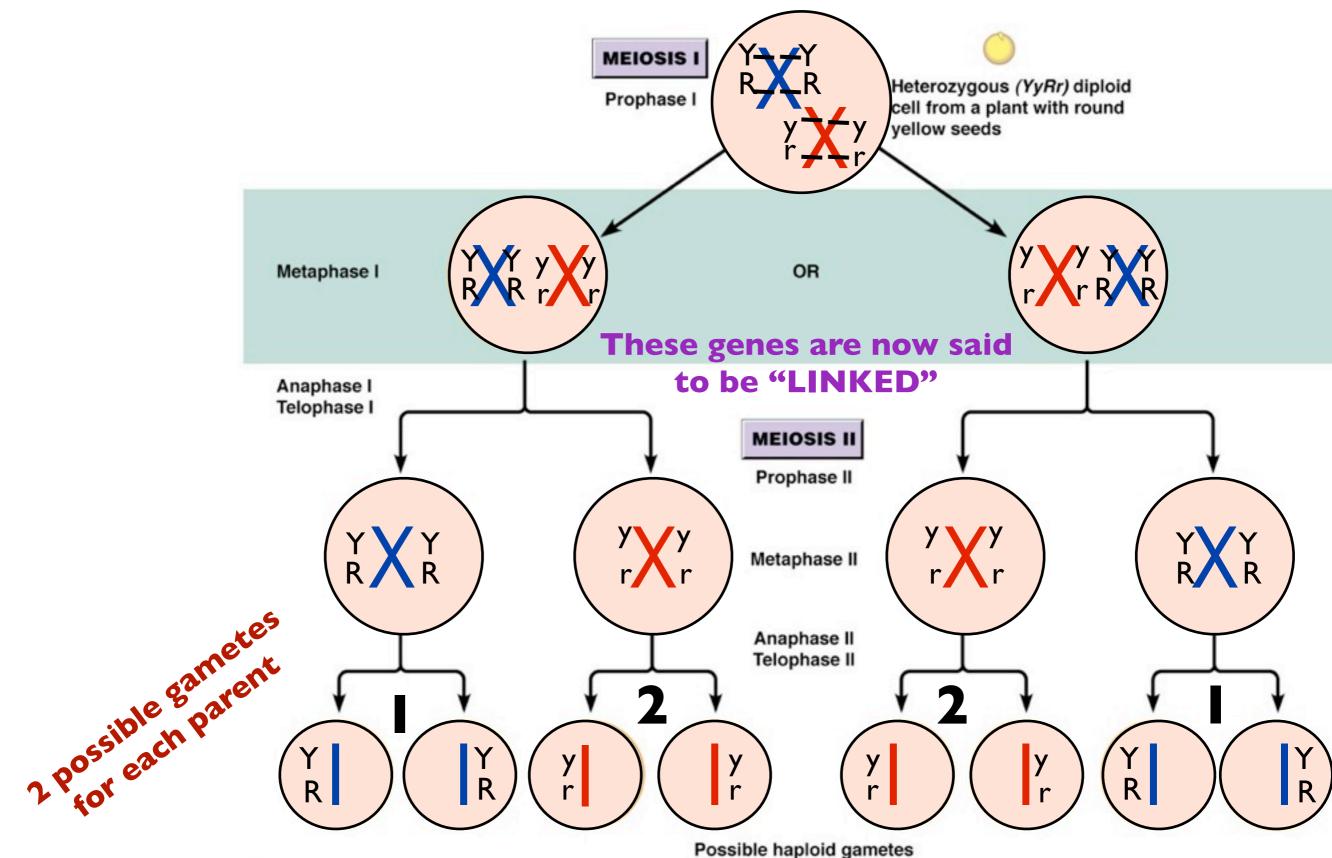


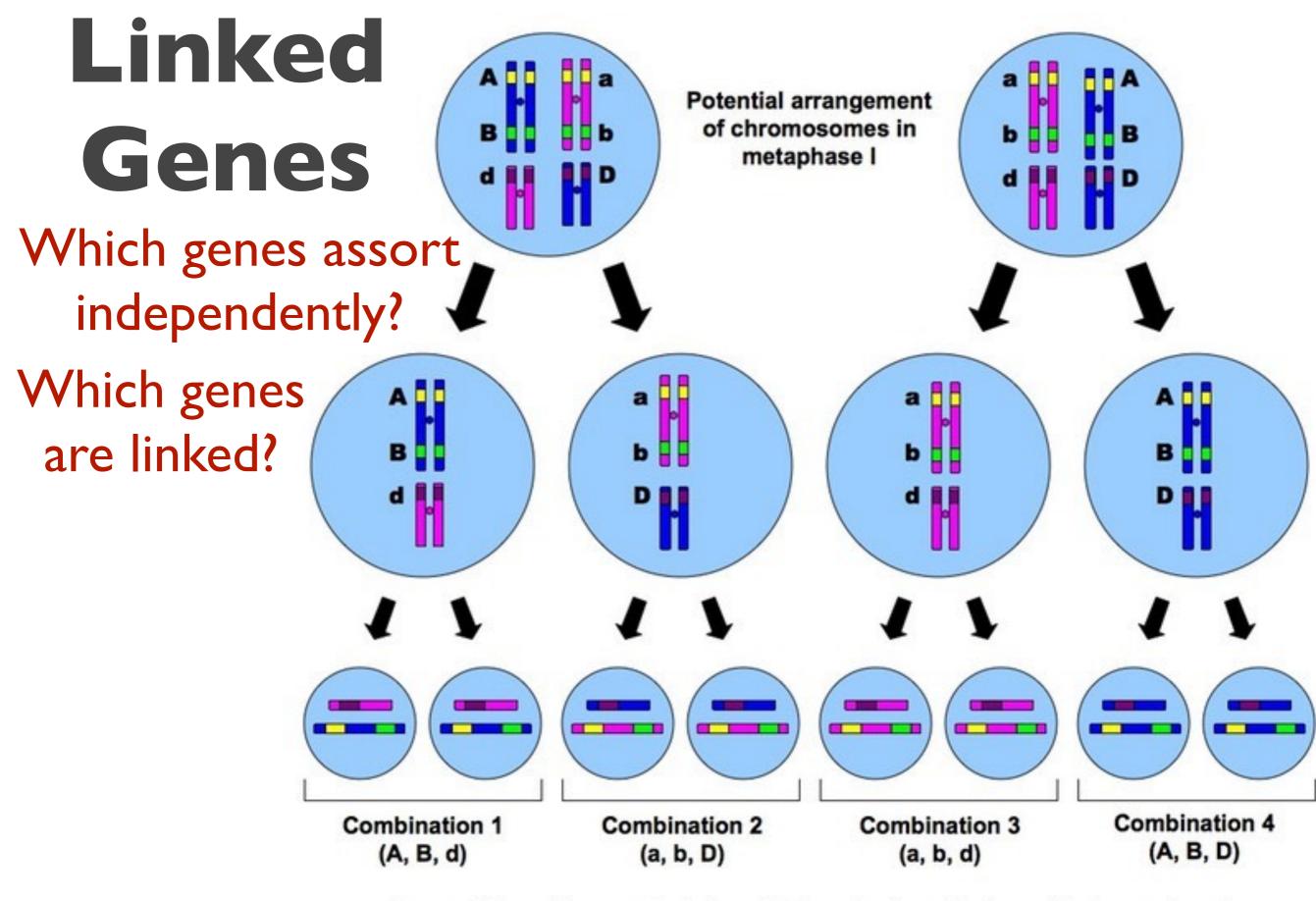
Thursday, February 9, 17

Mendel's results are supported when the different alleles (a and b) are carried on different chromosomes.



Now redraw the formation of gametes, put the different alleles on the same chromosome.





Genes A/B and D are unlinked and follow the law of independent assortment

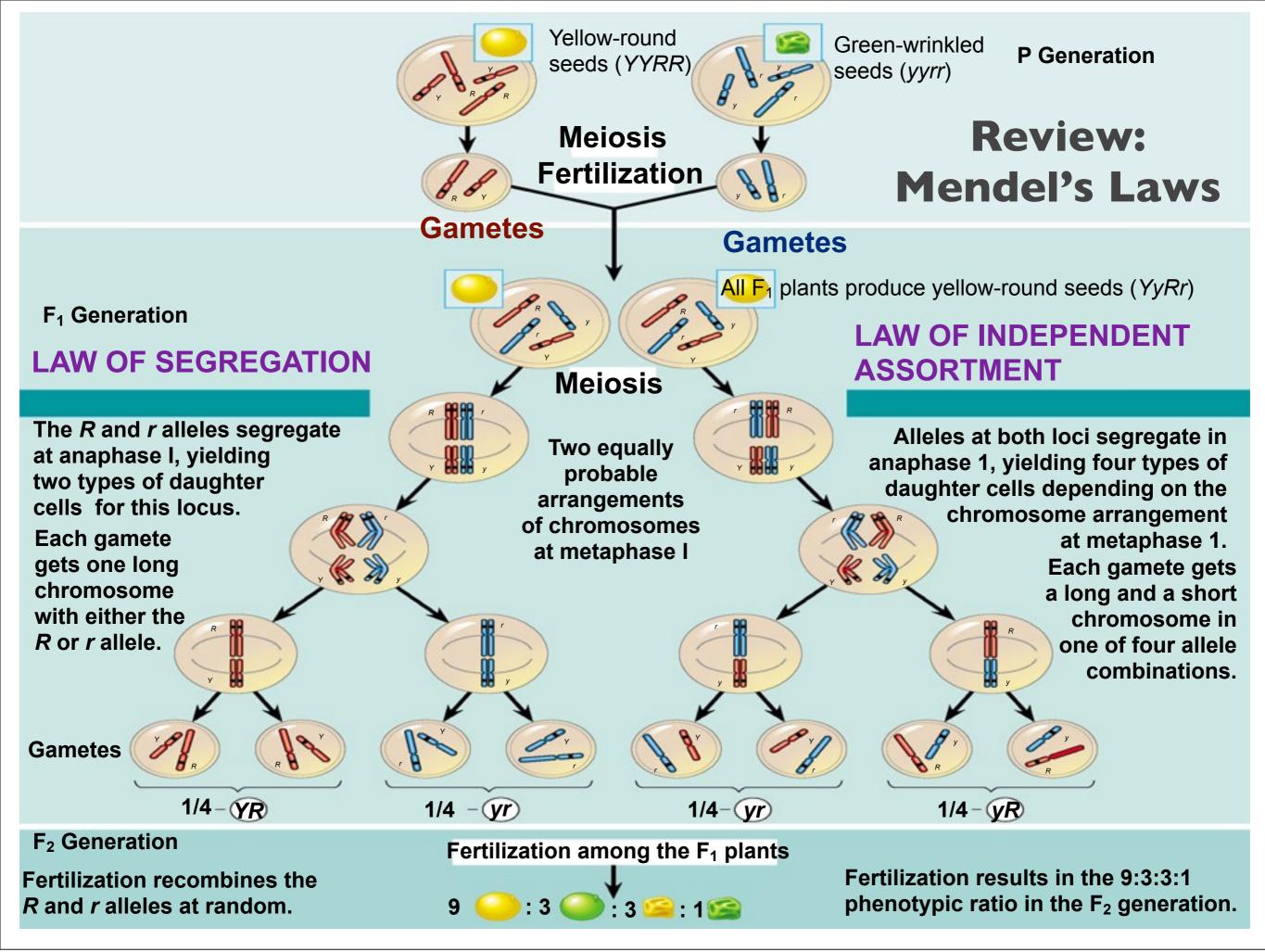
Genes A and B are linked and do not follow the law of independent assortment

Mendel's Model Continues

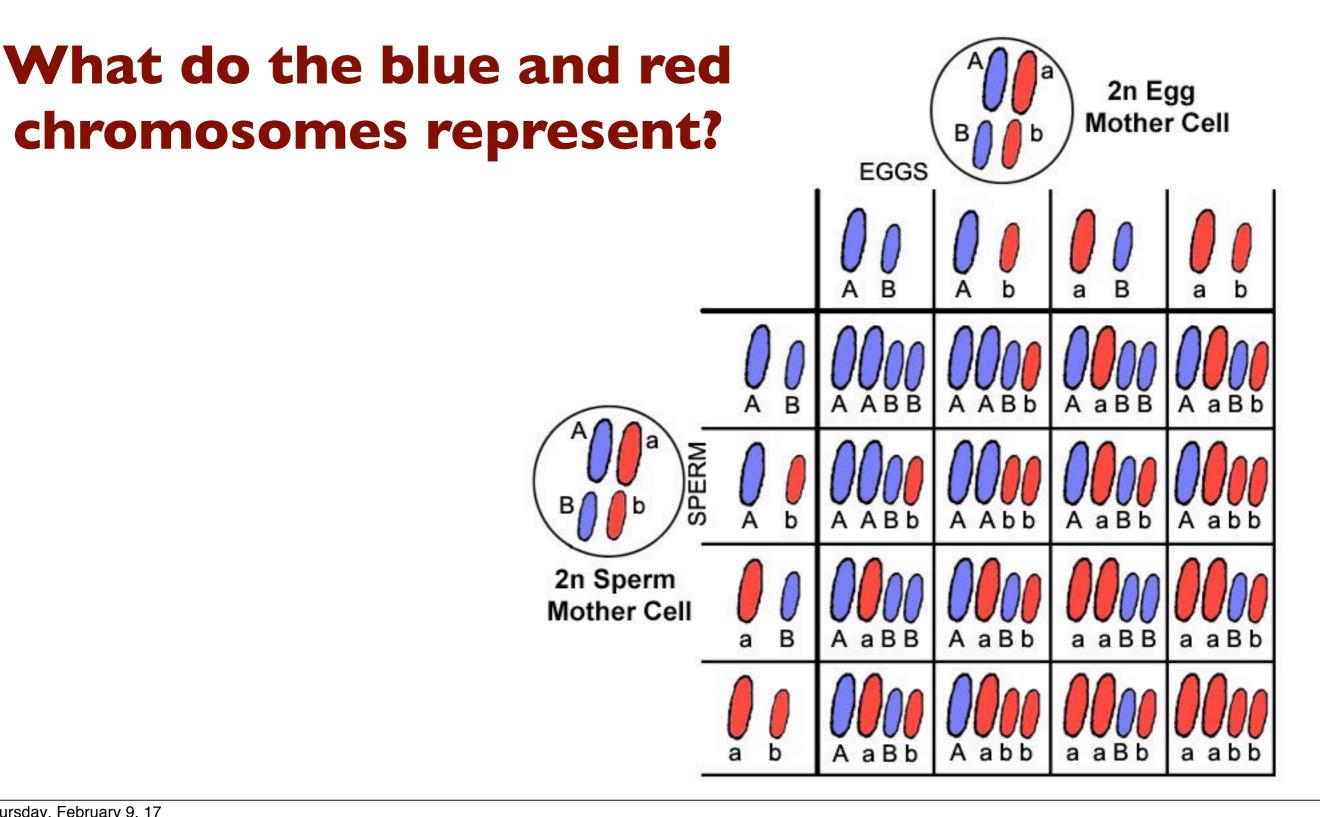
- From this Dihybrid cross, Mendel formed his 2nd Law of Inheritance.
- The **Law of Independent Assortment** states that each pair of alleles separate independently of each other pair of alleles during gamete formation.

PLEASE NOTE: This law only applies to allele pairs that are located on different chromosomes.

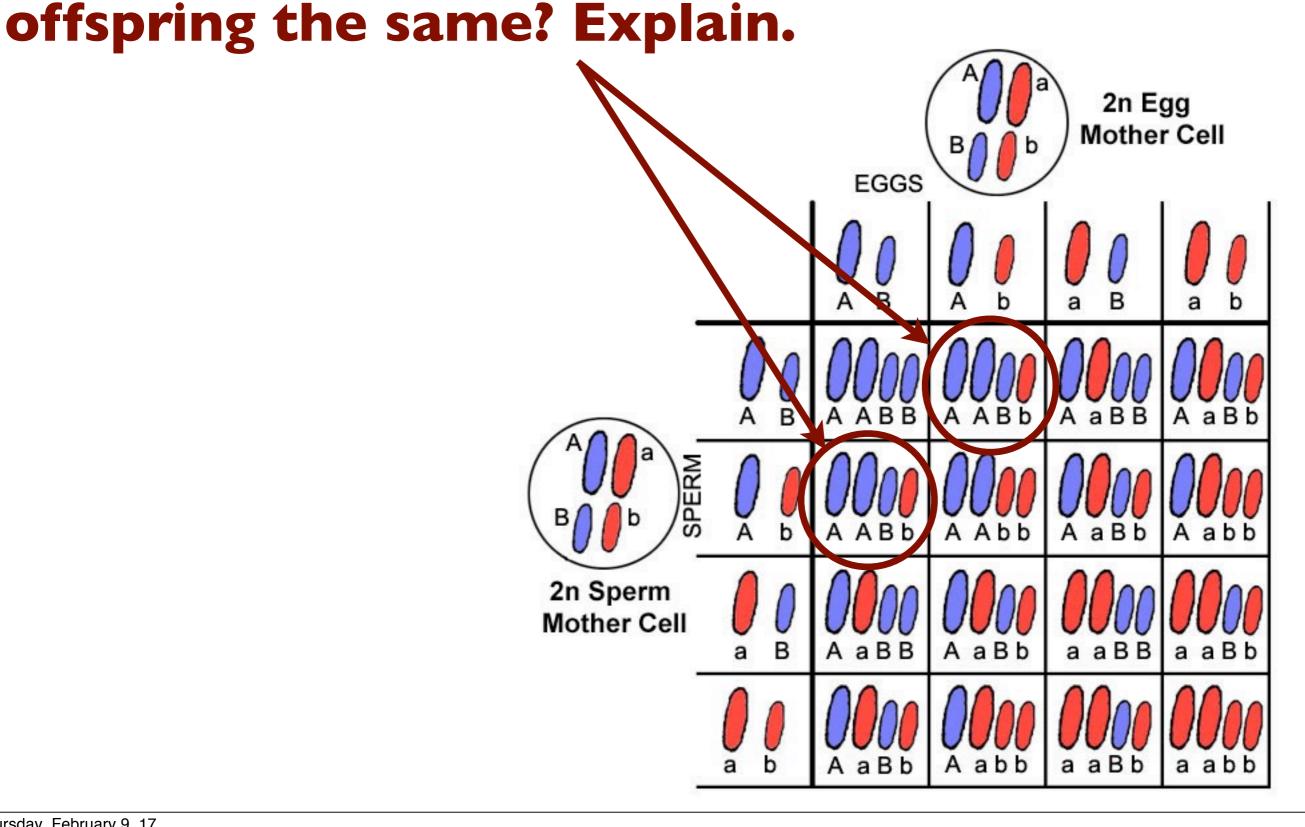
Earlier I said that Mendel was a little lucky. He was able to generate this law because every time he tracked two different allele pairs they happen to be on different chromosomes. Do think every allele pair has its own chromosome? I hope not! What would happen if Mendel had picked two allele pairs on the same chromosome? (rhetorical)

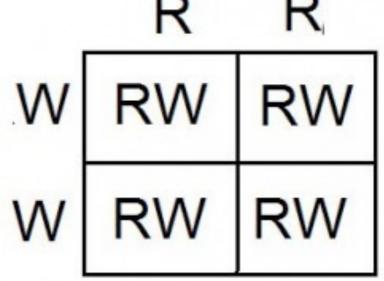


... Back to Punnet Squares If we know: Mom is AaBb and Dad is AaBb

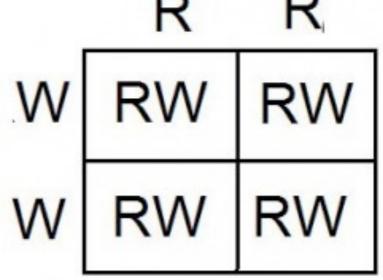


If we know: Mom is AaBb and Dad is AaBb Are these two possible



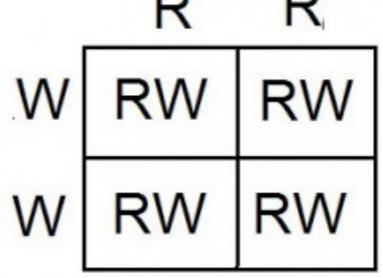






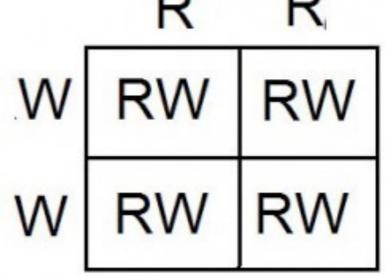


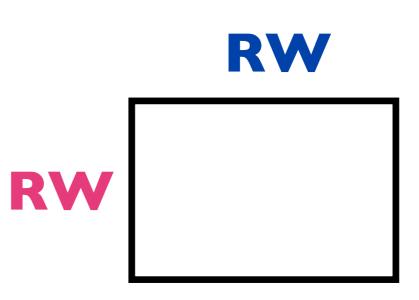




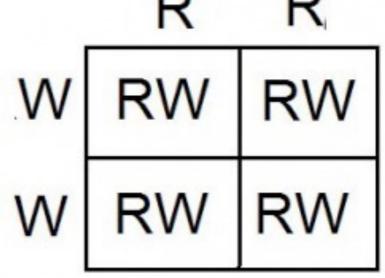


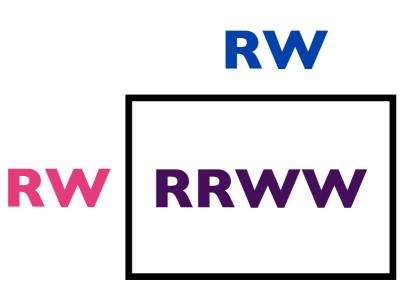






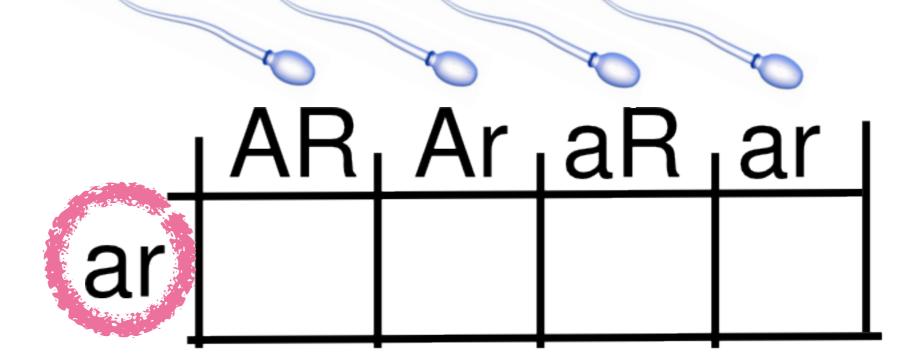
Can you fix it?



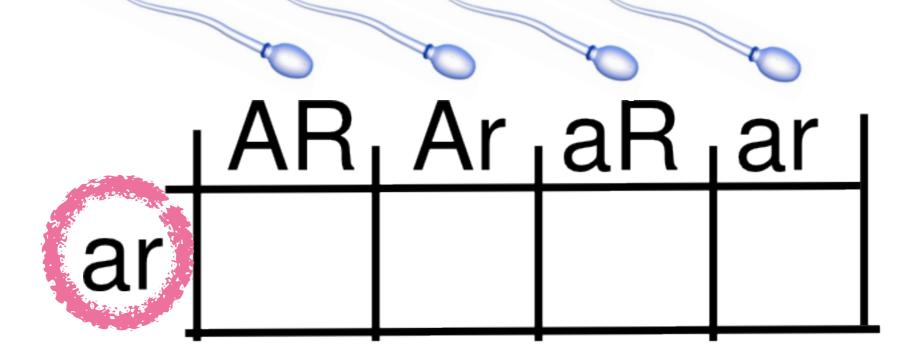


Can you fix it?

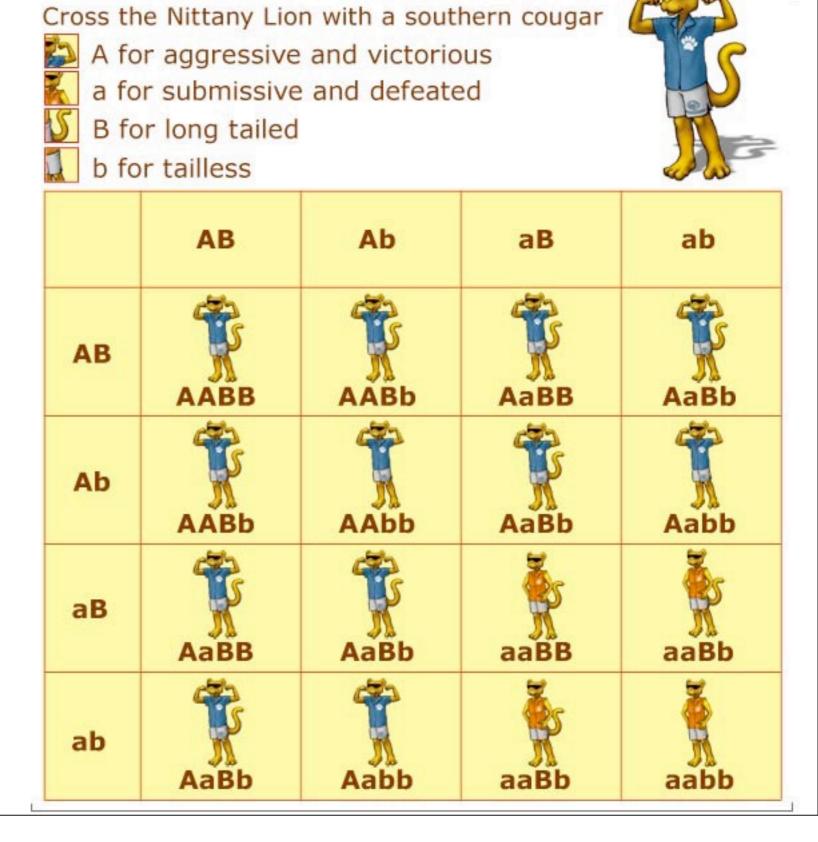
What are the parent's genotypes?

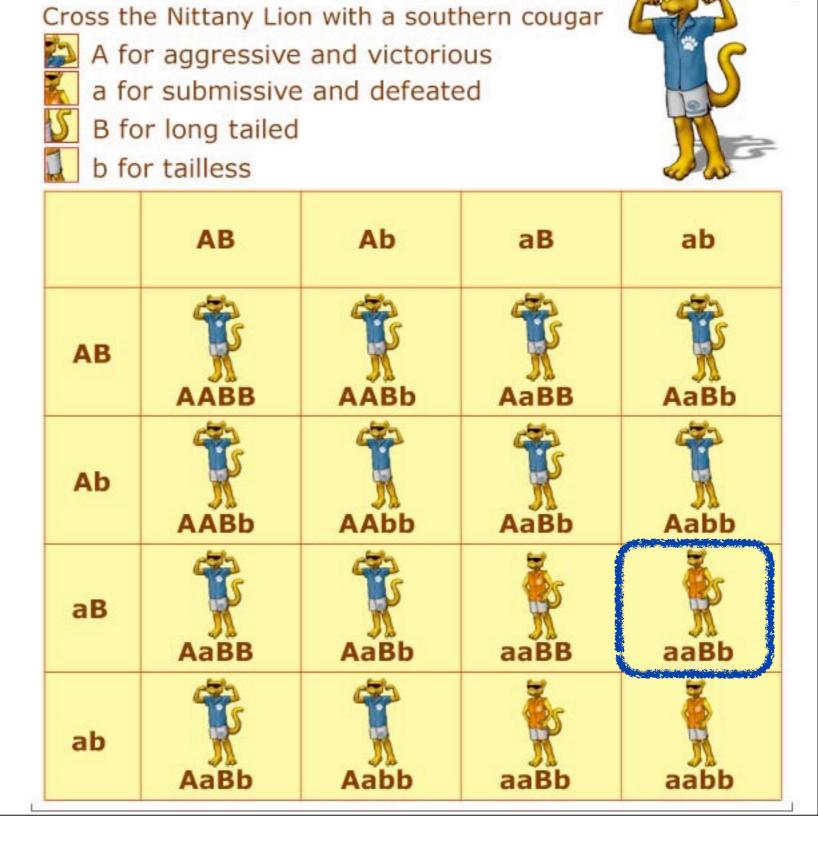


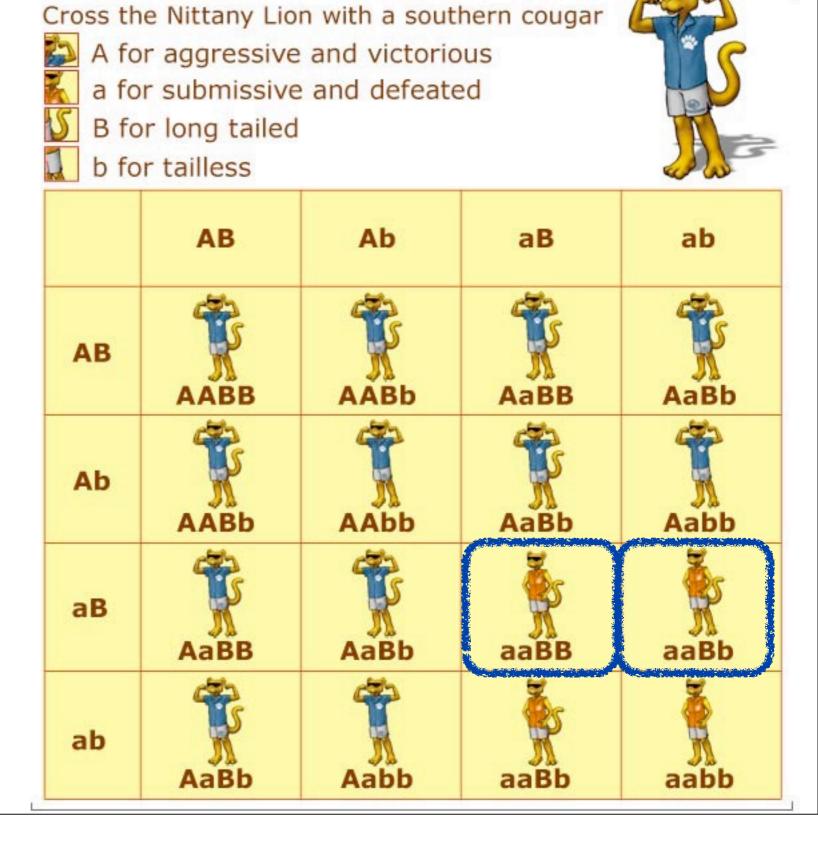
What are the parent's genotypes?

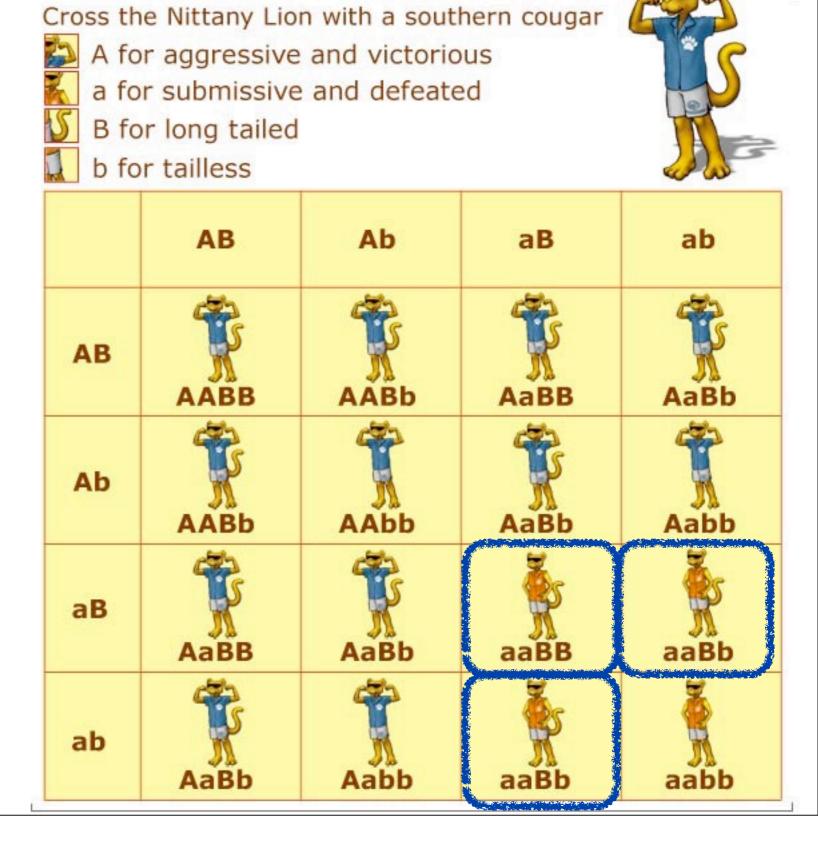


Mom is aarr and Dad is AaRr



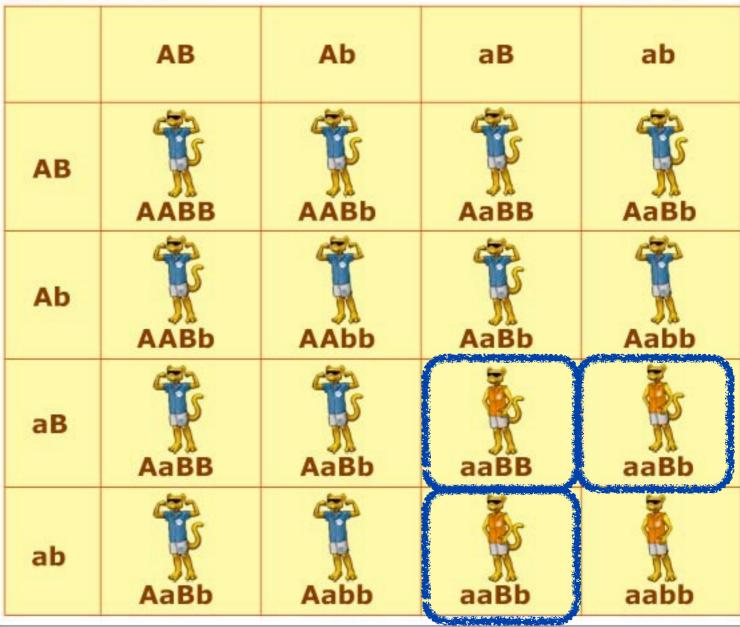






3/16 or 18.75%

Cross the Nittany Lion with a southern cougar A for aggressive and victorious a for submissive and defeated B for long tailed b for tailless



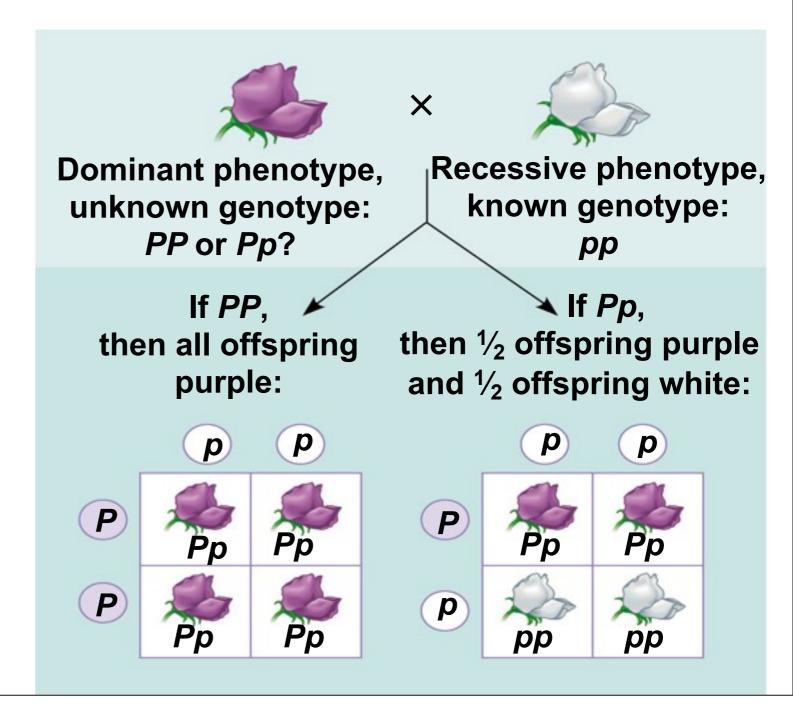
3/16 or 18.75%

Now, can you do it without the pictures?

Dihybrid Cross Cross the Nittany Lion with a southern cougar A for aggressive and victorious a for submissive and defeated B for long tailed b for tailless Ab AB ab aB AB AABB AaBb AABb AaBB Ab AABb AAbb AaBb Aabb aB aaBB AaBB aaBb AaBb ab aabb Aabb AaBb aaBh

The Test Cross

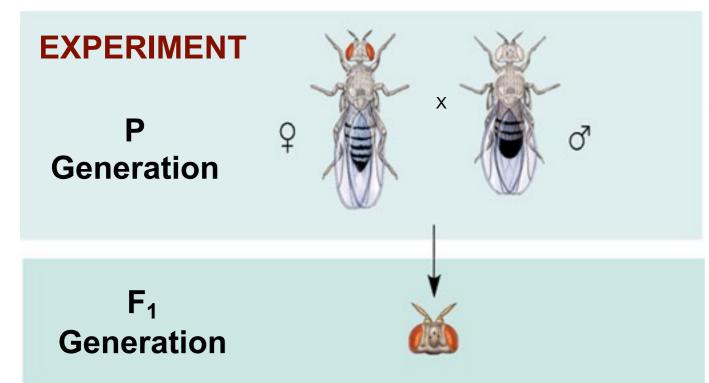
What if we find an organism with a dominant phenotype but we do not know its genotype. Can we determine whether its a hybrid or true-bred?



Sex Linked Inheritance

- Thomas Hunt Morgan, an embryologist from Columbia University, provided the first solid evidence that genes were in fact located on chromosomes.
 - Like Mendel his discovery was both insightful and a little lucky.
- After years of tedious work with fruit flies, Morgan provided the first support for the **chromosome theory of inheritance**, that specific genes are carried on specific chromosomes.
 - fruit flies breed quickly and have only 4 chromosomes
- In addition, he showed that genes located on the sex chromosomes exhibit a unique pattern of inheritance.

Morgan mated a wild-type (red-eyed) female with a mutant white-eyed male. The F₁ offspring all had red eyes.

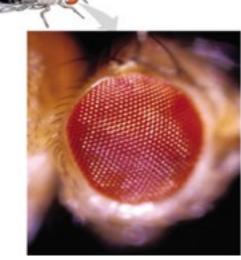


Morgan then bred an F_1 red-eyed female to an F_1 red-eyed male to produce the F_2 generation.



The F₂ generation showed a typical Mendelian 3:1 ratio of red eyes to white eyes. However, no females displayed the white-eye trait; they all had red eyes. Half the males had white eyes, and half had red eyes.

Morgan's Experiment

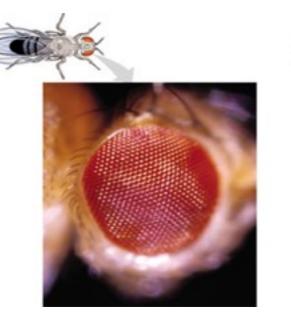


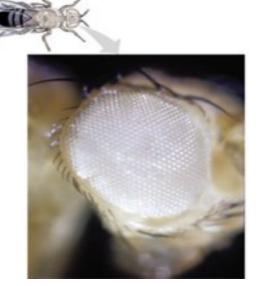
 w^+



Fruit Fly Genetic Symbols

Now called "wild type" instead of dominant





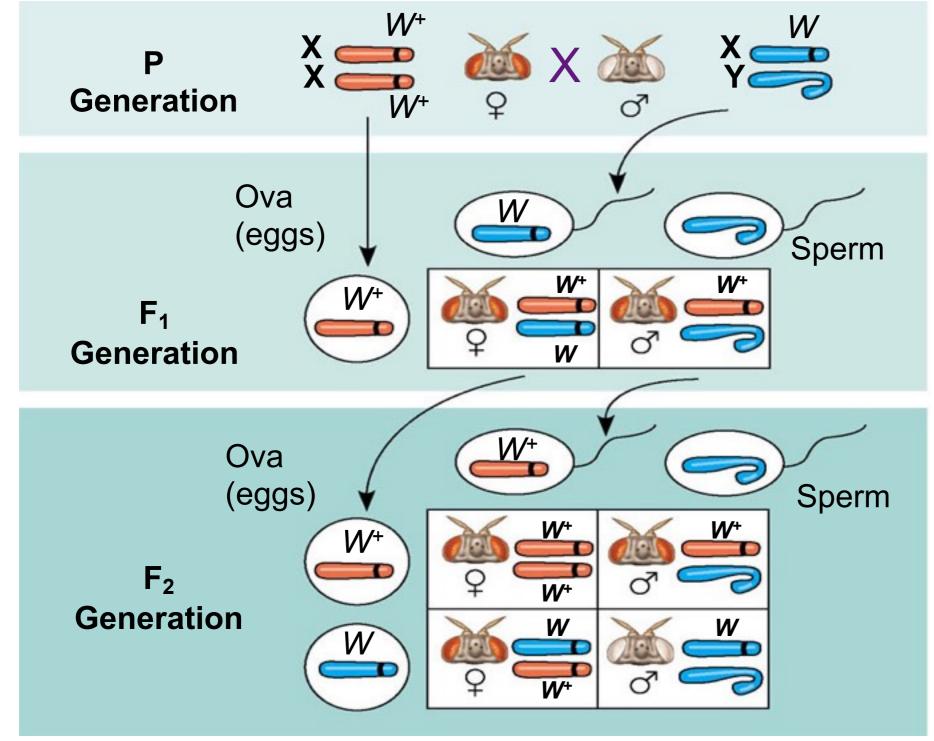
Now called "mutant" instead of recessive

(+) superscript now used instead of capital letter

V lower case letters still used for recessive allele

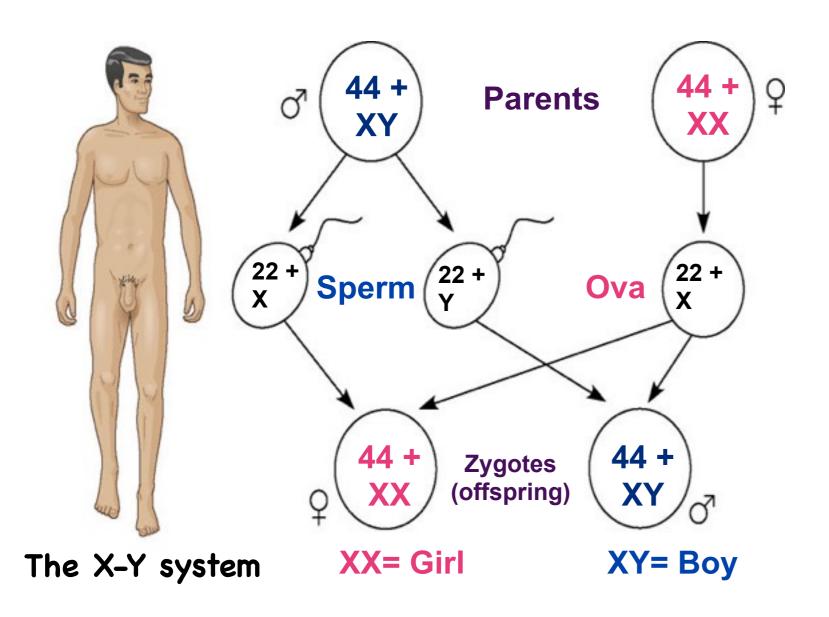
CONCLUSION

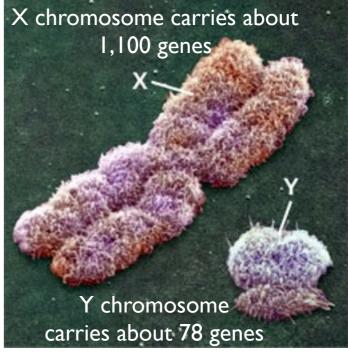
Since all F_1 offspring had red eyes, the mutant white-eye trait (*w*) must be recessive to the wild-type red-eye trait (*w*⁺). Since the recessive trait—white eyes—was expressed only in males in the F_2 generation, Morgan hypothesized that the eye-color gene is located on the X chromosome and that there is no corresponding locus on the Y chromosome, as diagrammed here.



Chromosomal Basis of Sex

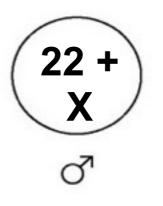
- There are two varieties of sex chromosomes X and Y.
- An organisms sex is determined by the presence or absence of certain sex chromosomes.
 X chromosome carries about





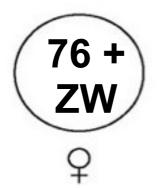
Other Systems of Sex Determination

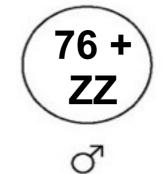




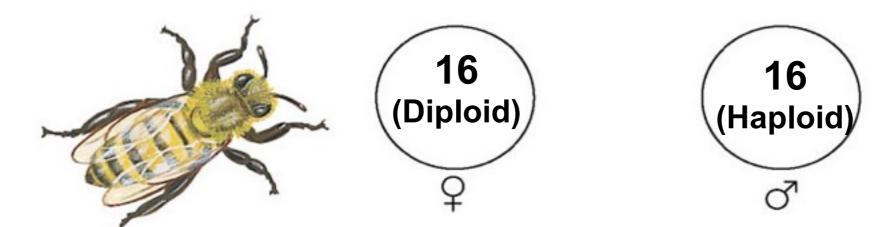
The X–0 system







The Z–W system

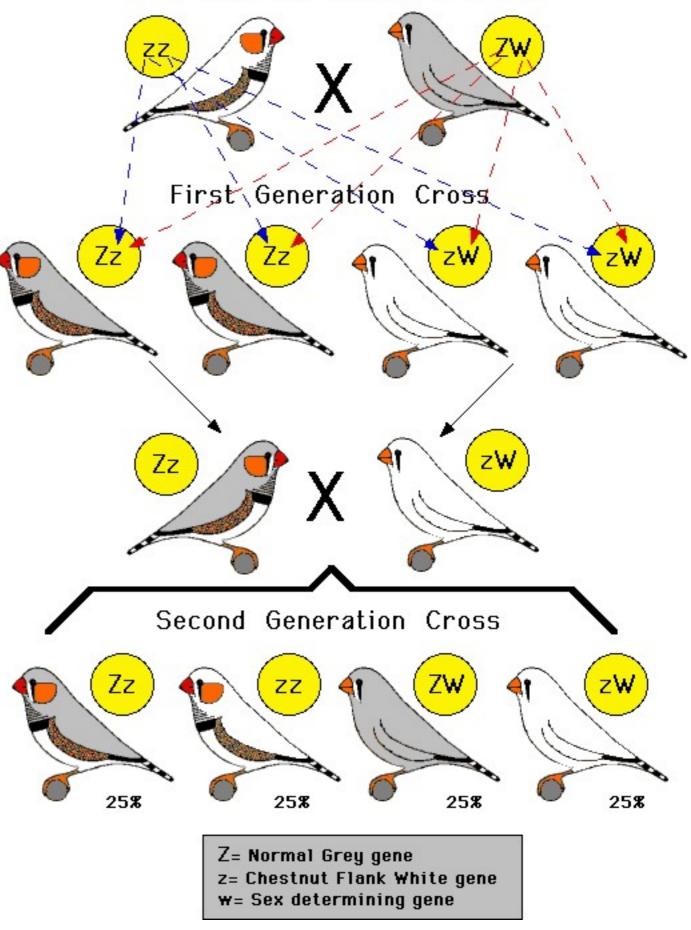


The haplo-diploid system

A gene located on any sex chromosome is said to be sex linked

Sex Linked Example on the ZW System

Sex linked Inheritance with a Chestnut Flanked White Cock and Normal Grey Hen

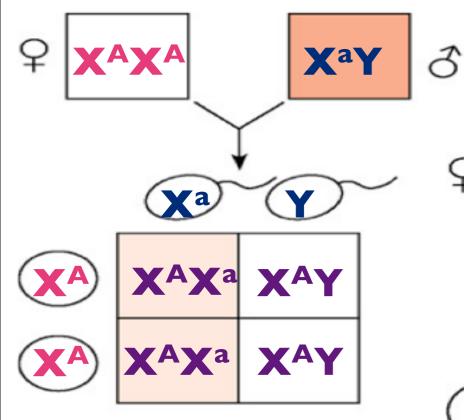


A gene located on any sex chromosome is said to be sex linked

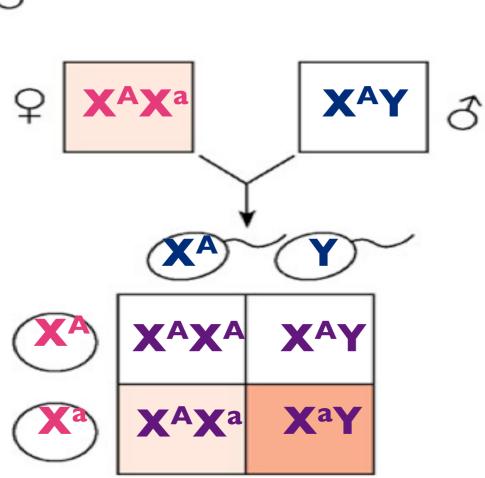
Inheritance of Sex Linked Traits

- Although a sex linked trait can be found on the X or Y chromosome, most genetic problems you will encounter will be "X-linked" traits.
 - Y linked traits are few and mainly sex determinate
- X-linked traits are far more numerous and some diseases are carried on this chromosome consequently most genetic problems are X-linked.
 - Duchenne Muscular Dystrophy, Hemophilia & Color Blindness
- Most importantly X-linked traits follow a unique pattern of inheritance, the same pattern seen in Morgan's fruit flies.

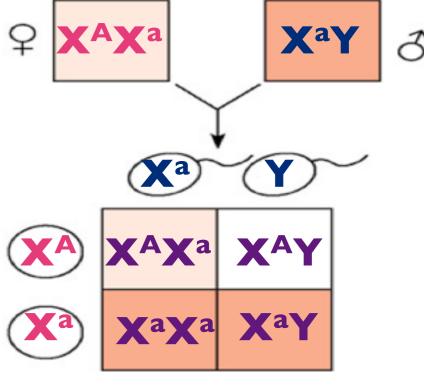
Inheritance of Sex Linked Traits



A father with the disorder will transmit the mutant allele to all daughters but to no sons. When the mother is a dominant homozygote, the daughters will have the normal phenotype but will be carriers of the mutation.



If a carrier mates with a male of normal phenotype, there is a 50% chance that each daughter will be a carrier like her mother, and a 50% chance that each son will have the disorder.



If a carrier mates with a male who has the disorder, there is a 50% chance that each child born to them will have the disorder, regardless of sex. Daughters who do not have the disorder will be carriers, where as males without the disorder will be completely free of the recessive allele. Essential knowledge 3.A.3: The chromosomal basis of inheritance provides an understanding of the pattern of passage (transmission) of genes from parent to offspring.

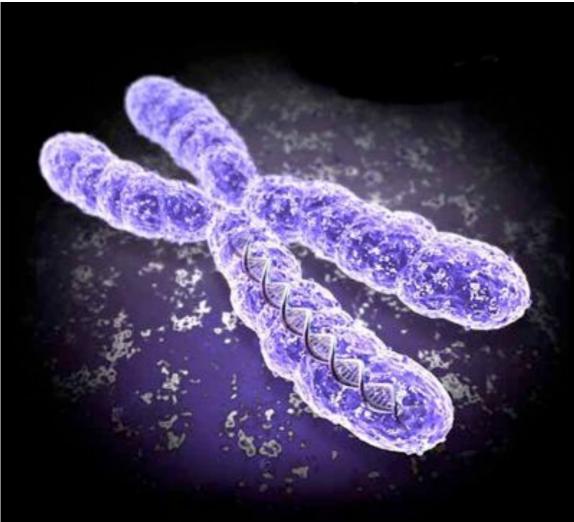
c. Certain human genetic disorders can be attributed to the inheritance of single gene traits or specific chromosomal changes, such as nondisjunction.

To foster student understanding of this concept, instructors can choose an illustrative example such as:

- Sickle cell anemia
- Tay-Sachs disease
- Huntington's disease
- X-linked color blindness
- Trisomy 21/Down syndrome
- Klinefelter's syndrome

Human Genetic Diseases

Main Idea: Many human diseases and disorders have a genetic basis. Understanding the genetic basis of disease helps us better predict and manage the disease in the future.





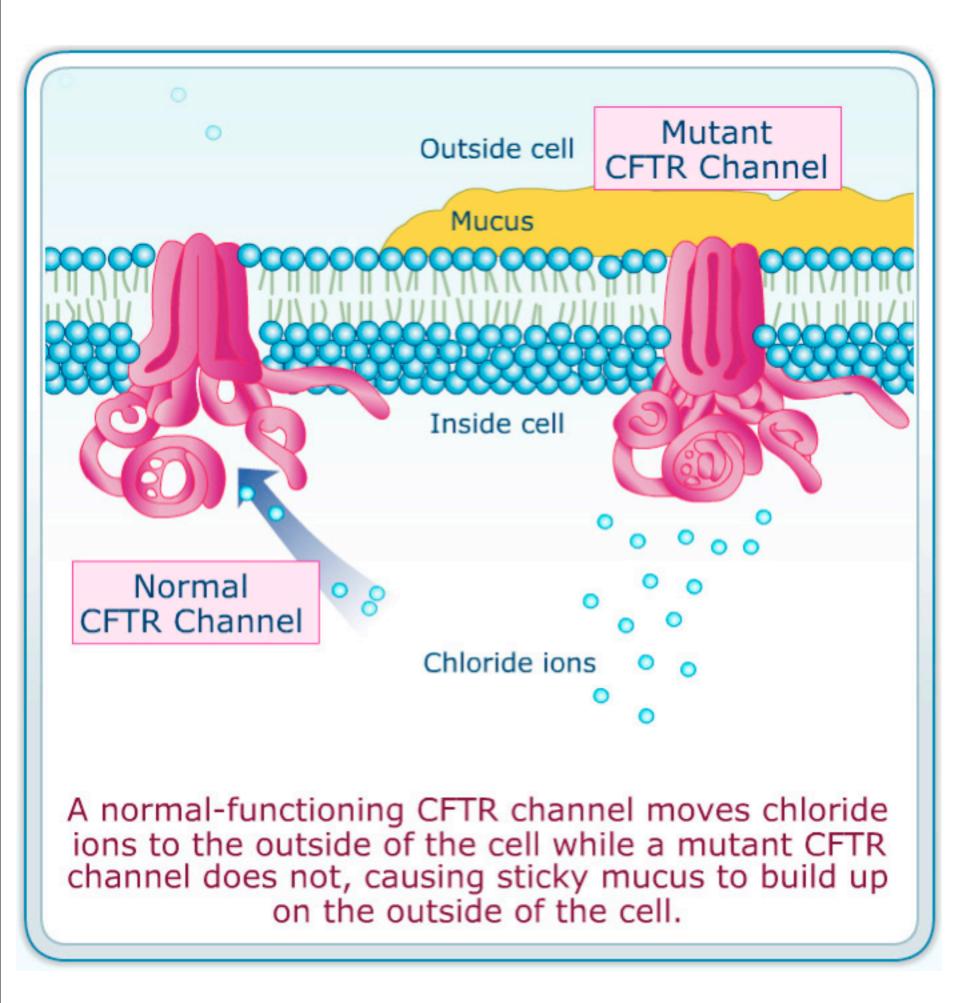
- Genetic disease and disorders range from mild phenotypes, like color blindness, to life threatening like Tay-Sachs disease.
- Some genetic disease occurs at the gene level, where a mutation results in a detrimental protein or level of protein.
 - These diseases can be dominant or recessive.
 - They are found on autosomes and sex chromosomes
- Other genetic disorders occur at the chromosomal level, where a mutation results in a too many chromosomes, too few chromosomes or broken chromosomes.

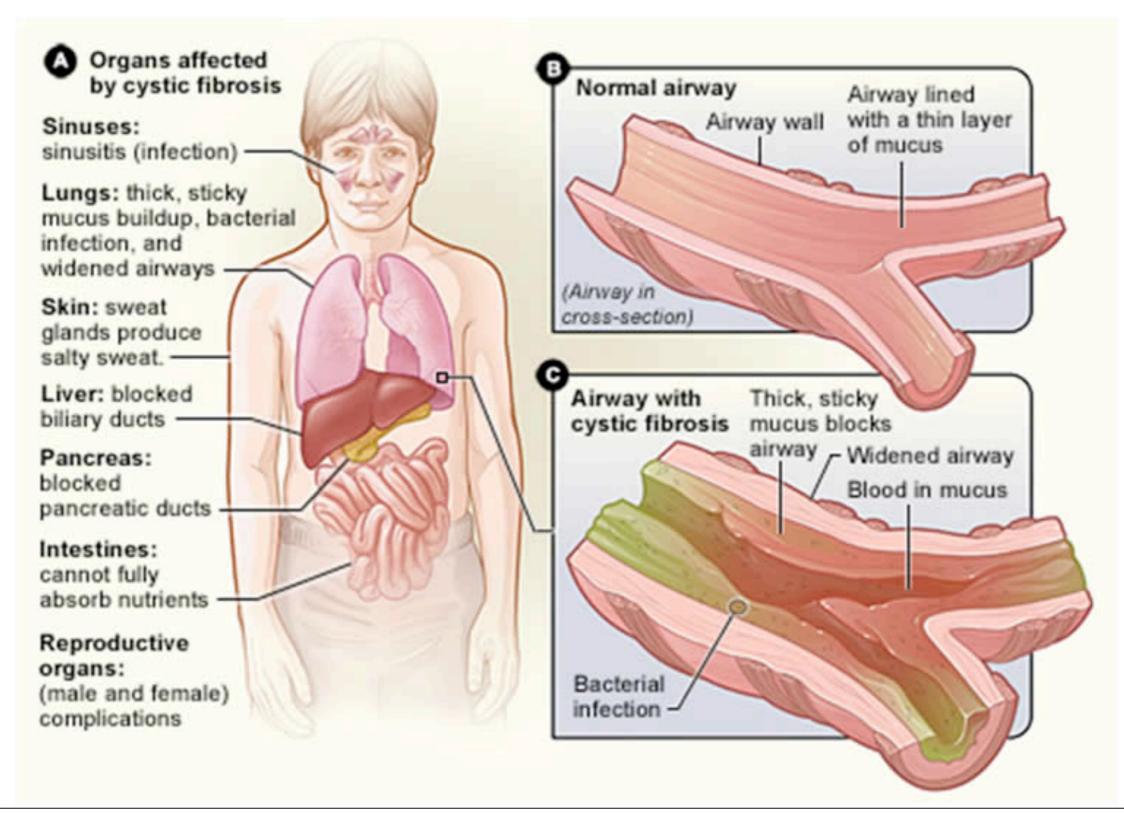
Recessive Disorders

- In general genetic disorders are not evenly distributed among all groups of people.
- When a disease causing allele is rare, it is unlikely that two carriers meet and mate.
- Because people with recent common ancestors are more likely to carry the same recessive alleles than unrelated people mating of close relatives produce more homozygous recessive offspring (diseased).
 - most societies and cultures have laws or taboos forbidding **consanguineous** marriages which may have evolved from empirical evidence over time.
 - many pure bred dog breeds today are so inbred they have greater incidence of physical and behavioral problems

Cystic Fibrosis

- The most common lethal genetic disease in the United States, strikes I in 2500 people of European descent.
- 4% of people with European descent are carriers for the trait.
- If untreated most children die before the age of 5.
- With treatment, more than 50% of those in the U.S. live into their 20's or 30's.
 - Treatment includes: antibiotics, daily pounding on the chest to clear mucous and other preventive treatments





Sickle Cell Anemia

- The most common genetic disease in people of African descent, strikes I in 400 people.
- About I in 10 African-Americans carry the trait.
- The high incidence stems from the partial resistance to malaria conferred by carrying the sickle cell trait thus being selected for in Africa where malaria is common.
- Regular blood transfusions can ward off brain damage in children and new drugs can help prevent and treat the disease other related problems but there is no cure.

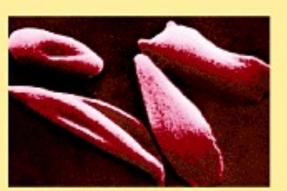
Two copies of the sickle-cell allele

All hemoglobin is the sickle-cell (abnormal) variety

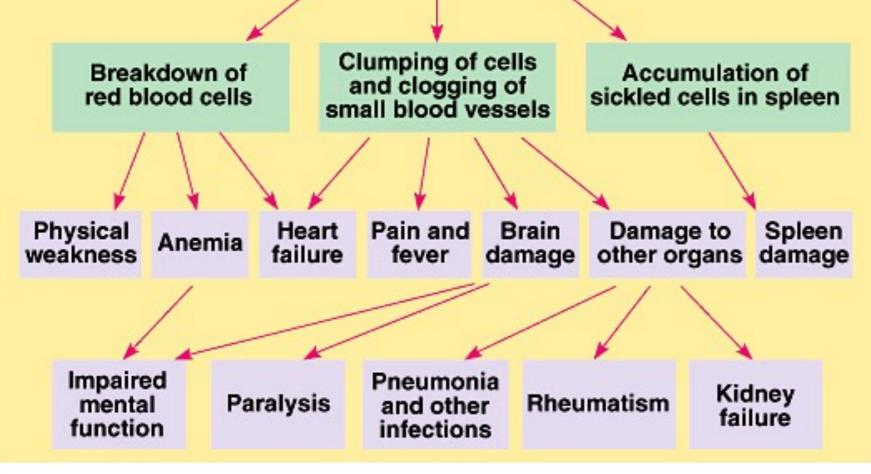
Abnormal hemoglobin crystallizes when oxygen content of blood is low, causing red blood cells to become sickle-shaped



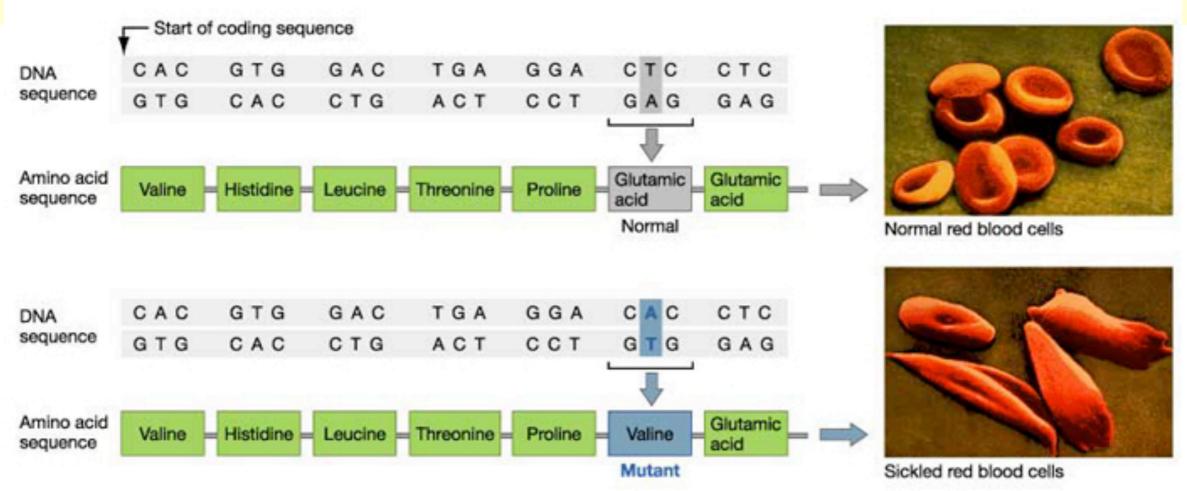
Normal cells



Sickled cells



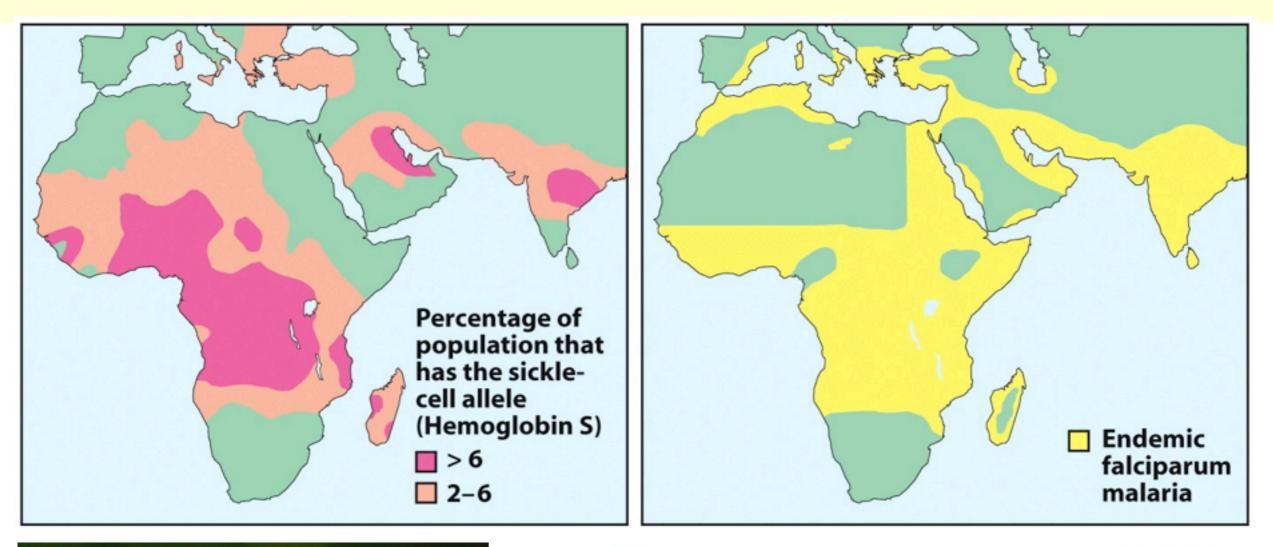
Sickle Cell Trait & Malaria



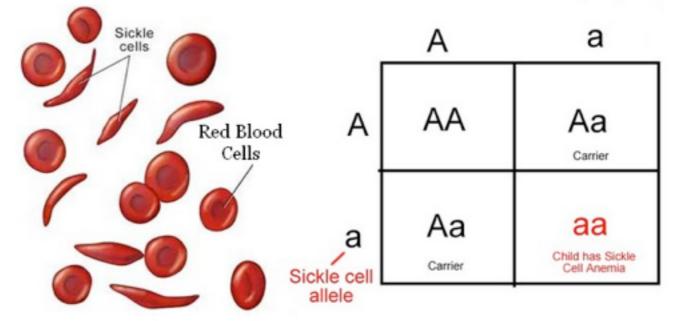
The change in amino acid sequence causes hemoglobin molecules to crystallize when oxygen levels in the blood are low. As a result, red blood cells sickle and get stuck in small blood vessels.

- This is a "substitution" mutation notice the thymine was switched with alanine.
- The normal beta subunit consists of 438 nucleotides and 146 amino acids.
- A change in 1 nucleotide, changes 1 amino acid resulting in sickle cell disease

Sickle Cell Trait & Malaria







Dominant Disorders

• Although many harmful alleles are recessive, a number genetic diseases are due to dominant alleles.



Achondroplasia

- A form of dwarfism that occurs in l in 25,000 people.
- The heterozygous individuals(Aa) are dwarfs, thus 99.9+% of the population is homozygous recessive(aa).
- The high incidence stems from the partial resistance to malaria conferred by carrying the sickle cell trait thus being selected for in Africa where malaria is common.
- Regular blood transfusions can ward off brain damage in children and new drugs can help prevent and treat the disease other related problems but there is no cure.

Huntington's Disease

- A rare lethal dominant allele located on the tip of chromosome #4, that effects 1 in 10,000 people.
- The timing of the disease expression is crucial in its persistence in the gene pool.
- Most lethal dominant alleles would be quickly eliminated from the gene pool because they offspring would die prior to passing on the trait.
- Huntington's, a degenerative disease of the nervous system has no effect until the person 35-45 years old, who has likely already reproduced and passed the gene on.

Aa X aa Offspring will have a 50% of inheriting the allele and the disease

Multifactorial Diseases

- The genetic disease discussed up to this point are caused by one or both alleles at one genetic locus.
- However many diseases have both a genetic component as well as an *environmental* component.
 - Cardiovascular Disease (#1 killer in U.S.),
 Cancer (#2), Diabetes (becoming epidemic),
 Alcoholism, Schizophrenia, Bipolar disorder
- To complicate matters the genetic component is often polygenic.
- So little is understood about the genetic component that the best public health strategy is to educate people about the environmental factors and promote healthy behavior.

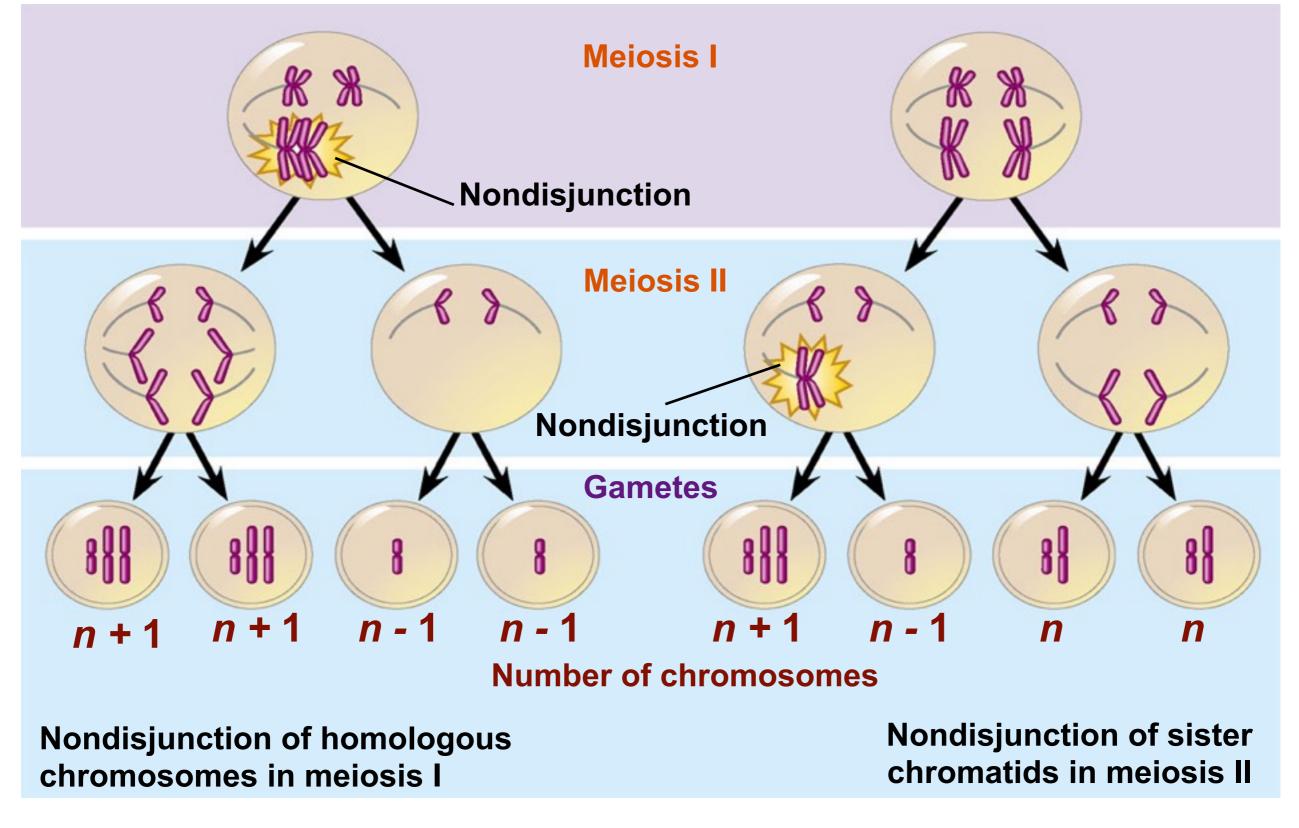
Chromosomal Disorders

- Large scale chromosomal changes can also effect an organisms phenotype and result in genetic disorders.
 - Errors in cell division can result in cells have too many chromosomes or too few chromosomes.
 - Physical and chemical disturbances can alter chromosome structure and function as well.
- These changes to the chromosome number or integrity result in genetic disorders.
- The disorders can vary in severity and plants tend to deal with these alterations better than animals.

Alteration in Chromosome Numbers

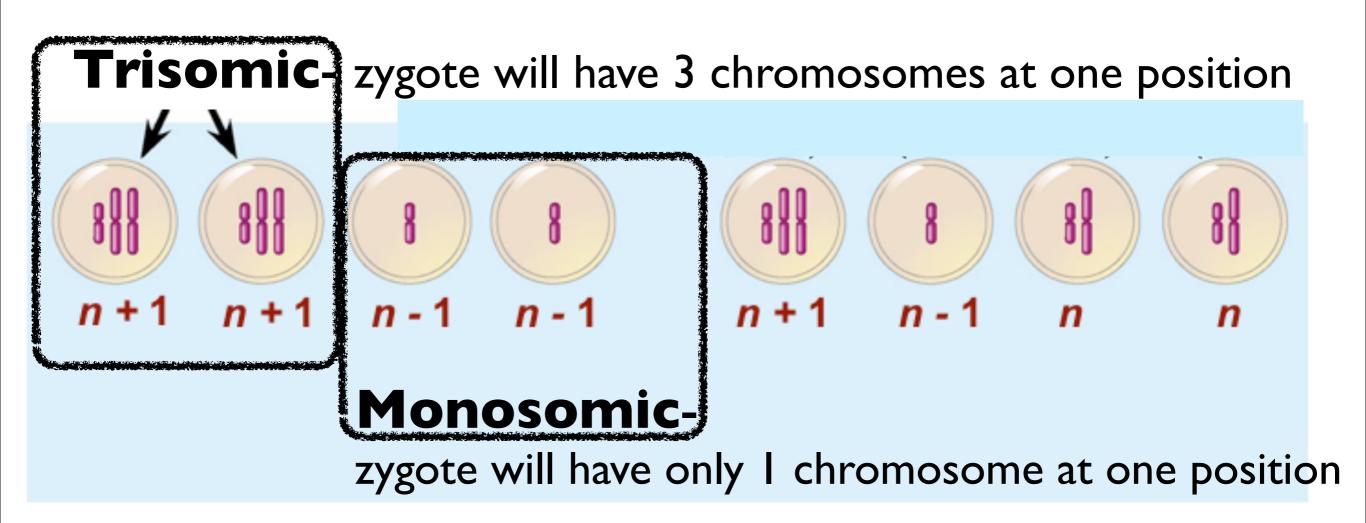
- Ideally chromosomes are distributed evenly and without error amongst daughter cells during meiosis.
- Occasionally errors occur, when, members of a pair of homologous chromosomes fail to separate during meiosis I or sister chromatids fail to separate during meiosis II it is called **nondisjunction**.
 - These errors in cell division result in some cells having too many chromosomes, while the other cells have too few chromosomes.
- Should any of these gametes fuse with a normal gamete the resulting zygote will also have an abnormal number of chromosomes

Nondisjunction



Nondisjunction can also occur in mitosis, during embryological development.

Thursday, February 9, 17



Aneuploidy- a condition where an individual has an abnormal number of chromosomes and it may involve more than one chromosome.

 Mitosis will consequently pass the anomaly to each and every cell of the body during development.

Alteration in Chromosome Number

- These alterations may be quite common but most of the time we never see the results of such alterations because the embryos spontaneously abort well before birth.
- When the embryo survives it results in a syndrome, a set of certain traits associated with that specific type of aneuploidy.
 - ex. Downs syndrome, Klinefelters, Turners

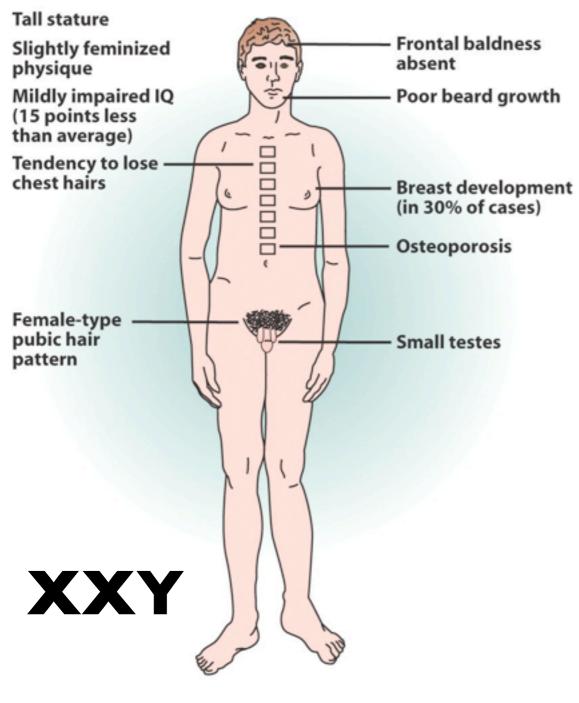
Downs Syndrome



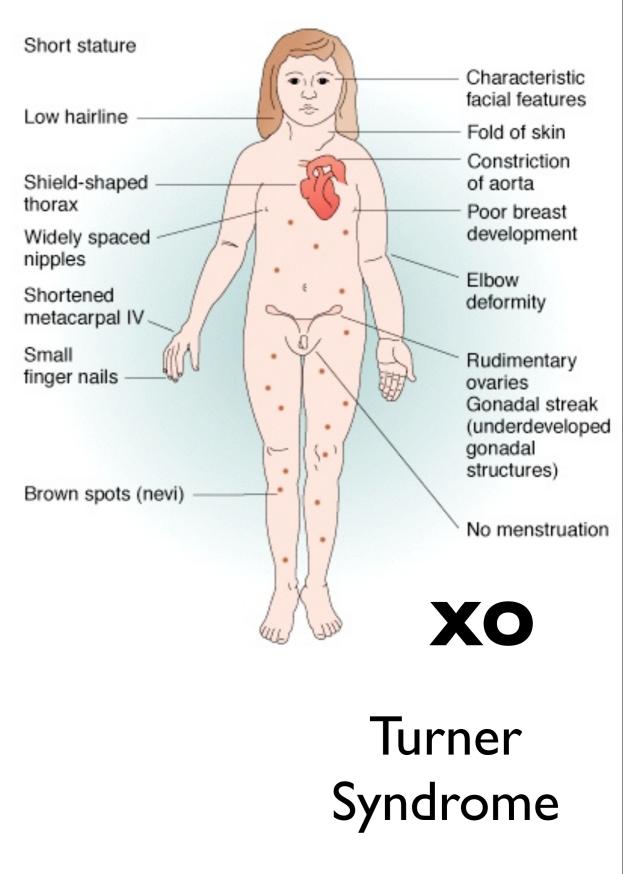
I in 700 births in U.S.

Thursday, February 9, 17

Two or the more common sex chromosome aneuploidy conditions



Klinefelter Syndrome



Thursday, February 9, 17

Aneuploidy in Sex Chromosomes

Table 1. Main features of numerical sex chromosome anomalies

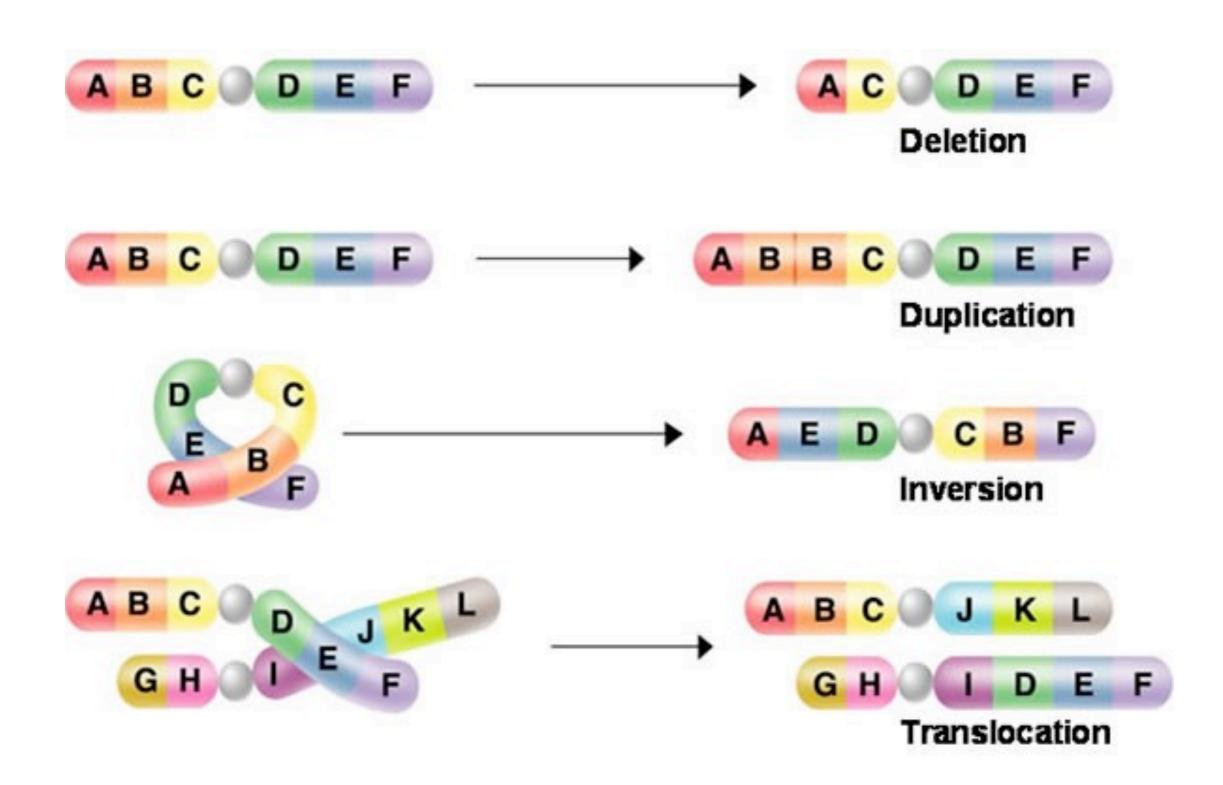
Karyotype	Incidence	Mental retardation	Behavioral disorders	Stature	Gonadal function	Congenital anomalies/ Additional medical problems	Ref.
45,X	1:2,130 F	-		Short	Hypergonadotropic hypogonadism	Dysmorphic picture, CV and renal anomalies, autoimmune disorders	1,2
47,XXY	1:576 M	Greater frequency when compared with normal men	Greater frequency when compared with normal men	Tall	Hypergonadotropic hypogonadism	Minor physical findings, varicose veins, DVT, <i>diabetes mellitus</i> , autoimmune disorders	1, 2,6
47,XYY	1:851 M	Greater frequency when compared with normal men	Greater frequency when compared with normal men	Tall	Usually normal	Minor physical findings	1,2,6
47,XXX	1:897 F	Greater frequency when compared with normal women	Greater frequency when compared with normal women	Tall	Usually normal Unknown frequency of premature ovarian failure	Minor physical findings, low frequency of genitourinary anomalies and seizures	1,2,6
48,XXXX 49,XXXXX	?	+	Variable	Short	Hypergonadotropic hypogonadism	Dysmorphic picture, CV anomalies	6,16
48,XXYY	1:18,000- 1:40,000 M	+	+	Tall	Hypergonadotropic hypogonadism	Dysmorphic picture, CV and renal anomalies, type II diabetes, seizures, DVT	3,6,7
48,XXXY	1:50,000 M	+	+	Tall	Hypergonadotropic hypogonadism	Dysmorphic picture, CV and renal anomalies, type II diabetes, seizures, DVT	4,6,7
49,XXXXY	1:85,000- 1:100,000 M	+	+	Short	Hypergonadotropic hypogonadism	Dysmorphic picture, CV and renal anomalies, type II diabetes, seizures, DVT	4,6,7

+ = present; - = absent; CV = cardiovascular; DVT = deep vein thrombosis; F = females; M = males.

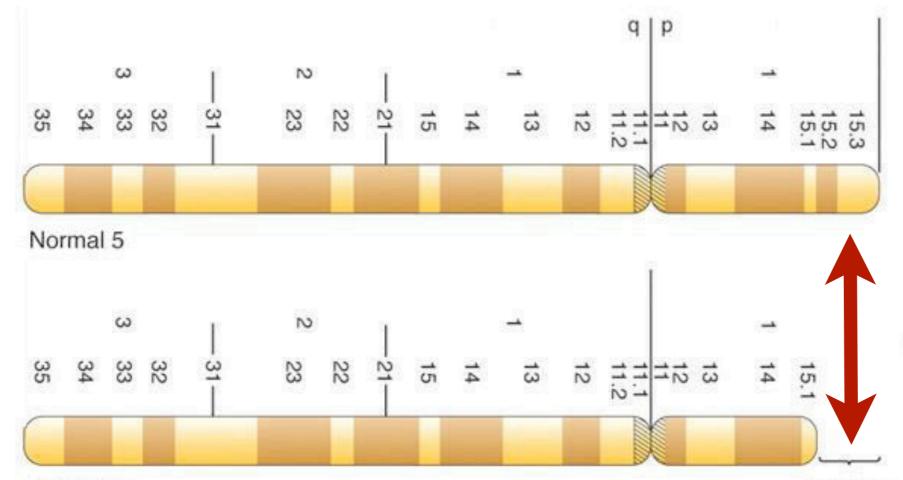
Alteration in Chromosome Structure

- Errors in meiosis or damaging agents can alter chromosome structure in 1 of 4 ways. (illustrated on next slide)
 - deletions, duplications, inversions & translocations
- These alterations may cause severe problems.
 - Cri du Chat, Chronic Myelogenous Leukemia, Burkitt's Lymphoma

Chromosomal Mutations



THE CRI DU CHAT SYNDROME



Deleted 5















Deletion

Thursday, February 9, 17

Essential knowledge 3.A.3: The chromosomal basis of inheritance provides an understanding of the pattern of passage (transmission) of genes from parent to offspring.

d. Many ethical, social and medical issues surround human genetic disorders.

To foster student understanding of this concept, instructors can choose an illustrative example such as:

• Reproduction issues

• Civic issues such as ownership of genetic information, privacy, historical contexts, etc.

Learning Objectives:

LO 3.12 The student is able to construct a representation that connects the process of meiosis to the passage of traits from parent to offspring. [See SP 1.1, 7.2]

LO 3.13 The student is able to pose questions about ethical, social or medical issues surrounding human genetic disorders. [See SP 3.1]

LO 3.14 The student is able to apply mathematical routines to determine Mendelian patterns of inheritance provided by data sets. [See SP 2.2]

Appendix: Genetic Problems

Main Idea: Genetic problems are common in many biology classes and exams. In this section I will address common genetic problems by type, offer solution tips and explain how statistical tools are used for evaluating results.



Chi-Squared Test

- Chi square, is a test for difference between two data sets.
- It can only be used on raw count data, not measurements or derived data.
- It should only be used to compare experimental data (actual results) with theoretical data (expected results).
- Is not reliable when sample size is below 20, the more data the better.
- Its goal is to test the **Null Hypothesis**
 - Null Hypothesis (H₀) states that there is no difference between the two data sets.

Side bar...

Know the meaning of the 3 different types of chi-square analysis techniques.

(1) Goodness-of-fit test is a chi-square test technique used to study similarities between proportions or frequencies between groupings (or classification) of categorical data. this is our application in genetics

(comparing a distribution of data with another distribution of data where the expected frequencies are known).

(2) **Tests of Independence** is a chi-square technique used to determine whether two characteristics (such as food spoilage and refrigeration temperature) are related or independent.

(3) **Test of Homogeneity** is a chi-square technique used to study whether different populations are similar (or homogeneous or equal) in reference to some characteristic or attribute (such as "do students national identity affects the time spend doing homework?").

Chi-Squared in Genetics

- In many genetic problems we are asked to make a prediction about fate of a certain gene or likelihood that an offspring receives a certain gene.
- Recall that punnet squares and rules of probability are the tools we use to make these predictions
 - For Instance in the dihybrid cross below, a Mendelian pattern of inheritance predicts a 9:3:3:1 phenotypic ratio.

AaBb X AaBb

- 9 = yellow/round
- 3 = green/round
- 3 = yellow/wrinkled
- I = green/wrinkled

- Where... A = yellow a = green
- B = round
- b = wrinkled

Chi-Squared in Genetics

- In reality, when crosses are actually carried out in the lab or in your potential genetic problems the actual phenotypic ratio never exactly matches the expected ratio.
- Now consider two possibilities
 - I.The actual results are "close enough" to the expected ratios to conclude that the <u>assumed pattern of inheritance</u> is at work in this case.
 - OR 2. The actual results are simply <u>due to chance or some</u> <u>other factor</u>, thus assumed pattern of inheritance has no data to support it.

How close is "close enough"? Hard to say going on instincts alone, our "gut feelings" are too subjective

Using Chi-Squared

- Chi square is a statistical application that objectively helps us decide between the two alternatives, in other words how close is "close enough".
- State your two hypotheses.
 - I. Null Hypothesis (H₀)-There is no difference between the two data sets. Your actual results are the same as your expected results.
 - 2. Alternative Hypothesis (H_A) -There is a difference between the two data sets. Your actual results are not what you predicted(put simply H_A is H₀ is not true).

Now use Chi-square and let it determine which of the two choices above is more likely

$$\mathbf{X}^{2} = \sum_{i=1}^{n} \frac{(O_{i} - E_{i})^{2}}{E_{i}}$$

Chi-Squared Equation

$$\mathbf{X}^{2} = \sum_{i=1}^{n} \frac{(O_{i} - E_{i})^{2}}{E_{i}}$$

where

 ${\rm X}^2$ = Pearson's cumulative test statistic, which asymptotically approaches a

 χ^2 distribution.

- O_i = an observed frequency;
- E_i = an expected (theoretical) frequency, asserted by the null hypothesis;
- n = the number of cells in the table.

Using Chi-Squared AaBb X AaBb

PREDICTED

- 9 = yellow/round
- 3 = green/round
- 3 = yellow/wrinkled
- I = green/wrinkled

$$\mathbf{X}^{2} = \sum_{i=1}^{n} \frac{(O_{i} - E_{i})^{2}}{E_{i}}$$

EXPERIMENTAL

- 441 = yellow/round
- 159 = green/round
- 143 = yellow/wrinkled
- 57 = green/wrinkled

The predicted ratios assume that two traits are assorting independently and as as result probability of creating a yellow/round seed is 9/16, etc.

The experimental ratios are not perfect matches of the predicted percentages.

SO...

Are these traits following the presumed pattern of inheritance?

Thursday, February 9, 17



- I. State H_0 and H_A hypotheses
- 2. Calculate chi squared
- 3. Calculate degrees of freedom
- 4. Find P value for your X² value
- 5. Reject or Accept your H₀

I. State H_0 and H_A hypotheses

- I. Null Hypothesis (H₀)-There is no difference between the two data sets. Your actual results are the same as your expected results.
- 2. Alternative Hypothesis (H_A) -There is a difference between the two data sets. Your actual results are not what you predicted (put simply H_A is H₀ is not true).

PREDICTED

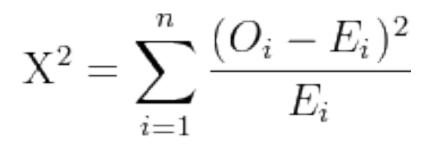
- 9 = yellow/round
- 3 = green/round
- 3 = yellow/wrinkled
- I = green/wrinkled

EXPERIMENTAL

- 441 = yellow/round
- 159 = green/round
- 143 = yellow/wrinkled
- 57 = green/wrinkled

2. Calculate chi squared

Expected: (9/16)(800)= 450 (3/16)(800)= 150 (3/16)(800)= 150 (1/16)(800)= 50



Total
800441 = yellow/round
159 = green/round
143 = yellow/wrinkled
57 = green/wrinkled

Category	0	Ε	0-Е	(O-E) ²	<u>(O-E)²</u> E
yellow/round	441	450	-9	81	0.18
green/round	159	150	9	81	0.54
yellow/wrinkled	143	150	7	49	0.33
green/round	57	50	7	49	0.98
	∑ 800				2.03

3. Calculate degrees of freedom



Category

yellow/round

green/round

yellow/wrinkled

green/round

n = 4 categories degrees of freedom = n-l thus 4-l = 3 d.f.

$X^2 = 2.03$

4. Find P value for X² value

df	$\chi^2_{0.005}$	$\chi^{2}_{0.01}$	$\chi^{2}_{0.025}$	$\chi^{2}_{0.05}$	$\chi^{2}_{0.10}$	$\chi^{2}_{0.90}$	$\chi^{2}_{0.95}$	$x_{0.975}^2$	$\chi^{2}_{0.99}$	$\chi^{2}_{0.995}$
1	0.000039	0.00016	0.00098	0.0039	0.0158	2.71	3.84	5.02	6.63	7.88
2	0.01	0.0201	0.0506	0.1026	0.2107	4.61	5.99	7.38	9.21	10.6
3	0.0717	0.115	0.216	0.352	0.584	6.25	7.81	9.35	11.34	12.84
4	0.207	0.297	0.484	0.711	1.064	7.78	9.49	11.14	13.28	14.86
5	0.412	0.554	0.831	1.15	1.61	9.24	11.07	12.83	15.09	16.75
6	0.676	0.872	1.24	1.64	2.20	10.64	12.59	14.45	16.81	18.55
7	0.989	1.24	1.69	2.17	2.83	12.02	14.07	16.01	18.48	20.28
8	1.34	1.65	2.18	2.73	3.49	13.36	15.51	17.53	20.09	21.96
9	1.73	2.09	2.70	3.33	4.17	14.68	16.92	19.02	21.67	23.59
10	2.16	2.56	3.25	3.94	4.87	15.99	18.31	20.48	23.21	25.19

Accept H₀

Reject H₀

$X^2 = 2.03$

5. Reject or Accept your H₀

df	$\chi^{2}_{0.005}$	$\chi^{2}_{0.01}$	$\chi^{2}_{0.025}$	$\chi^{2}_{0.05}$	$\chi^{2}_{0.10}$	$\chi^{2}_{0.90}$	$\chi^{2}_{0.95}$	$\chi^{2}_{0.975}$	$\chi^{2}_{0.99}$	$\chi^2_{0.995}$
1	0.000039	0.00016	0.00098	0.0039	0.0158	2.71	3.84	5.02	6.63	7.88
2	0.01	0.0201	0.0506	0.1026	0.2107	4.61	5.99	7.38	9.21	10.6
3	0.0717	0.115	0.216	0.352	0.584	6.25	7.81	9.35	11.34	12.84
4	0.207	0.297	0.484	0.711	1.064	7.78	9.49	11.14	13.28	14.86
5	0.412	0.554	0.831	1.15	1.61	9.24	11.07	12.83	15.09	16.75
6	0.676	0.872	1.24	1.64	2.20	10.64	12.59	14.45	16.81	18.55
7	0.989	1.24	1.69	2.17	2.83	12.02	14.07	16.01	18.48	20.28
8	1.34	1.65	2.18	2.73	3.49	13.36	15.51	17.53	20.09	21.96
9	1.73	2.09	2.70	3.33	4.17	14.68	16.92	19.02	21.67	23.59
10	2.16	2.56	3.25	3.94	4.87	15.99	18.31	20.48	23.21	25.19



Reject H₀

There is no difference between the two data sets.

Your actual results are the same as your expected results.

Thursday, February 9, 17

Analysis of Different Types of Genetic Problems

coming next year...

In the meantime I have sample problems of every type that you may encounter on the test. You can find them on the website under the homework tab.