

**Topic 1:**  
**Cracking the Mystery of Heredity**

Sssshhhh ... listen to the pea plants

# Genetics

The study of heredity, how traits are passed from parent to offspring is full of mystery



or

=



or





## How Can This Be?



A Kermode  
“Spirit” Bear

- The details of heredity remained a mystery for many years
- Many thought the parents' traits blended in the next generation ... much like mixing red and white paint to get pink



- Can you think of any human traits that are NOT inherited in this way???

• The first person to begin solving the mystery of heredity in the 1800s was an Austrian monk named Gregor Mendel



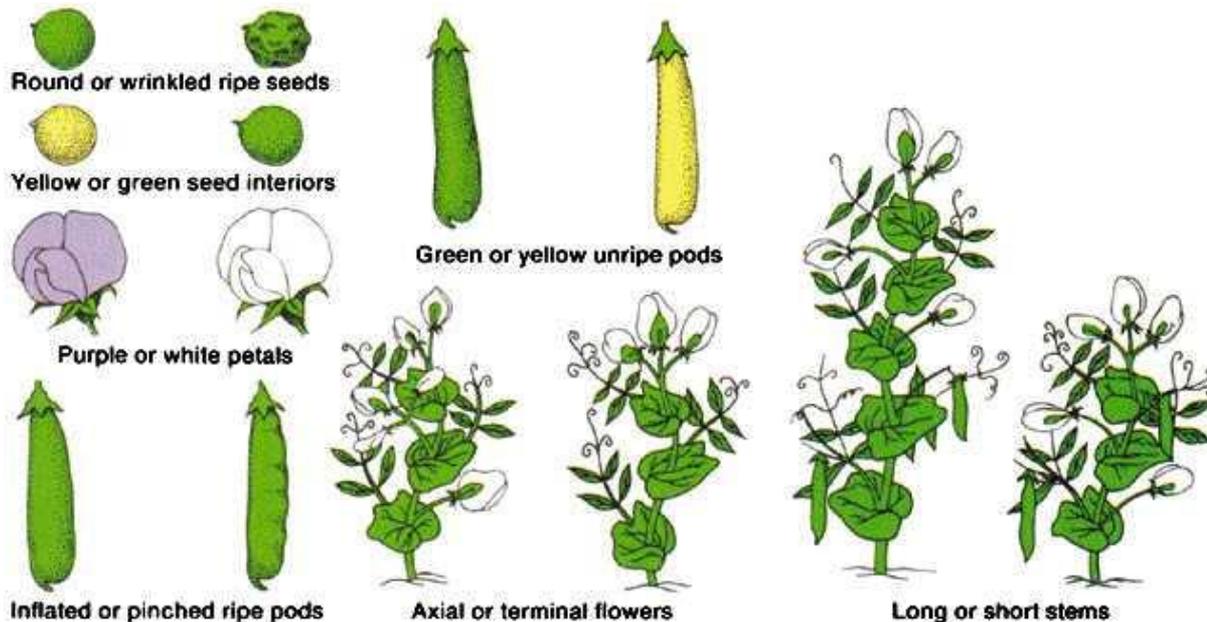
**“Father of Genetics”**



# Mendel's Experiments

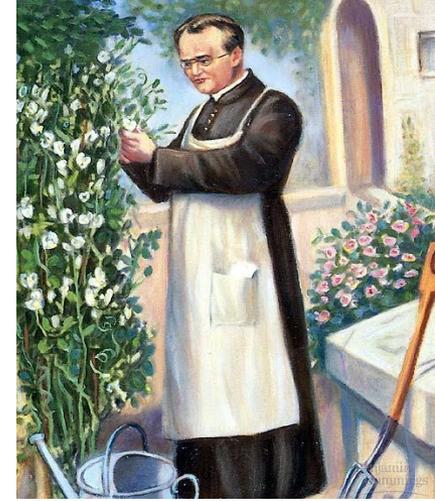


- Mendel's understanding of heredity came from several years observing how traits are inherited in pea plants
- He chose to follow 7 simple traits that occur in 2 opposite forms through 2 generations of breeding experiments.

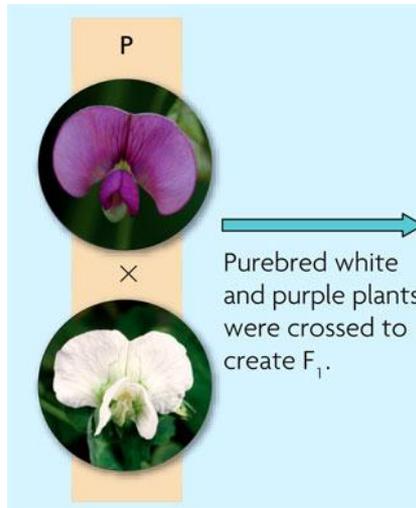




# Mendel's Experiments

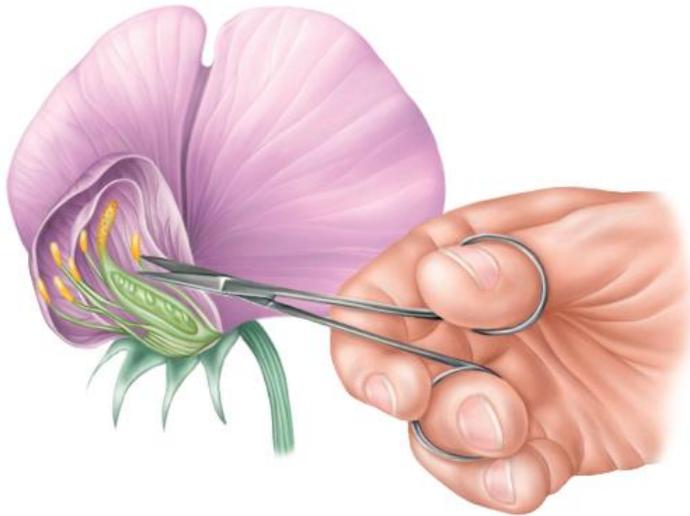


- Mendel's experiments began by hand cross-pollinating parent plants (P generation) that breed pure generation after generation for 2 opposite forms of a trait
- (e.g. purple flowers x white flowers)

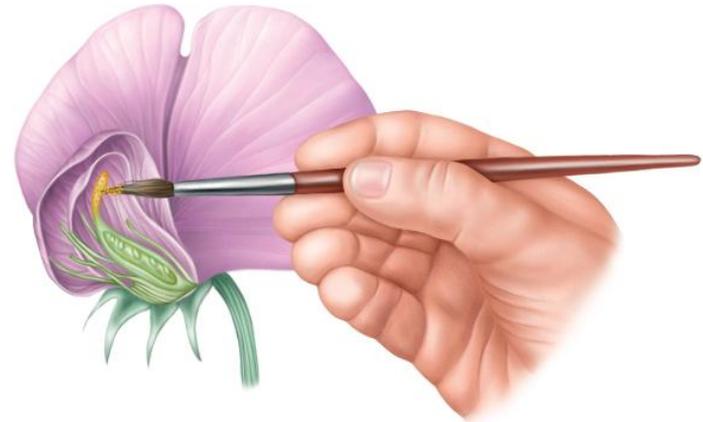


## 6.3 Mendel and Heredity

- Mendel used pollen to fertilize selected pea plants.
  - P generation crossed to produce F<sub>1</sub> generation
  - interrupted the self-pollination process by removing male flower parts



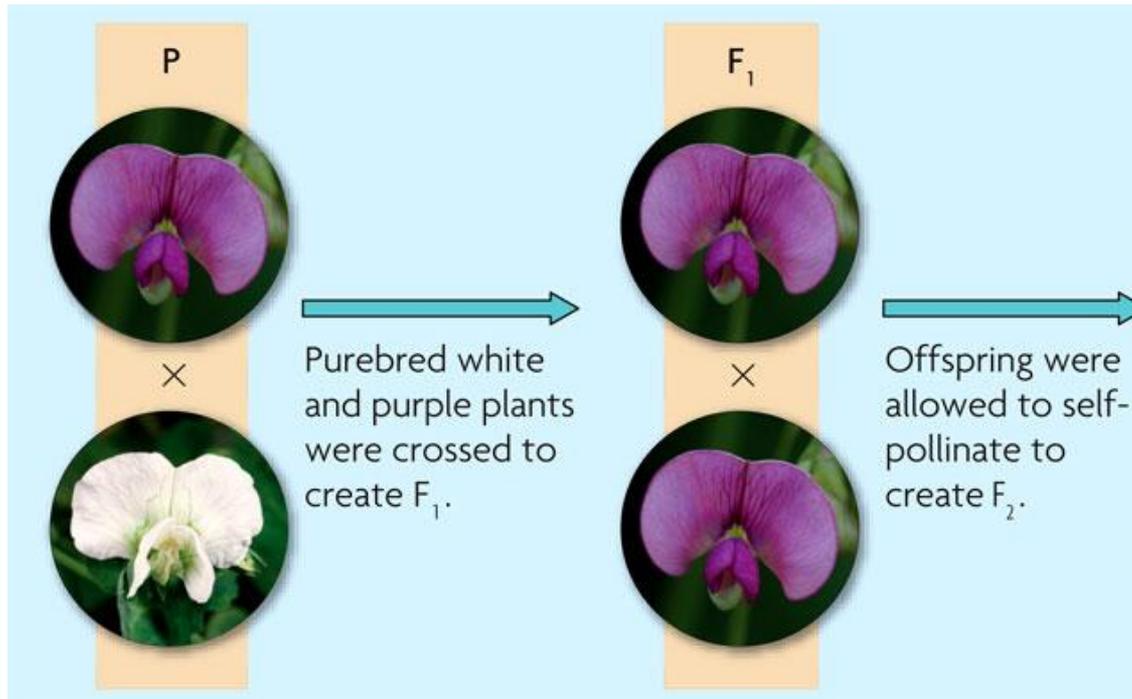
**Mendel controlled the fertilization of his pea plants by removing the male parts, or stamens.**



**He then fertilized the female part, or pistil, with pollen from a different pea plant.**

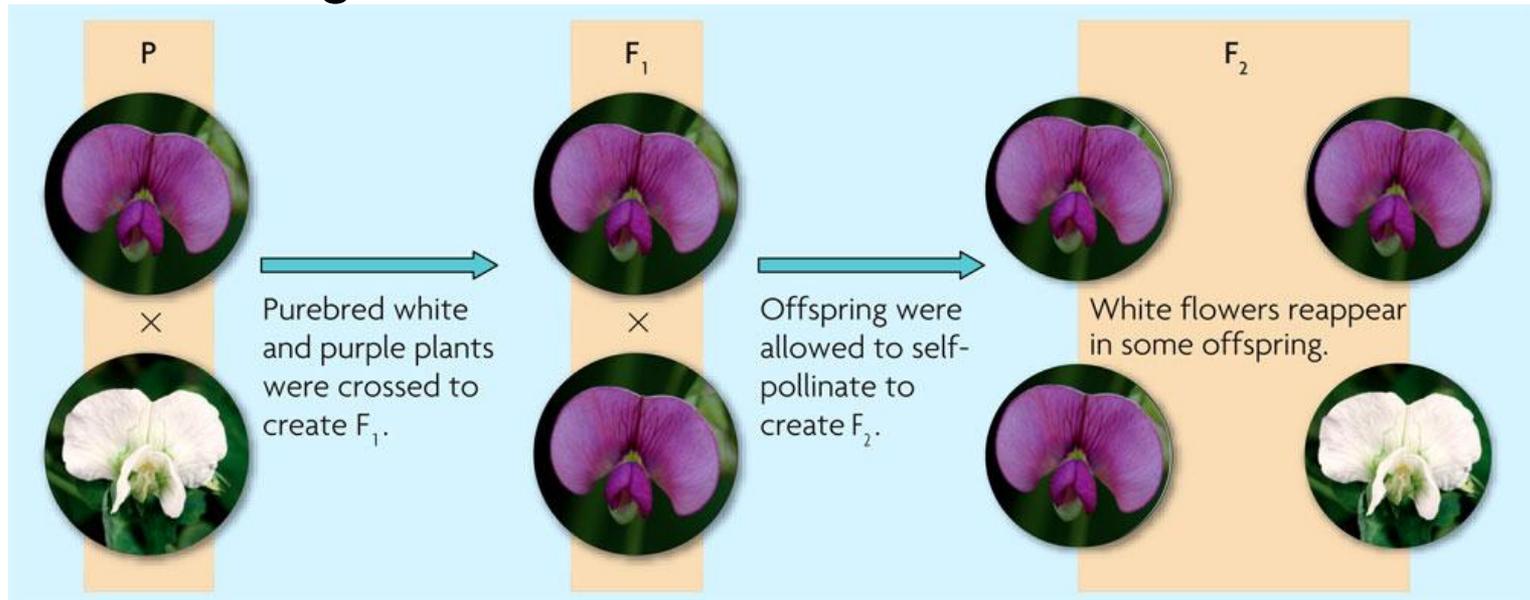
## 6.3 Mendel and Heredity

- The next generation ( $F_1$ ) had all purple flowers
- Mendel wondered what had happened to the **white** trait???
- He allowed the purple flowered  $F_1$  generation to self-pollinate the next generation to see what would happen?



## 6.3 Mendel and Heredity

- The second generation ( $F_2$ ) had many purple flowers but some white flowers.....HOW???
- Mendel suggested that each pea plant trait is determined by inheriting 2 gene codes, one from each parent
- Mendel was the first to crack this inheritance mystery when he proposed that one gene code is Dominant and the other gene code is recessive and remains “hidden” unless both genes are recessive



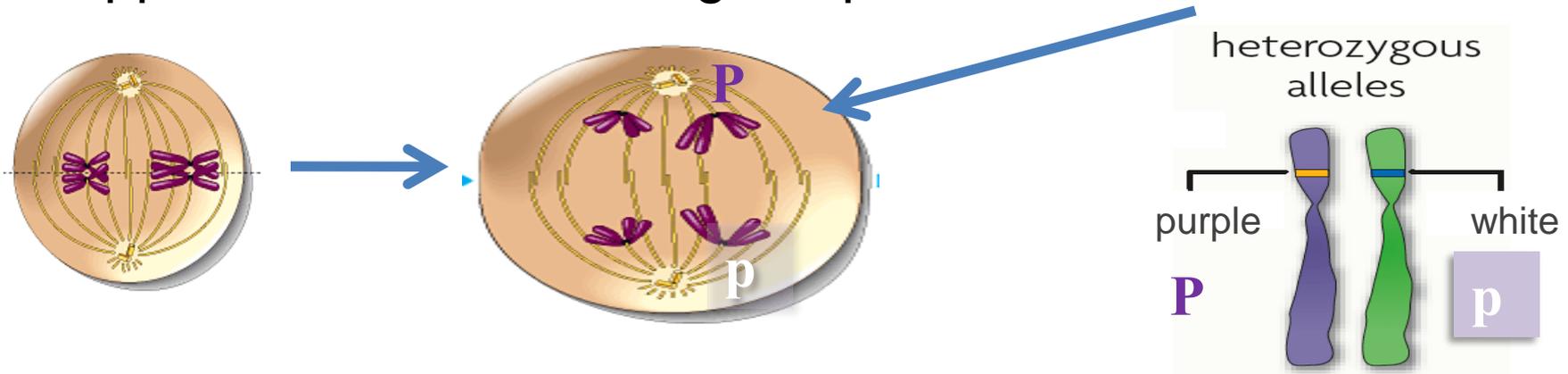
## 6.3 Mendel and Heredity

- Mendel repeated his experiment for other traits and noticed **patterns** in the first and second generations of his crosses.
- Do **YOU** notice any patterns in his F<sub>2</sub> data below ???

FIGURE 6.10 MENDEL'S MONOHYBRID CROSS RESULTS			
F <sub>2</sub> TRAITS	DOMINANT	RECESSIVE	RATIO
Pea shape	5474 round	1850 wrinkled	2.96:1
Pea color	6022 yellow	2001 green	3.01:1
Flower color	705 purple	224 white	3.15:1
Pod shape	882 smooth	299 constricted	2.95:1
Pod color	428 green	152 yellow	2.82:1
Flower position	651 axial	207 terminal	3.14:1
Plant height	787 tall	277 short	2.84:1

## 6.3 Mendel and Heredity

- Mendel wondered why the recessive trait consistently seemed to hide during the F<sub>1</sub> generation but then reappeared in the F<sub>2</sub> generation only 1/4th of the time???
- He reasoned that this consistent **3:1** ratio in the traits follows the rules of math probability. This means that each time a parent creates a gamete, they must randomly send only 1 of their 2 gene codes just like flipping a coin. Today we understand why Mendel's "coin toss" idea was correct because we can see the two genes move in opposite directions during Anaphase 1 of Meiosis.



## 6.3 Mendel and Heredity

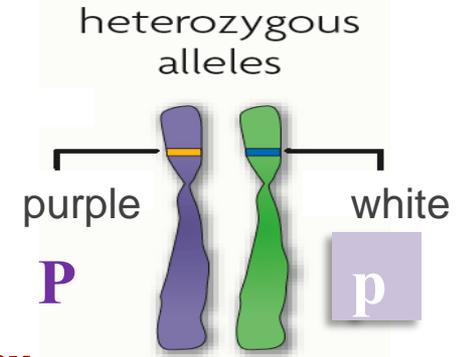
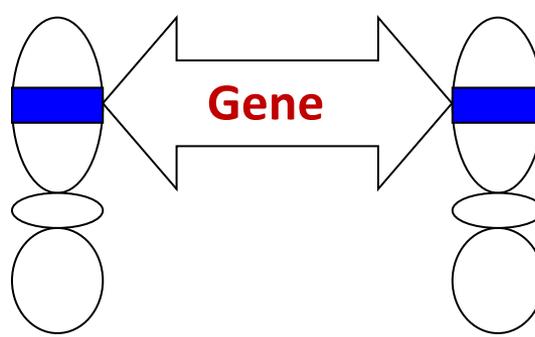


### Mendel's Legacy

- Mendel's legacy is that his work laid the foundation for understanding heredity
- His ideas about heredity correctly explain how many traits are inherited, not only for pea plants, but also for all sexually reproducing organisms from insects to humans.

Punnett Squares  
AND  
Human Inherited Disorders

**Topic 2: Inheritance TERMS**

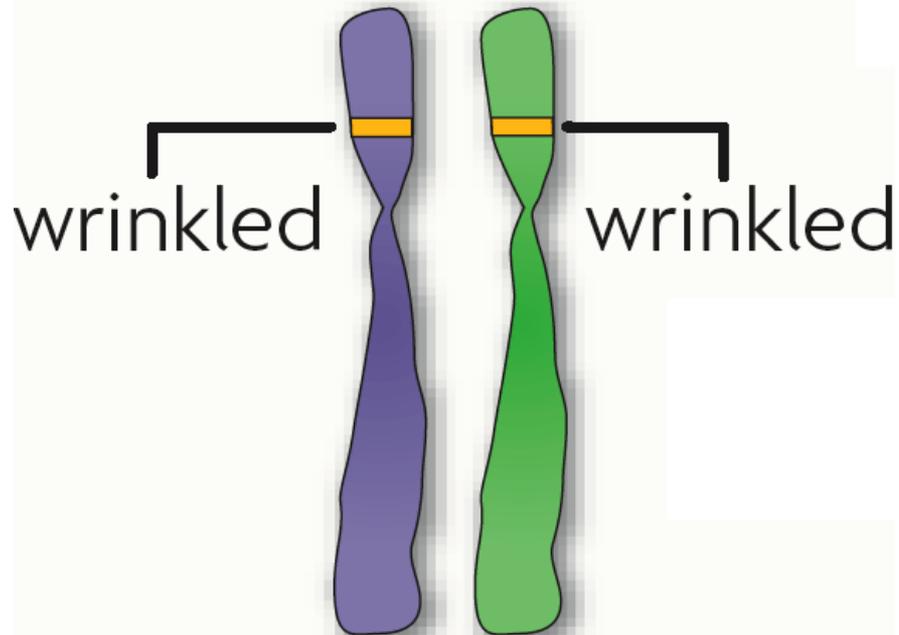


- **Gene:** a segment of DNA or a chromosome “ Mailbox ” with coded information for a particular trait or characteristic. Because animals are diploid (i.e., chromosomes in homologous pairs), there are 2 gene “Mailboxes” for each trait...one inherited from each parent.
- **Allele:** Actual DNA “ letters ” or N-base codes found in the gene “Mailbox”. One allele is inherited from each parent for each genetic trait.
  - Ex: **Gene** = Hexosaminidase A Lysosome enzyme for lipid recycling ;  
**Alleles** = normal vs mutated
- **Gene Notation:** letters are used to represent traits.  
 Rule: 1 letter for each trait

- ⊙ **Dominant:** alleles which hide or mask the information carried by recessive alleles. Notation = CAPITAL letters
  - > Ex: H = normal lysosome enzyme allele
- ⊙ **Recessive:** alleles which are only expressed or observed when NOT paired with a Dominant allele (i.e., need 2 recessive alleles to observe the trait. Notation = lowercase letters
  - > Ex: h = mutated lysosome enzyme allele
- ⊙ **Genotype:** the 2 alleles inherited for each genetic trait → the actual genetic info.
  - > **Homozygous:** 2 copies of the same allele for a trait. Ex: **HH** or **hh**
  - > **Heterozygous:** 2 different alleles for a trait Ex: **Hh**
- ⊙ **Phenotype:** physical appearance of the expressed trait which can be seen.
  - > Ex: have the disease or are normal and symptom free? (i.e., sick or healthy?)

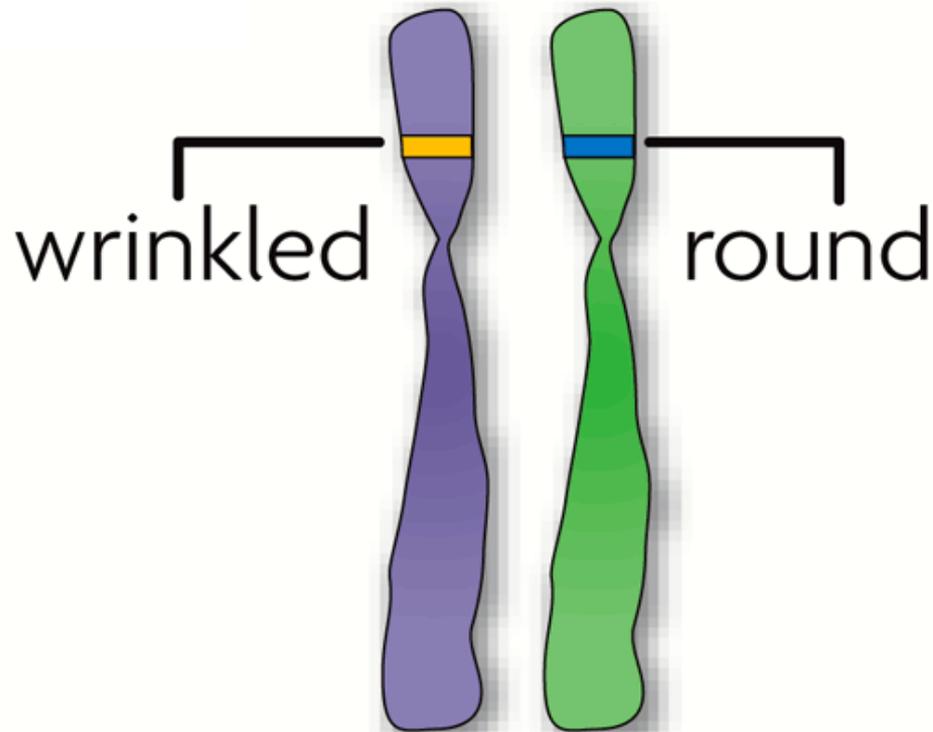
# Let's Review

- Homozygous or heterozygous?



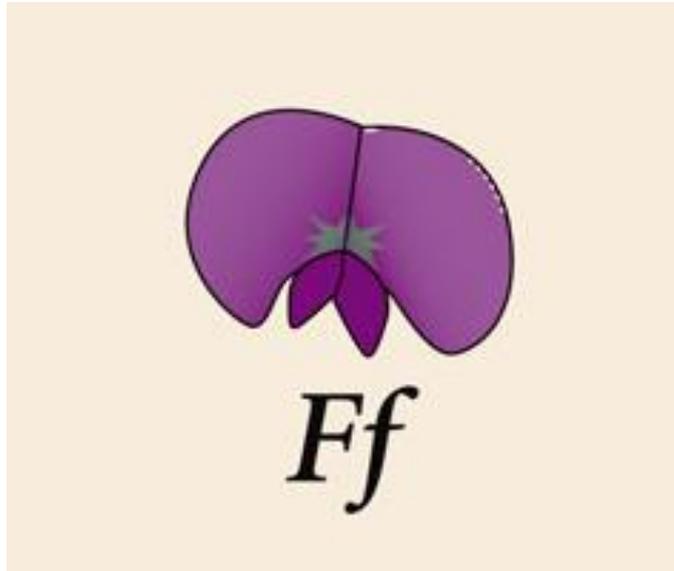
# Let's Review

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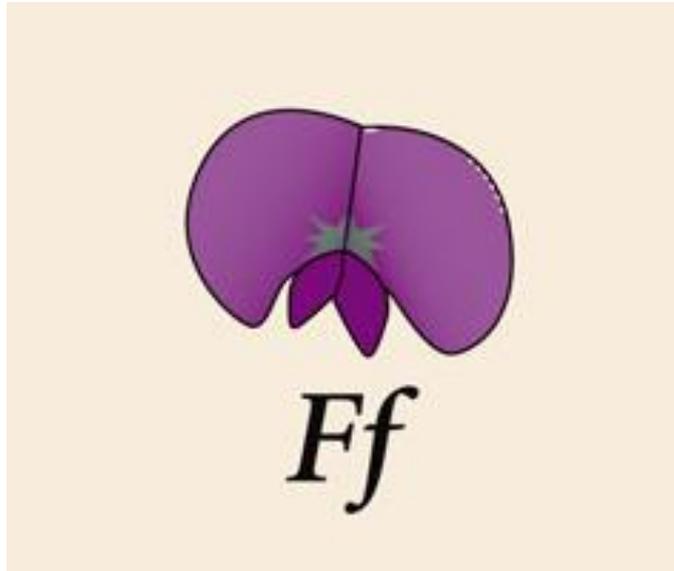
# Let's Review

- What is this flower's **genotype**?



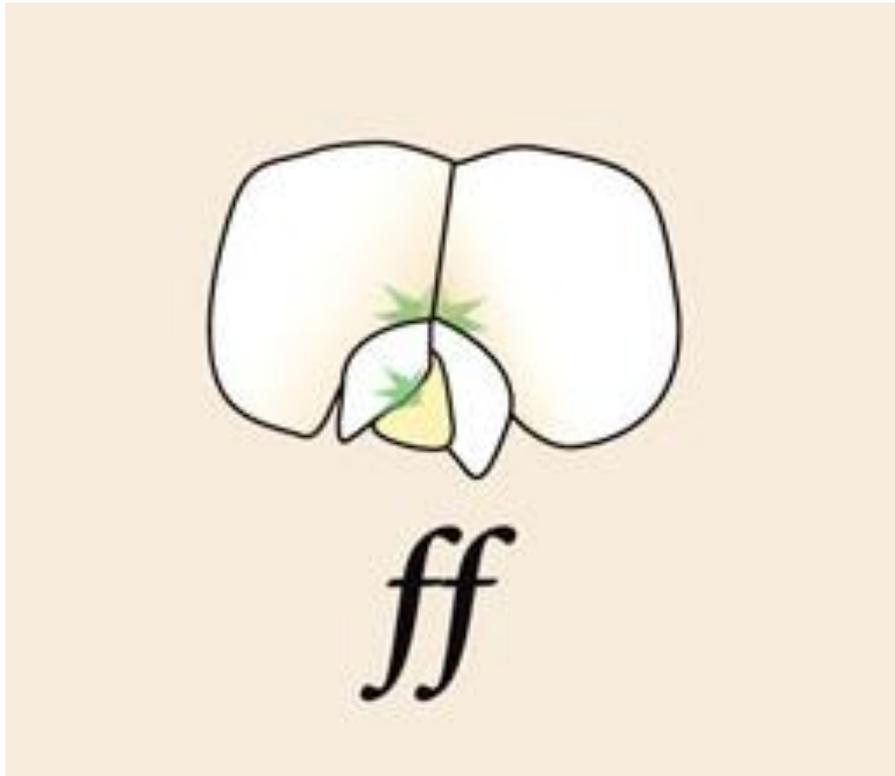
# Let's Review

- What is this flower's **phenotype**?



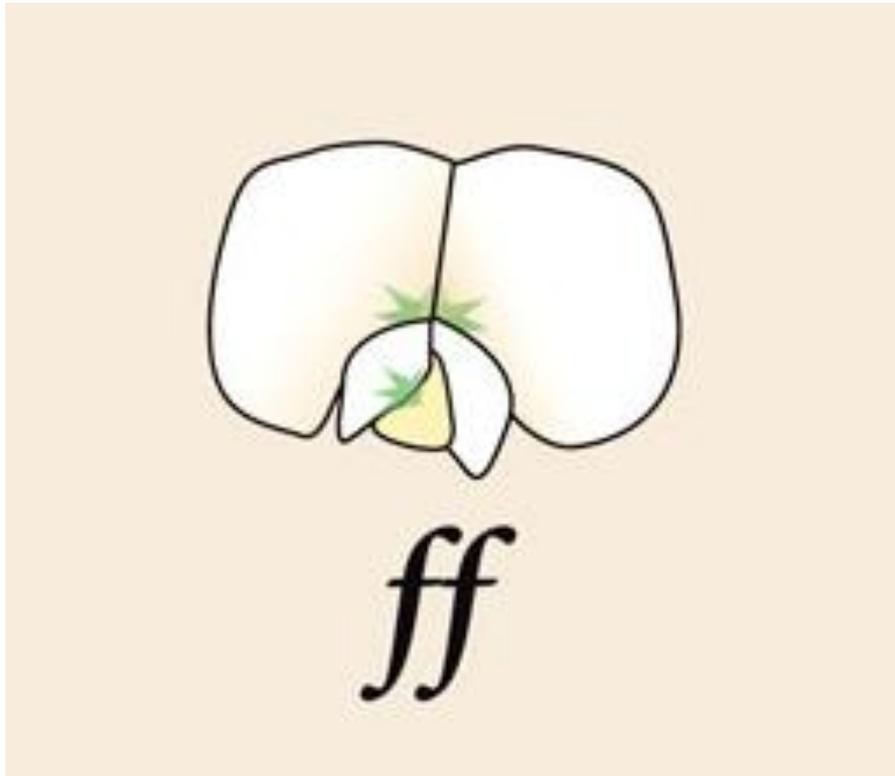
# Let's Review

- What is this flower's **phenotype**?



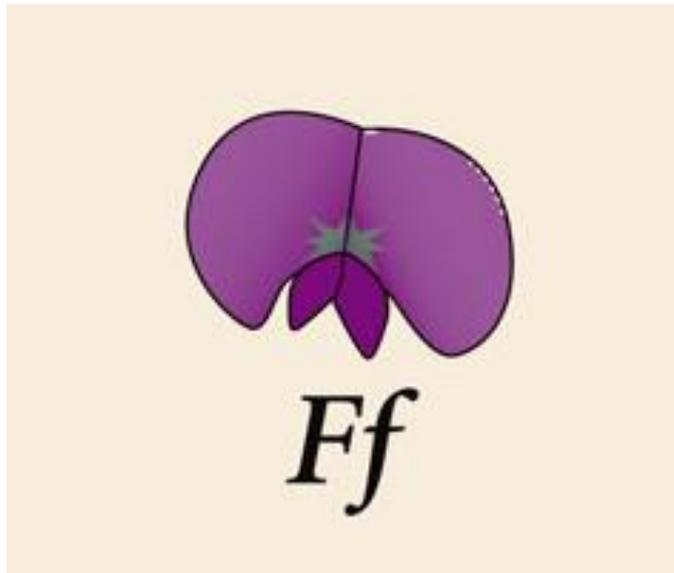
# Let's Review

- What is this flower's **genotype**?

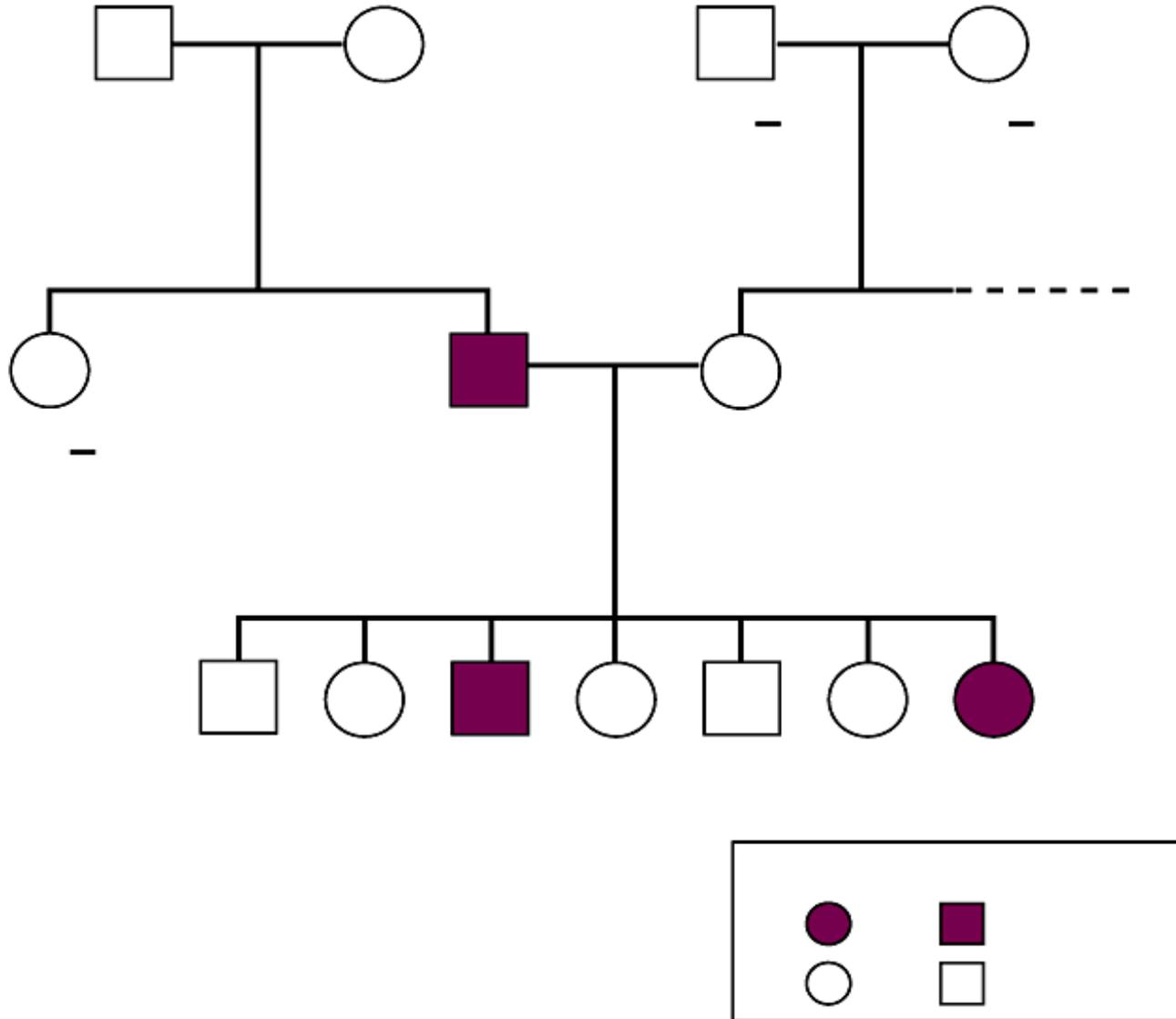


# Let's Review

- Is this flower **homozygous or heterozygous?**



- **Pedigree:** a family tree picture showing how a certain trait is inherited over several generations



# CAUTION

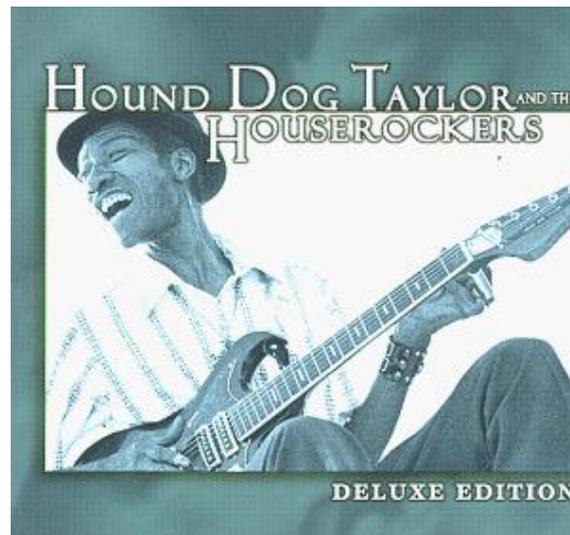
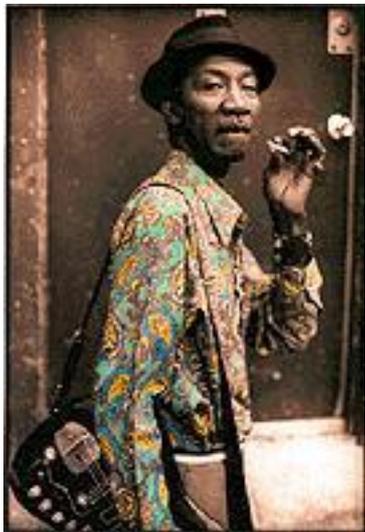
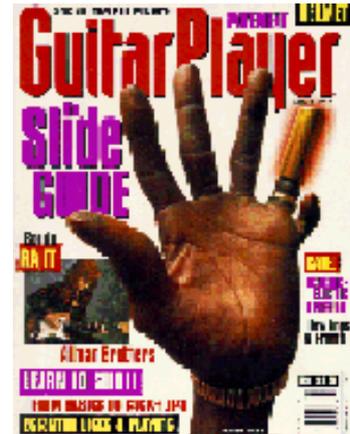
- Dominant alleles are usually frequently seen traits, but they are not necessarily always common in the gene pool
- Example = **Polydactyly** in humans (extra digits) is a dominant allele



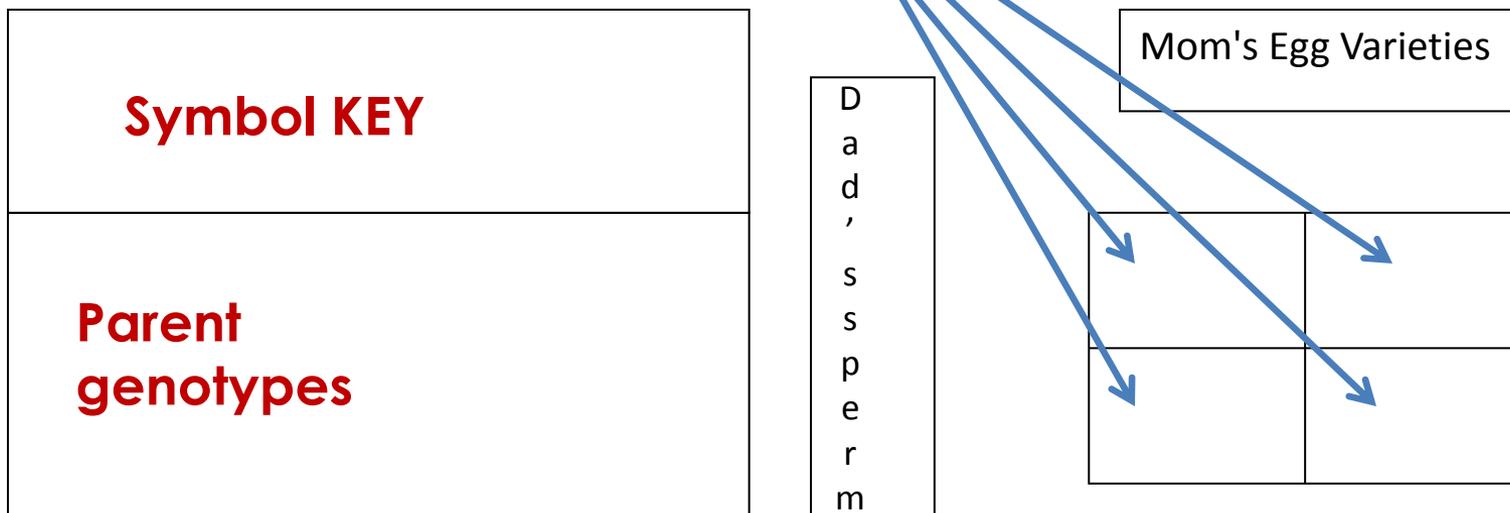
# More Polydactyly



# Hound Dog Taylor



- **Punnett square:** a box like diagram used to predict the probability of expected outcomes for a genetic cross
- Punnett square **Quick Tips:**
  - Define your Dominant & recessive letter symbols: make a key
  - Write each Parent's genotype
  - Fill in Mom's egg and Dad's sperm varieties along the PS sides
  - Complete the PS boxes to see all the possible genetic cross outcomes or kid's genotypes



# Topic 3: ONE-Trait Crosses

- The inheritance of many **human traits** follows Mendel's principles and the rules of probability



Figure 9.8A

# A Punnett Square can predict the chances for inheriting a variety of simple human traits

Example 1: What is the chance that a child will have freckles if **DAD** is homozygous for freckles and **MOM** does not have freckles? 4/4

Gene Notation Symbols Used

**F** = Freckles

**f** = NO freckles

Parent Genotypes:

**MOM** ff

**DAD** FF

	<b>f</b>	<b>f</b>
<b>F</b>	<b>Ff</b>	<b>Ff</b>
<b>F</b>	<b>Ff</b>	<b>Ff</b>

Dominant Traits	Recessive Traits
 Freckles	 No freckles
 Widow's peak	 Straight hairline
 Free earlobe	 Attached earlobe

# A Punnett Square can predict the chances for inheriting a variety of simple human traits

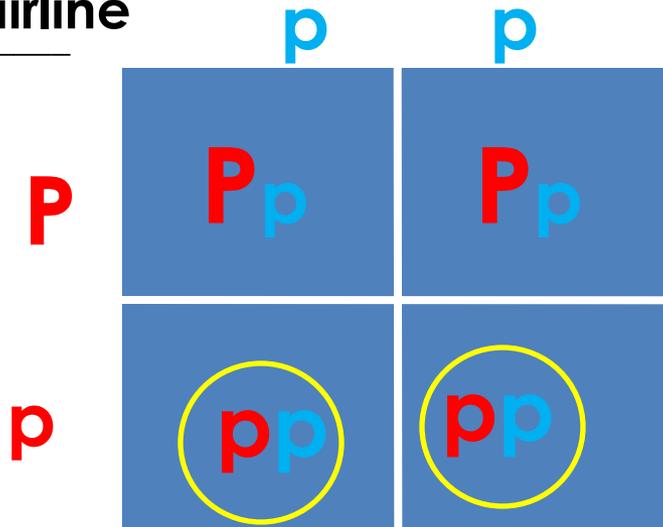
Example 2: What is the chance that a child will have a straight hairline if **DAD** is heterozygous for widow's peak and **MOM** has a straight hairline? \_\_\_\_\_ **2/4**

Gene Notation Symbols Used

**P** = Widow's Peak

**p** = Straight hairline

Parent Genotypes:  
**MOM** pp  
**DAD** Pp



Dominant Traits	Recessive Traits
 Freckles	 No freckles
 Widow's peak	 Straight hairline
 Free earlobe	 Attached earlobe

- The inheritance of many human genetic diseases also follows Mendel's principles and the rules of probability



# Genetic disorders are caused by **Dominant** or **Recessive** mutations

## SOME AUTOSOMAL DISORDERS IN HUMANS

Disorder	Major Symptoms	Incidence	Comments
<b>Recessive disorders</b>			
Albinism	Lack of pigment in skin, hair, and eyes	$\frac{1}{22,000}$	Very easily sunburned
Cystic fibrosis	Excess mucus in lungs, digestive tract, liver; increased susceptibility to infections; death in infancy unless treated	$\frac{1}{1,800}$ Caucasians	See Modules 9.9 and 12.11
Galactosemia	Accumulation of galactose in tissues; mental retardation; eye and liver damage	$\frac{1}{100,000}$	Treated by eliminating galactose from diet
Phenylketonuria (PKU)	Accumulation of phenylalanine in blood; lack of normal skin pigment; mental retardation	$\frac{1}{10,000}$ in U.S. and Europe	See Module 9.10
Sickle-cell disease (homozygous)	Sickled red blood cells; damage to many tissues	$\frac{1}{500}$ African Americans	Alleles are codominant; see Modules 9.13–9.15
Tay-Sachs disease	Lipid accumulation in brain cells; mental deficiency; blindness; death in childhood	$\frac{1}{3,500}$ Jews from central Europe	See Module 4.12
<b>Dominant disorders</b>			
Achondroplasia	Dwarfism	$\frac{1}{25,000}$	See Module 9.9
Alzheimer's disease (one type)	Mental deterioration; usually strikes late in life	Not known	
Huntington's disease	Mental deterioration and uncontrollable movements; strikes in middle age	$\frac{1}{25,000}$	See Modules 9.9 and 12.11
Hypercholesterolemia	Excess cholesterol in blood; heart disease	$\frac{1}{500}$ are heterozygous	Incomplete dominance; see Module 9.12

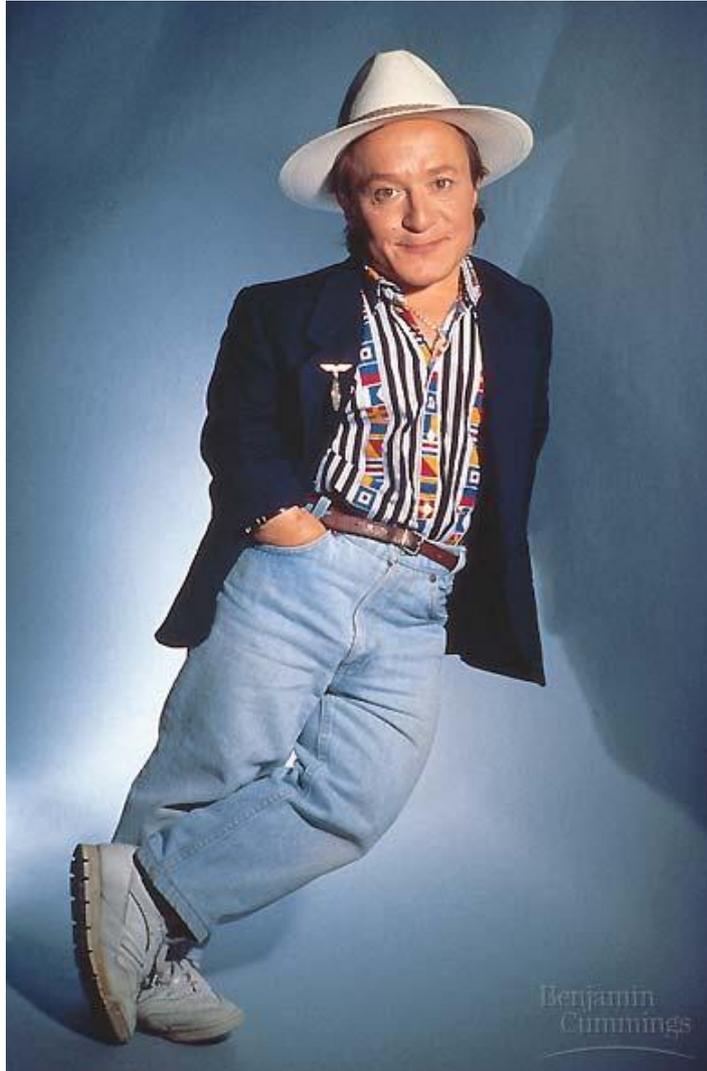
# Albinism...a recessive mutation

**albino  
Africans**





- Dwarfism .... A dominant mutation



Example 3: XP disorder is caused by a **recessive** mutation. What is the chance that a couple will have a child affected with XP if MOM is a heterozygous carrier and DAD has XP? 1/2

Gene Notation Symbols Used:

**R** : Normal RE code

**r** : Broken code

Parent Genotypes:

MOM: **Rr**

DAD: **rr**

Mom's Alleles

**R**

**r**

**r**

**Rr**

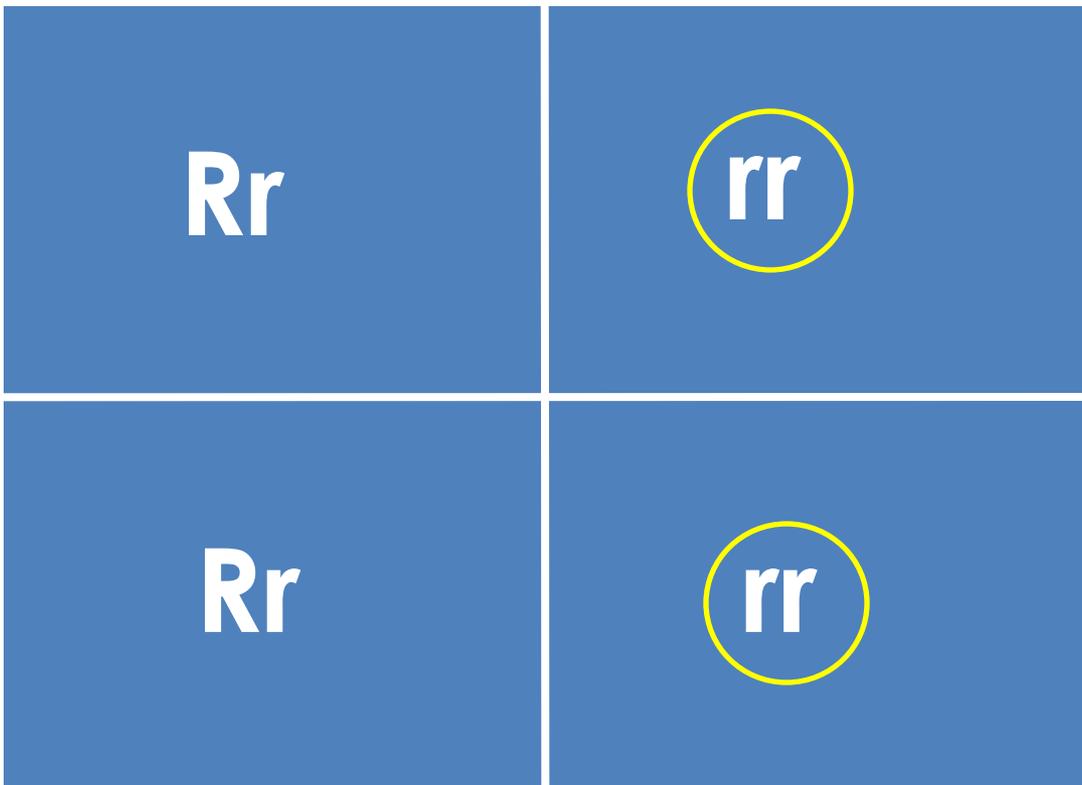
**rr**

Dad's Alleles

**r**

**Rr**

**rr**



Example 4: Huntington's Disease (HD) is caused by a **Dominant** mutation. What is the chance that a couple will have a child affected with HD if MOM is heterozygous for HD and DAD is homozygous normal and disease free? 1/2

Gene Notation Symbols Used:

**H** : **Broken code**

**h** : **Normal code**

Parent Genotypes:

MOM: **Hh**

DAD: **hh**

Dad's  
Alleles

Mom's  
Alleles

	<b>H</b>	<b>h</b>
<b>h</b>	<b>Hh</b>	<b>hh</b>
<b>h</b>	<b>Hh</b>	<b>hh</b>

Example 5: Dwarfism(HD) is caused by a **Dominant** mutation. What is the chance that a couple who are both dwarfs will have a child with NORMAL height if MOM and DAD are heterozygous for dwarfism? 1/4

Gene Notation Symbols Used:

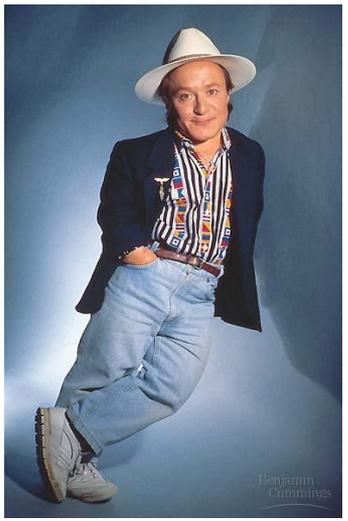
**D** : Broken code

**d** : Normal code

Parent Genotypes:

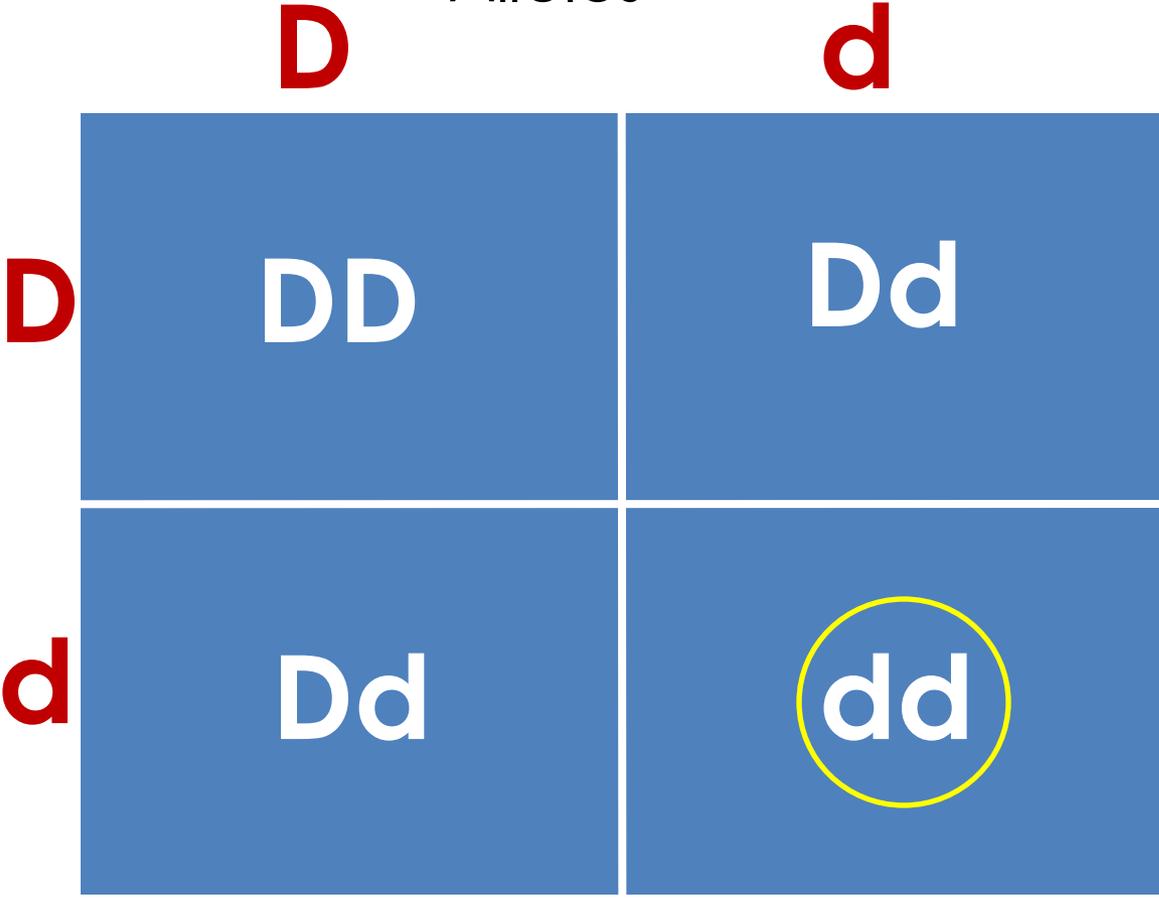
MOM: **Dd**

DAD: **Dd**



Dad's Alleles

Mom's Alleles



# Let's Review

- Genetic diseases usually happen in 1 of 2 ways:

1) A person inherits 1 or more mutated DNA codes => broken proteins inside cells that don't work correctly

**Examples?**

Remember:

A) If the mutation is Dominant => any person who inherits at least 1 mutation will show the disease

B) If the mutation is Recessive => only people who inherit 2 mutations will show the disease

C) If the mutation is Recessive => people who inherit only 1 mutation will be healthy carriers who could pass the mutation to their kids

2) A person inherits the wrong # of chromosomes => confusion during development

**Examples?**

- Notice anything wrong with this karyotype?

**Trisomy 21**

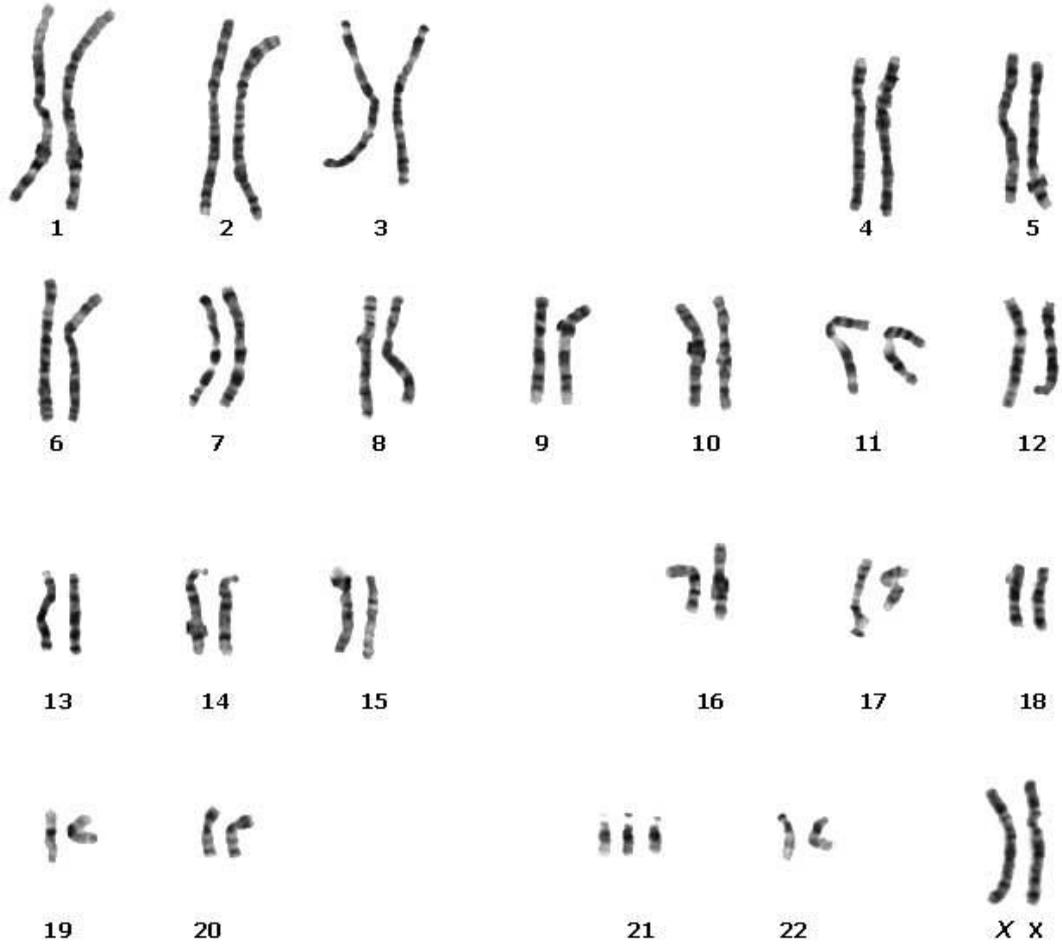


Figure 8.20Ax

# An extra copy of chromosome 21 causes Down Syndrome

- This karyotype shows three number 21 chromosomes

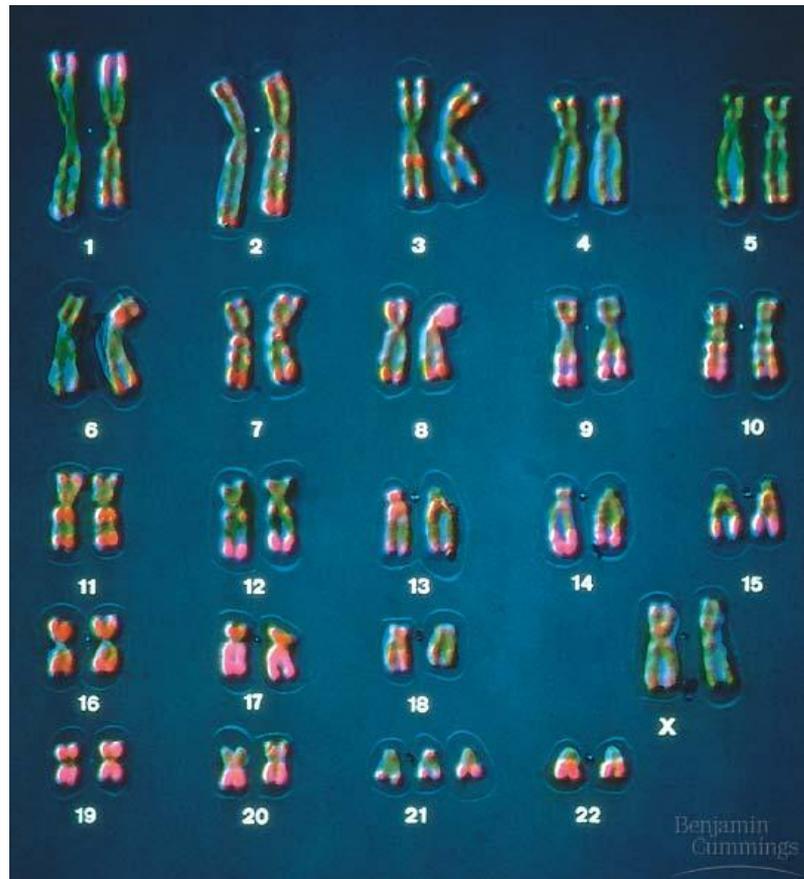


Figure 8.20A, B

- The chance of having a Down syndrome child goes up with MOM's age ...WHY?

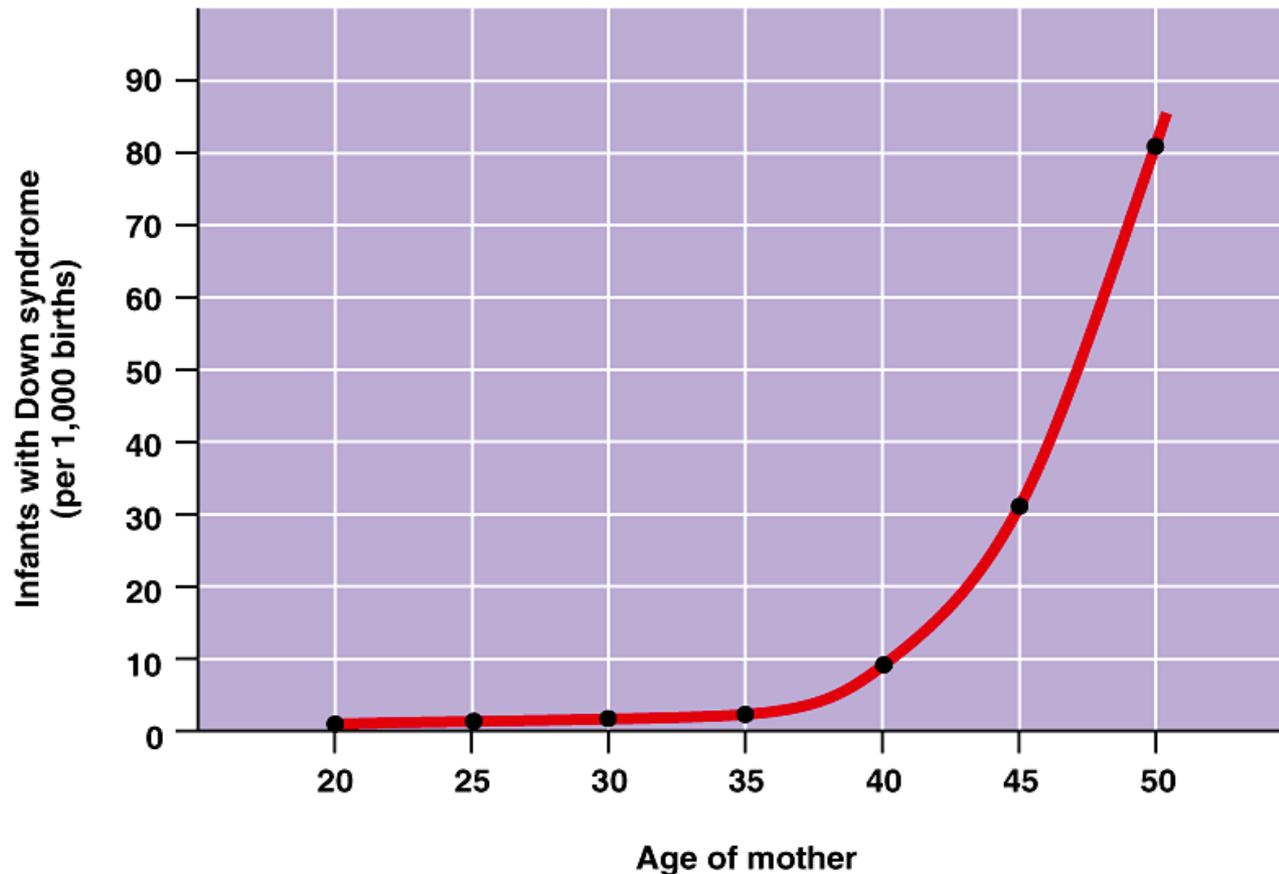


Figure 8.20C

# Geneticists use the testcross to determine unknown genotypes

- The offspring of a testcross often reveal the unknown **genotype** of a **Dominant** individual
- HOW: breed the unknown dog (BLACK) with a dog showing the **recessive** trait (chocolate)

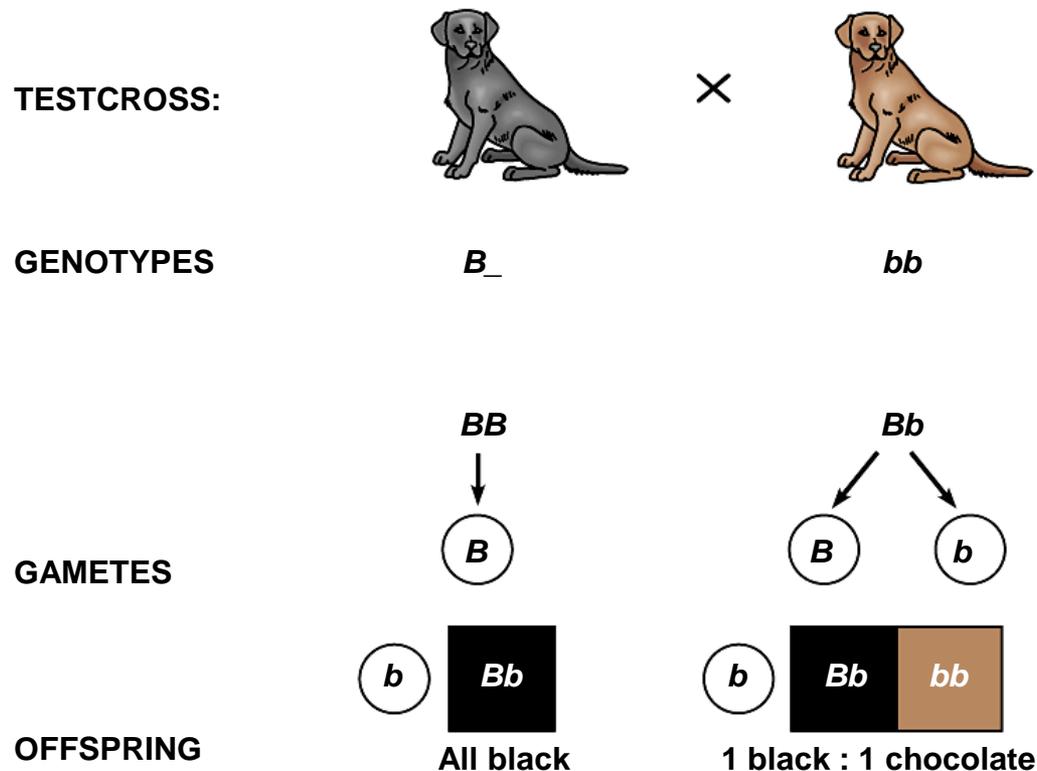


Figure 9.6

# Lethal allele combinations: A “double” dose of certain alleles is fatal during development

- Cause of many miscarriages?
- Examples:
  - Mexican Hairless Dogs



Mexican Hairless dogs contain the dominant lethal allele and are heterozygous (Hh). If a dog inherited both dominant alleles for the hairless trait and was homozygous (HH), it would die as an embryo. If a dog were homozygous recessive (hh) it would have hair.



Why don't dog breeders usually cross 2 Mexican hairless dogs together ???

Complete the Punnett square to find out?

Female's genotype: Hh (hairless)

Male's genotype: Hh (hairless)

	H	h
H	HH	Hh
h	Hh	hh



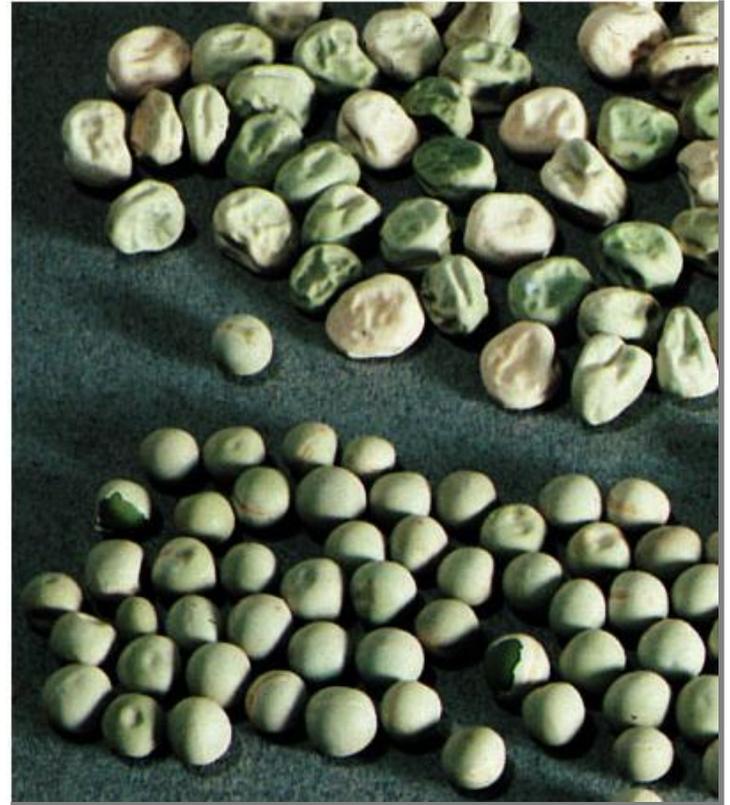
**Result:**  
**Fatal miscarriage**  
**chance = 1/4**

## **Topic 4: TWO-Trait Crosses**

**Mendel continued his studies with pea plants by following the inheritance of 2 different traits at the same time**

# Dihybrid cross

- Mendel's experiments continued by following the inheritance of 2 different characters
  - seed color and seed shape
  - dihybrid crosses



# Introducing the law of independent assortment that applies when tracking two characters at once

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## ■ Law of independent assortment

- Each pair of alleles segregates independently of the other pairs of alleles during gamete formation
- What are the 4 possible gamete types that are created in equal amounts during Meiosis for the genotype ***RrYy*** ?

**Hint: time to FOIL**

1) ***RY***   2) ***Ry***   3) ***rY***   4) ***ry***

# The **Law of Independent Assortment** is revealed by tracking two characteristics at once (AKA Dihybrid Cross)

Mom's genotype: BbRr (Black fur & running mouse)

possible egg varieties: BR Br bR br

Dad's genotype: BbRr (Black fur & running mouse)

possible sperm varieties: BR Br bR br

	BR	Br	bR	br
BR	BBRR	BBRr	BbRR	BbRr
Br	BBRr	BBrr	BbRr	Bbrr
bR	BbRR	BbRr	bbRR	bbRr
br	BbRr	Bbrr	bbRr	bbrr

Phenotype Probabilities:

-  Black fur, Running:  $\frac{9}{16}$
-  Black fur, waltzing:  $\frac{3}{16}$
-  Brown fur, Running:  $\frac{3}{16}$
-  Brown fur, waltzing:  $\frac{1}{16}$

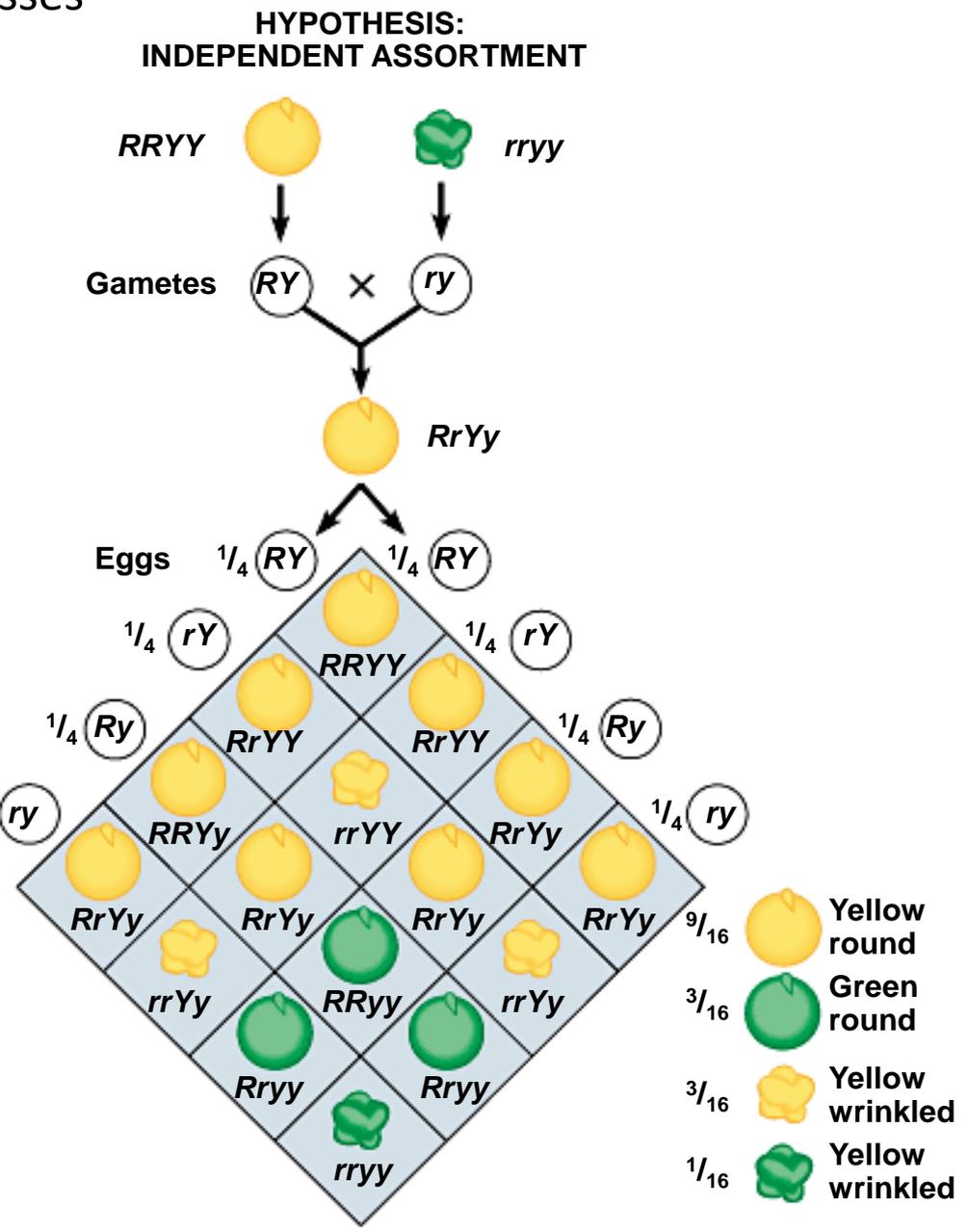
B = black fur in mice b = brown fur R = running mice r = waltzing mice

- When Mendel studied controlled crosses for **2 traits** over 2 generations he discovered consistent F<sub>2</sub> phenotype fractions:

- Show both dominant traits 9/16
- Show 1 dominant & 1 recessive 3/16
- Show other dominant & recessive 3/16
- Show both recessive traits 1/16

- WHY? .... Mendel again concluded that the rules of math probability will explain these F<sub>2</sub> phenotype fractions only if the alleles for 1 trait segregate independently of the other trait's allele pairs during Meiosis gamete formation

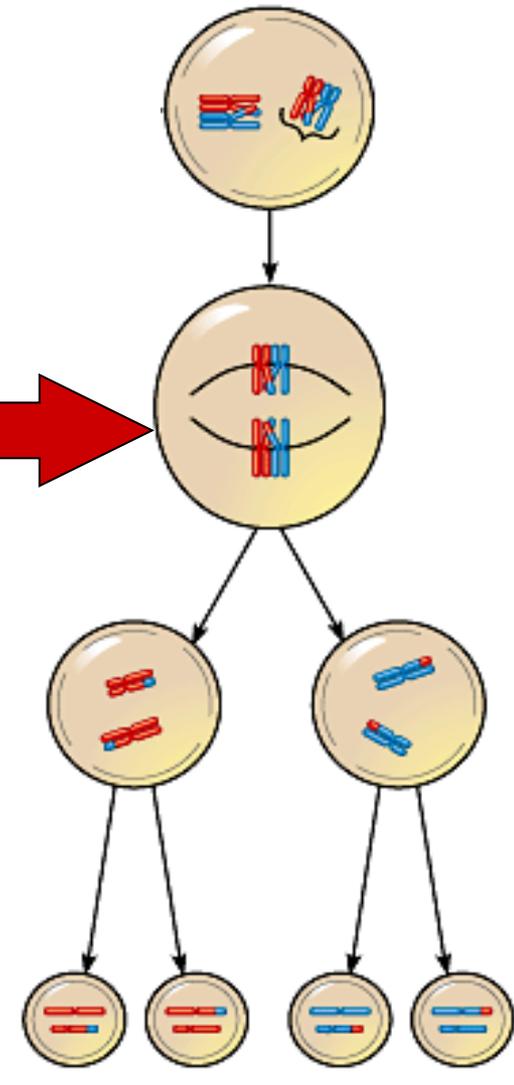
- This is known as the Law of Independent Assortment



# Law of Independent Assortment

- Which stage of meiosis creates the law of independent assortment?

Metaphase 1



## EXCEPTION

- Won't work if genes are on **same chromosome** & close together
- This means the two genes travel together during meiosis instead of going separate ways



**Hypothesis: Dependent assortment**

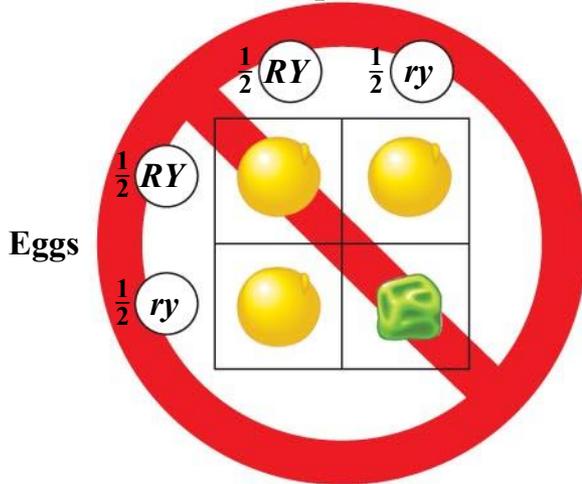
**P generation**



**F<sub>1</sub> generation**



Sperm

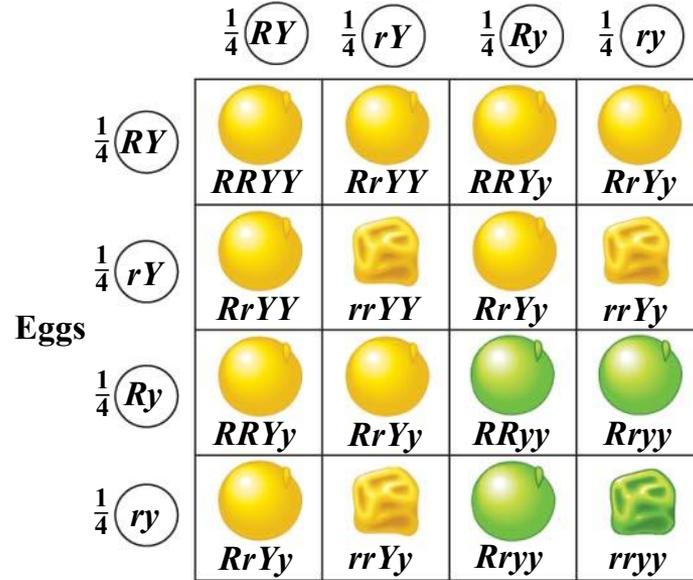


**Hypothesized  
(not actually seen)**

**Hypothesis: Independent assortment**



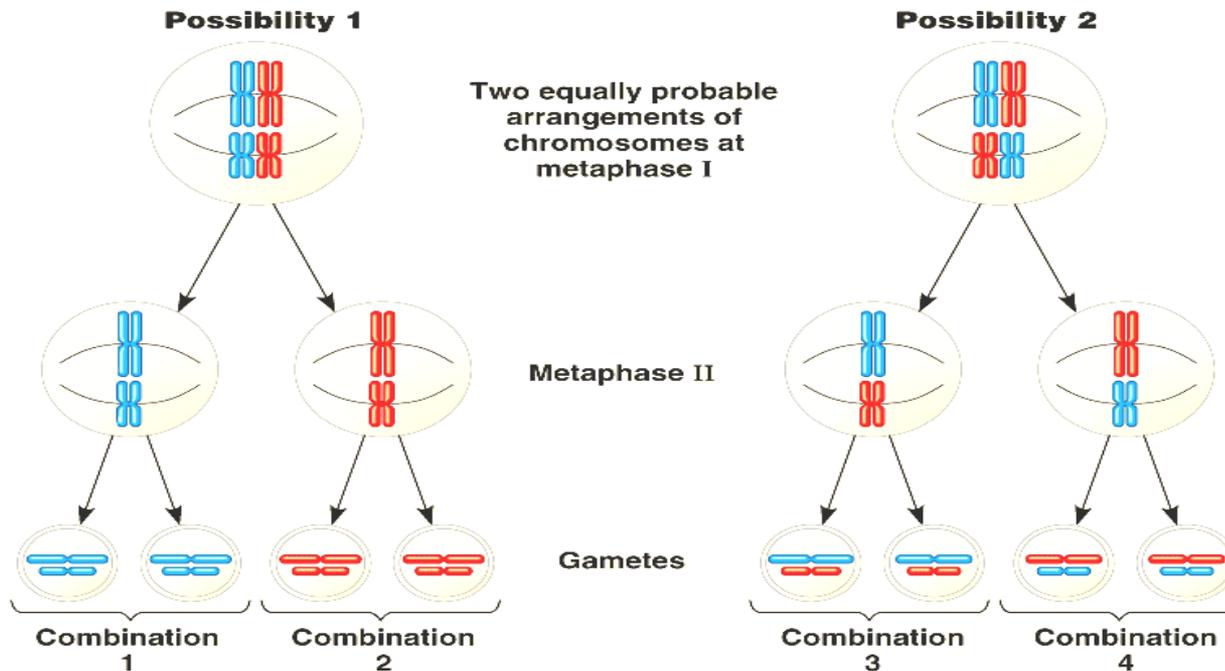
Sperm



**Actual results  
(support hypothesis)**

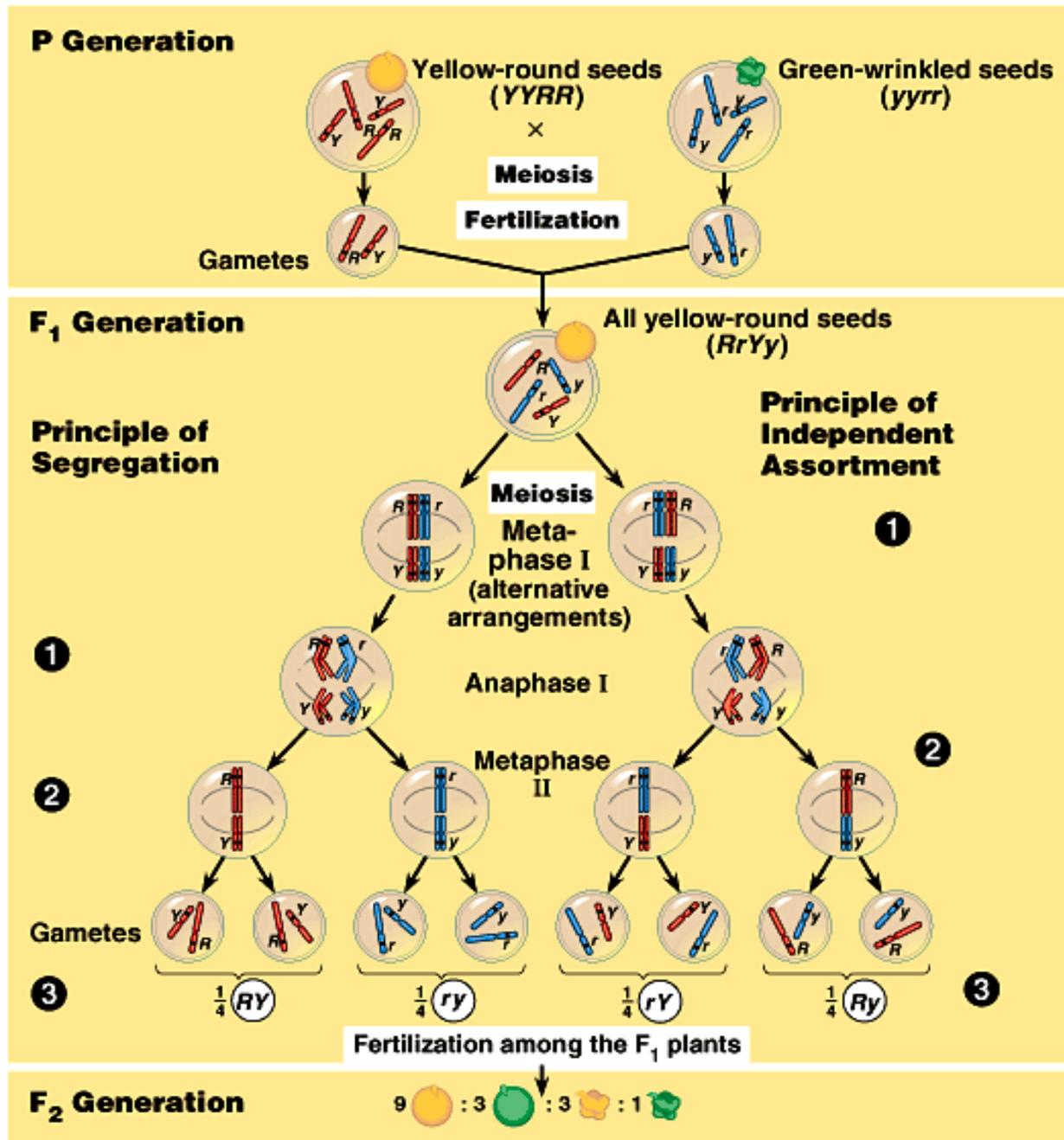
- $\frac{9}{16}$   Yellow round
- $\frac{3}{16}$   Green round
- $\frac{3}{16}$   Yellow wrinkled
- $\frac{1}{16}$   Green wrinkled

Basically, for any 2 traits that you might pay attention to as you go through Meiosis to make a gamete, the 2 chromosomes carrying those alleles can line up in different random ways during Metaphase I, and that means that the gamete produced might have both alleles from your DAD, both alleles from your MOM, or 1 allele from MOM and 1 allele from DAD.



The chromosomal basis of Mendel's laws...

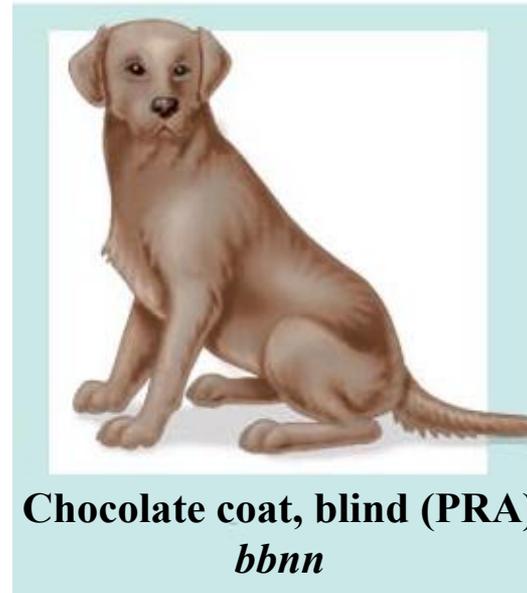
Trace the genetic events through meiosis, gamete formation & fertilization to offspring



Look what happens when you cross a lab that breed pure for BLACK & NORMAL VISION with one that breeds pure for CHOCOLATE & BLIND



**X**



**= ?**

ALL the  $F_1$  pups would look like this



**X**



**= ?**

Look what we get if the  $F_1$  pups breed together

# F<sub>2</sub> phenotypes

9/16

3/16

3/16

1/16



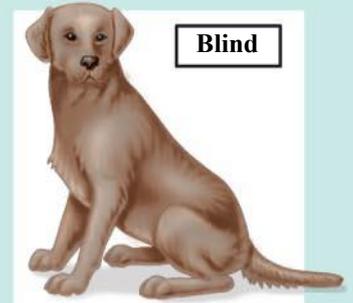
Black coat, normal vision  
*B\_N\_*



Black coat, blind (PRA)  
*B\_nn*



Chocolate coat, normal vision  
*bbN\_*



Chocolate coat, blind (PRA)  
*bbnn*

Phenotypes  
Genotypes

Mating of heterozygotes  
(black, normal vision)

Phenotypic ratio  
of offspring

*BbNn* × *BbNn*



9 black coat,  
normal vision

3 black coat,  
blind (PRA)

3 chocolate coat,  
normal vision

1 chocolate coat,  
blind (PRA)

Conclusion: Mendel's Dihybrid results with pea plants also apply to animals like dogs

1. A horse that is heterozygous for black coat color and trotting gait is mated with a horse with a chestnut coat color and pacing gait.

\*\* Black coat color is dominant over chestnut and trotting gait is dominant over pacing gait

<b>Gene notation</b>	<u><b>B</b></u> = <b>Black coat</b>
	<u><b>b</b></u> = <b>Chestnut coat</b>
<b>Symbols used</b>	<u><b>T</b></u> = <b>Trotting gait</b>
	<u><b>t</b></u> = <b>Pacing gait</b>
Parent MOM =	<u><b>BbTt</b></u>
Genotypes DAD =	<u><b>bbtt</b></u>
Mom's egg varieties:	<b>BT Bt bT bt</b>
Dad's sperm varieties:	<b>bt</b>

	BT	bT	Bt	bt
bt	BbTt	bbTt	Bbtt	bbtt

Genotypes

Phenotypes

BbTt

**Black & Trotting**

bbTt

**Chestnut & Trotting**

Bbtt

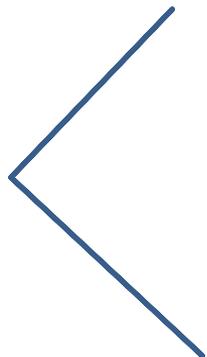
**Black & Pacing**

bbtt

**Chestnut & Pacing**

1/4

1/4



Sheila is right-handed and albino (and her mother is left-handed). Doug is left-handed and has normal skin pigmentation, but both of his maternal grandparents are albino. What is the probability of Sheila and Doug having a right-handed child with normal skin pigmentation?

\*\* Right-handedness is dominant to left-handedness and normal skin pigmentation is dominant over the albino skin condition.

<b>Gene notation</b>	<u><b>R</b></u> = <b>Right-handed</b>
	<u><b>r</b></u> = <b>Left handed</b>
<b>Symbols used</b>	<u><b>N</b></u> = <b>Normal pigment</b>
	<u><b>n</b></u> = <b>albino</b>

Parent	MOM = <u><b>Rrnn</b></u>
Genotypes	DAD = <u><b>rrNn</b></u>

Mom's egg varieties:	<b>Rn</b> <b>rn</b>
Dad's sperm varieties:	<b>rN</b> <b>rn</b>

	Rn	rn		
rN	RrNn	rrNn		
rn	Rrnn	rrnn		

What is the probability of Sheila and Doug having a right-handed child with normal skin pigmentation? 1/4

# Review: Mendel's laws of heredity

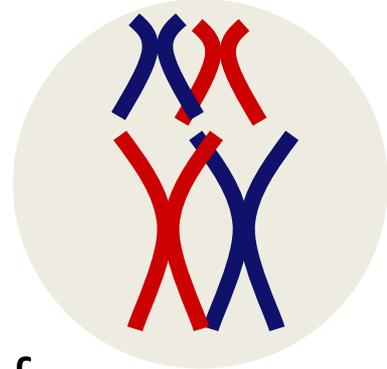
- Law of segregation

- Applies to inheritance of single traits
- **Answers the question:** Why don't 2 alleles ever end up in the same gamete?
- each allele **segregates** (goes a different direction during Anaphase 1) into separate gametes

- Law of independent assortment

- Applies to inheritance of 2 traits
- genes on separate chromosomes assort into gametes independently
- Happens because of the random arrangement of chromosome pairs during Metaphase 1 (Mom..Mom..Dad..Mom...etc, lineup is different each time)
- **EXCEPTION:** won't work if the 2 traits are located as neighbors on the same chromosome = linked genes

metaphase1



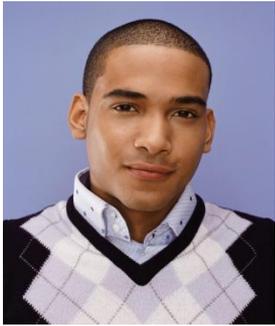
# **Topic 5: Deeper Mysteries of Heredity**

**AKA ... Beyond what  
Mendel could explain**

# 6.3 Mendel and Heredity

## KEY CONCEPT

**Phenotype is affected by many different factors.**



Some traits are determined by more than the simple interaction of dominant and recessive alleles

- **Incomplete Dominance** happens when a trait is a blend of the two alleles
- Example 1: Crossing green and steel blue betta fish creates a blended Royal blue fish



Green

+



Steel Blue

=



Royal blue

Some traits are determined by more than the simple interaction of dominant and recessive alleles

- Another example of **Incomplete Dominance** happens with some flowers
- Example 2: Crossing red and white flowers creates a blended Pink flower



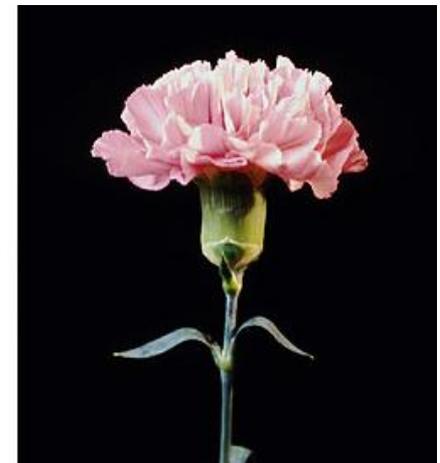
**Red**

+



white

=



**Pink**

- Hypercholesterolemia is an example of **Incomplete dominance** in humans

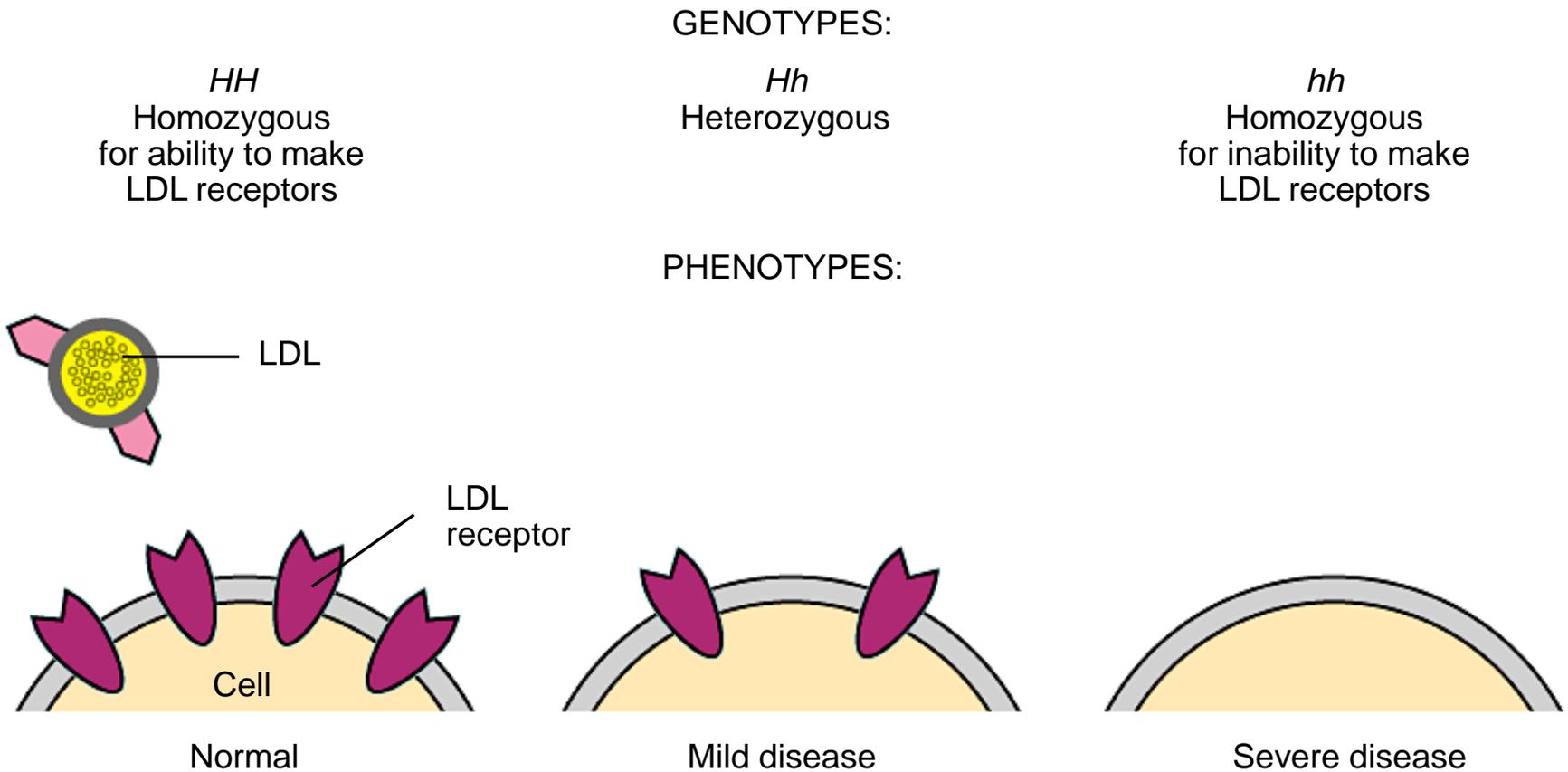


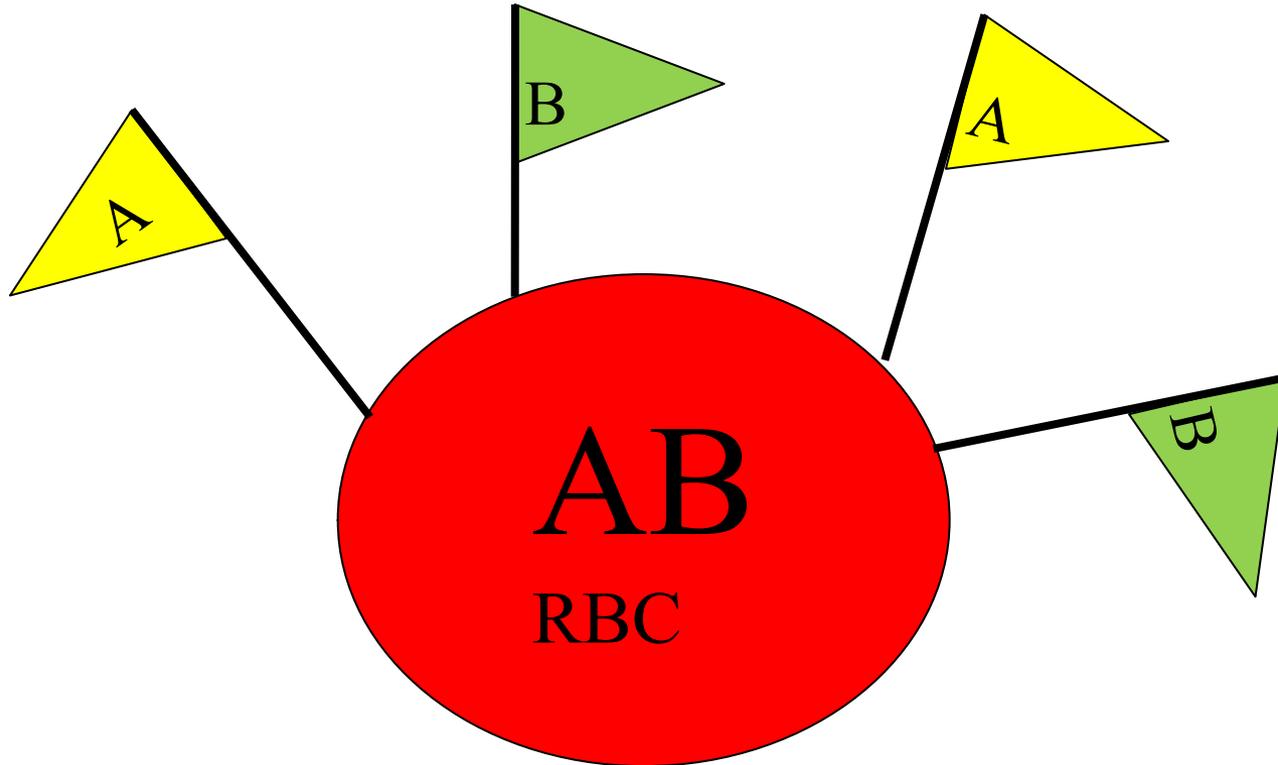
Figure 9.12B

Some traits are determined by more than the simple interaction of dominant and recessive alleles

- **Codominance** is another more complex situation that happens when two alleles are both expressed together at the same time
- Example 1: Some people inherit an A blood allele from one parent and a B blood allele from the other parent and end up expressing both codes with AB blood

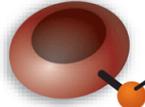
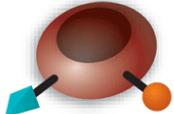
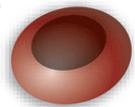
**A allele** + **B allele** = **AB blood**

- Sometimes the 2 alleles for a trait both show at the same time = **Codominance**
- Examples:      Type AB blood in humans



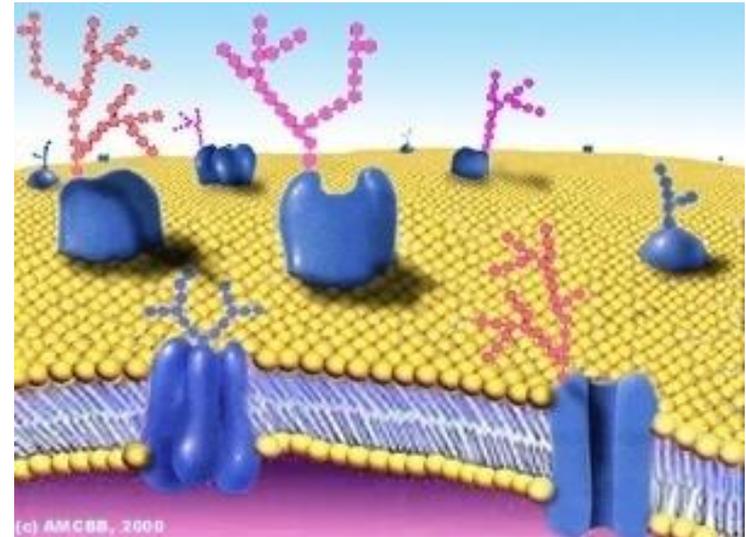
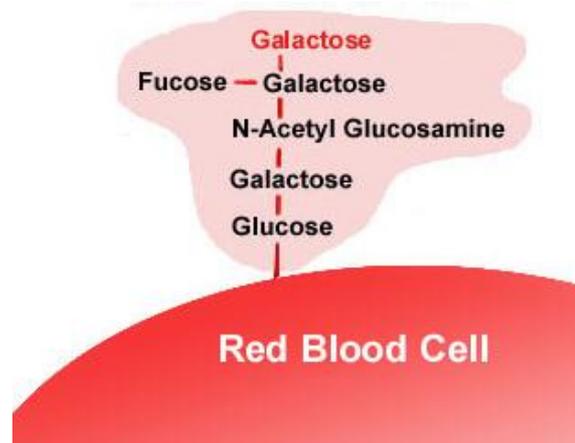
## 6.3 Mendel and Heredity

- Codominant alleles will both be completely expressed.

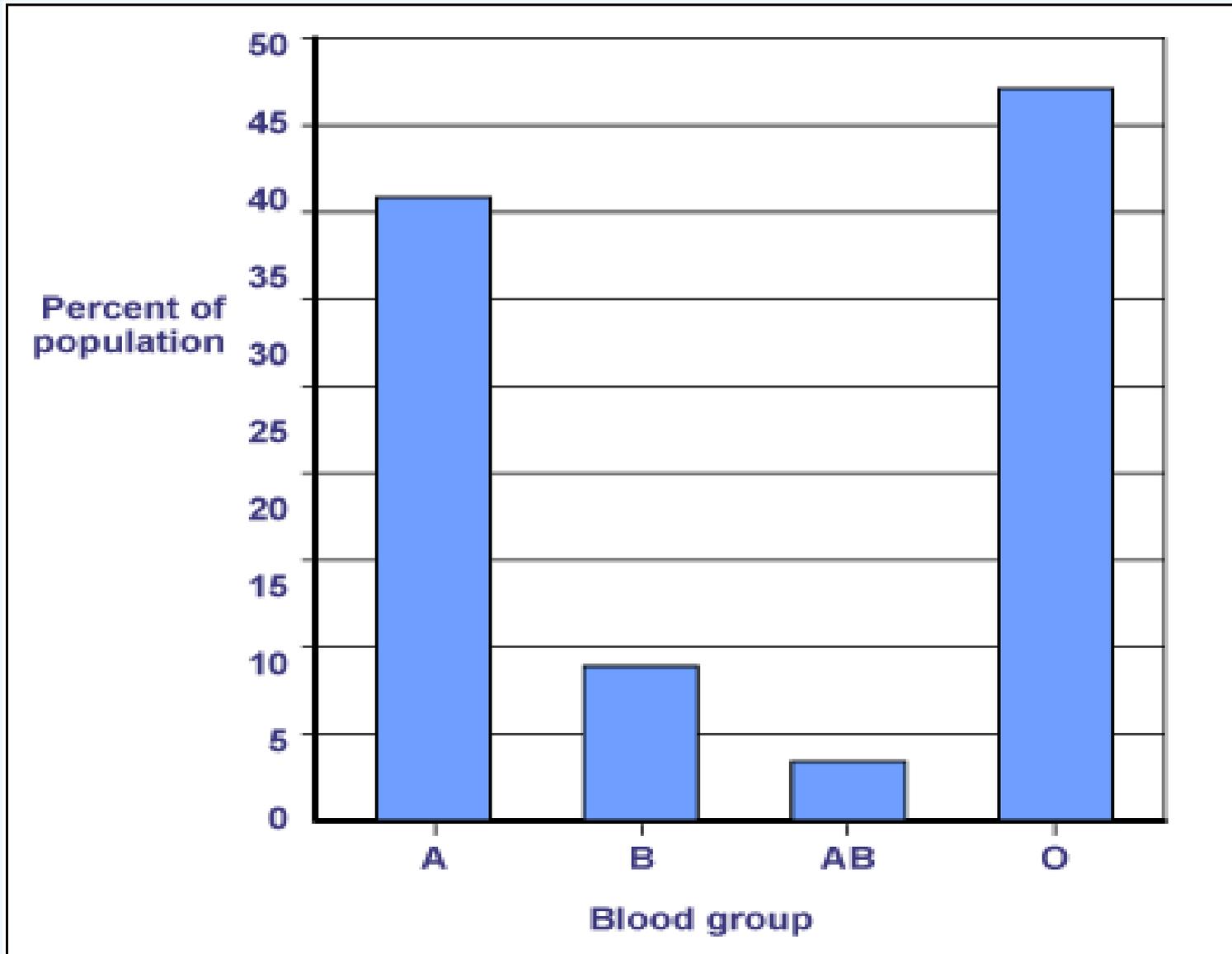
PHENOTYPE (BLOOD TYPE)		GENOTYPES
A	antigen A 	$I^A I^A$ or $I^A i$
B	 antigen B	$I^B I^B$ or $I^B i$
AB	both antigens 	$I^A I^B$
O	no antigens 	$ii$

Many genes have more than two alleles in the population = Multiple Alleles

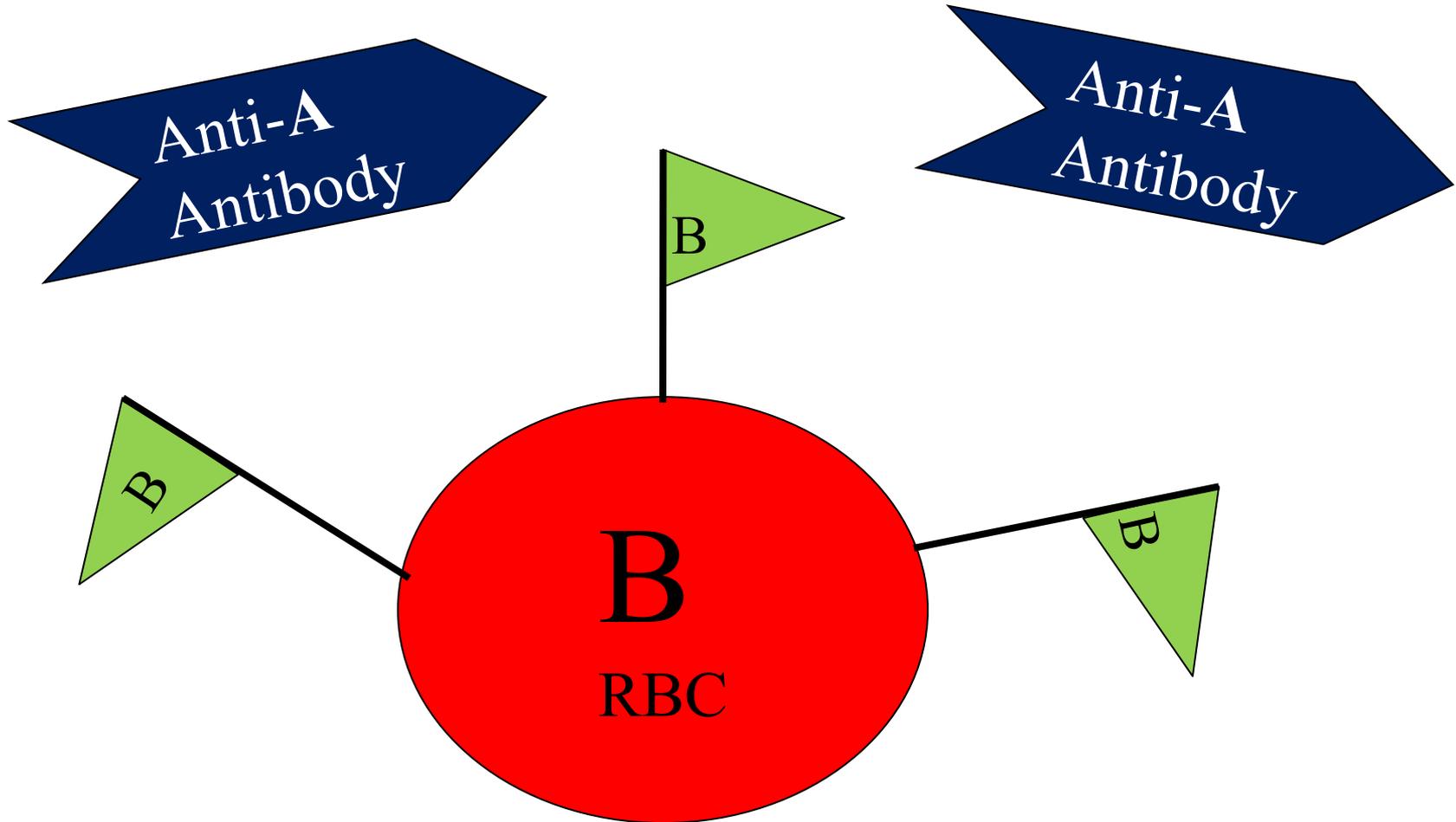
- Human ABO blood types are determined by 3 alleles in the human gene pool:
- The alleles for A and B blood types are codominant, and have the following symbols:  $I^A$   $I^B$
- The O allele is recessive =  $i$



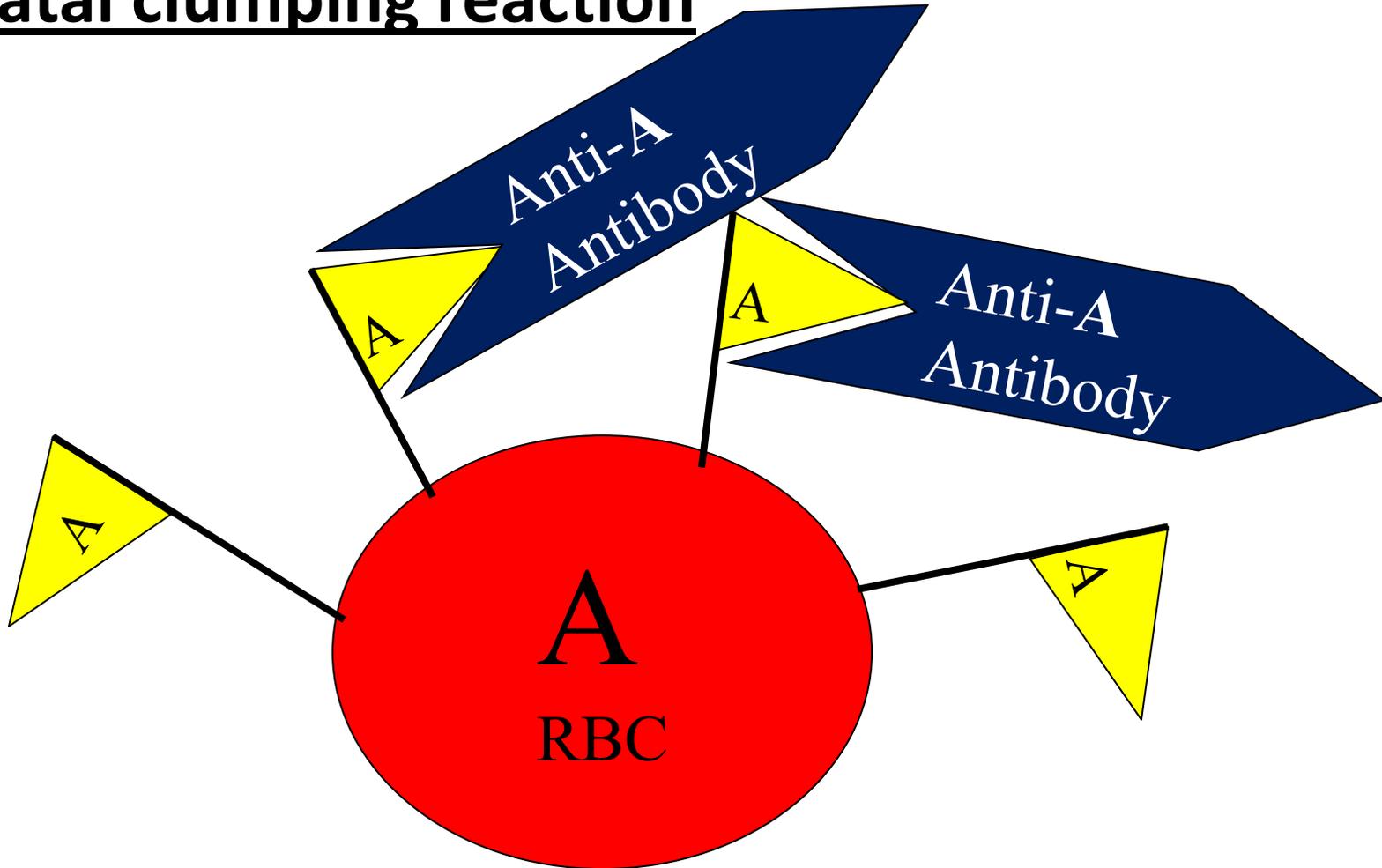
# Blood Type Frequencies



- People with B blood are born with antibodies against A type blood



- If B blood people receive A blood during a transfusion, the anti-A antibodies will cause a **fatal clumping reaction**

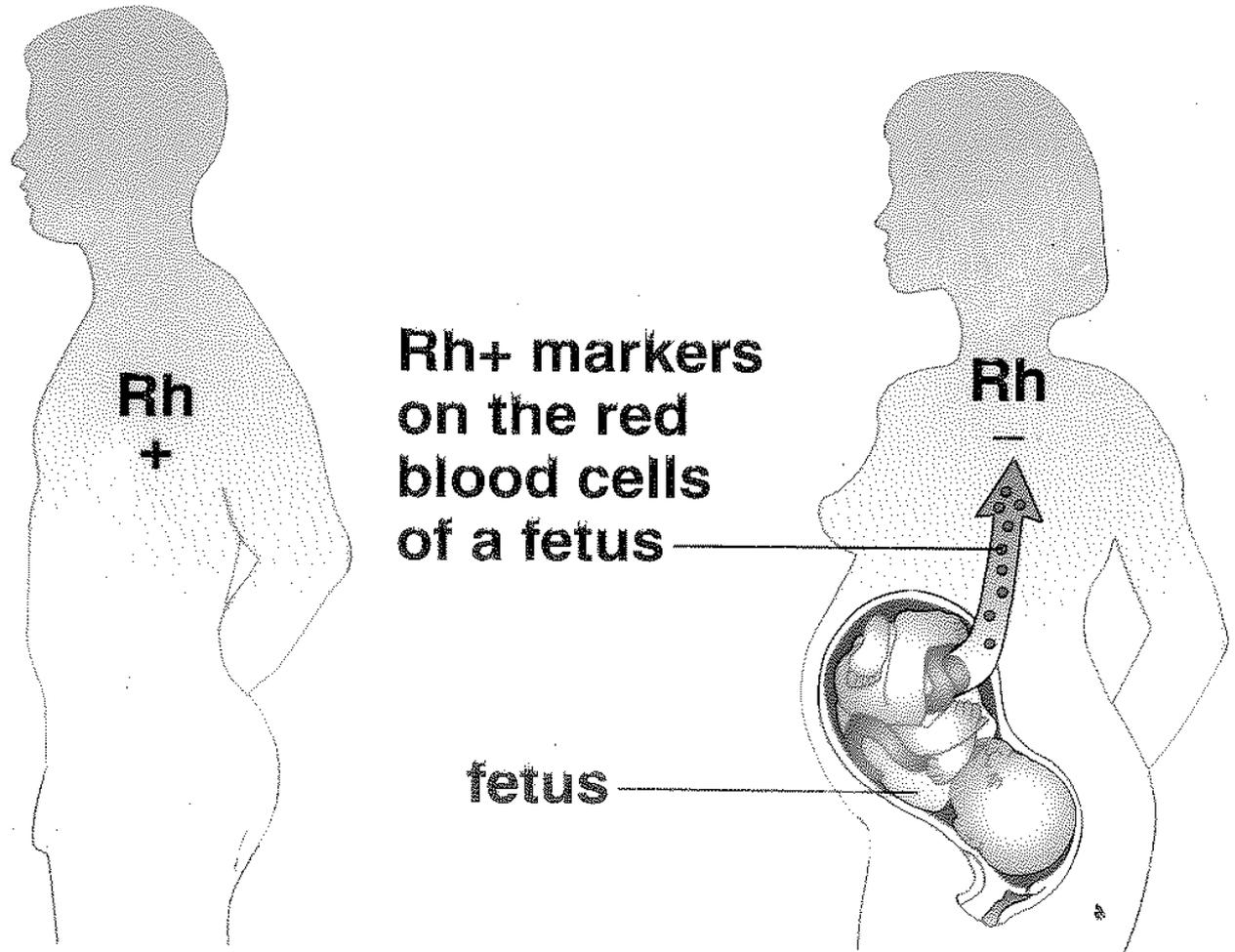


# Blood donation

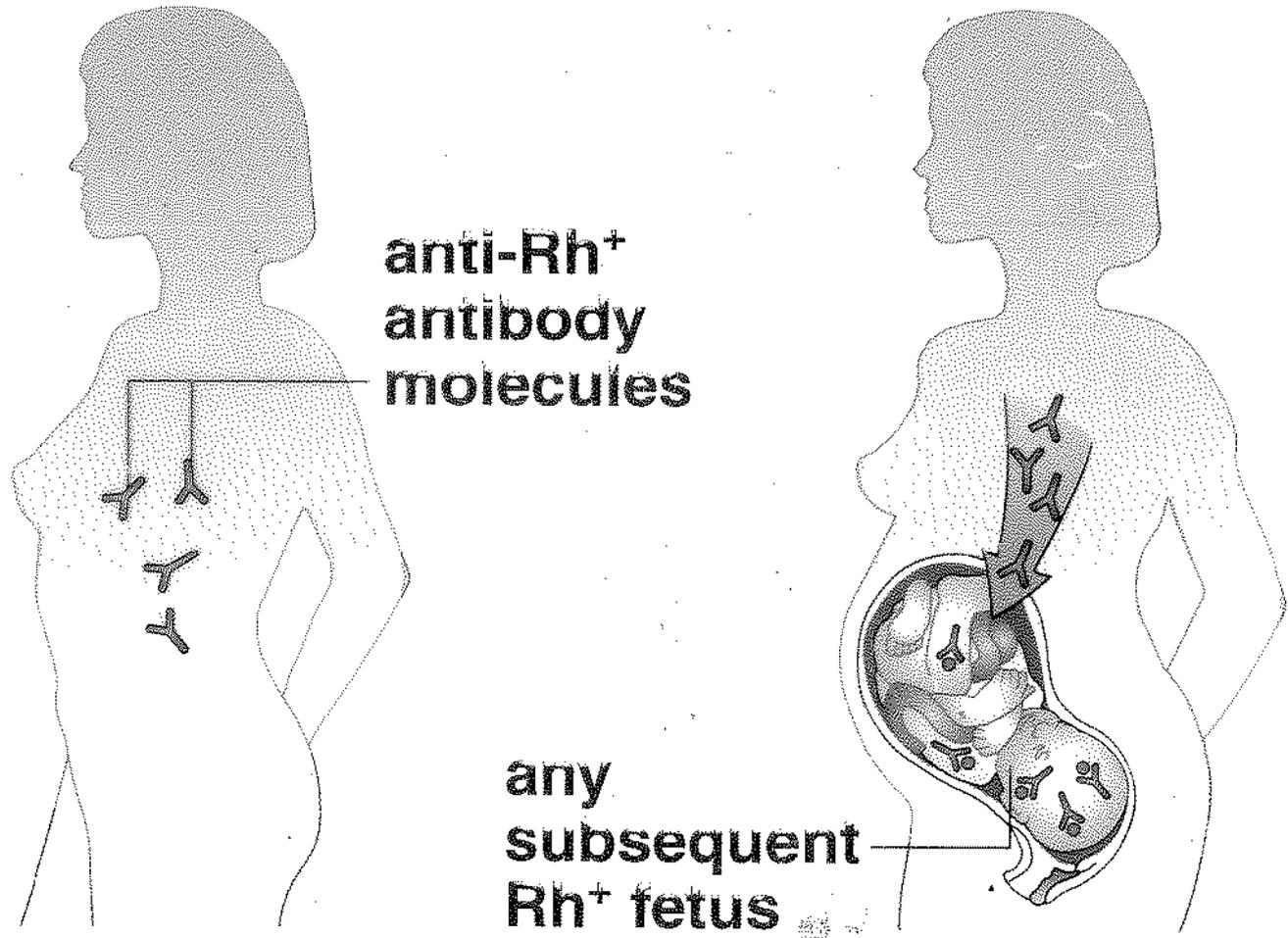
What types of blood can a person with **O** blood NOT receive ???

(a) Phenotype (blood group)	(b) Genotypes (see p.258)	(c) Antibodies present in blood serum	(d) Results from adding red blood cells from groups below to serum from groups at left			
			A	B	AB	O
<b>A</b>	$I^A I^A$ or $I^A i$	Anti-B				
<b>B</b>	$I^B I^B$ or $I^B i$	Anti-A				
<b>AB</b>	$I^A I^B$	—				
<b>O</b>	$ii$	Anti-A Anti-B				

# So what is the RH factor ??? = (+ or -)



Rh troubles happen for the 2<sup>nd</sup> baby if MOM is **RH -**



# Blood Type Punnett Square:

- List all the possible blood genotypes and phenotypes and their corresponding fractions for children conceived from a male heterozygous for type A blood with a woman with homozygous type B blood.

$$\text{MOM} = \frac{I^B I^B}{}$$

$$\text{DAD} = \frac{I^A i}{}$$

	$I^B$	
$I^A$	$I^A I^B$	
$i$	$I^B i$	

Genotypes	fractions	Phenotypes	fractions
$I^A I^B$	$1/2$	Type AB blood	$1/2$
$I^B i$	$1/2$	Type B blood	$1/2$

Some traits are determined by more than the simple interaction of dominant and recessive alleles

- Another example of **Codominance** happens with some horses and cows
- Example 2: Crossing black and white horses creates a **Roan** horse



**Black**

+



white

=



**Roan**



**Roan**

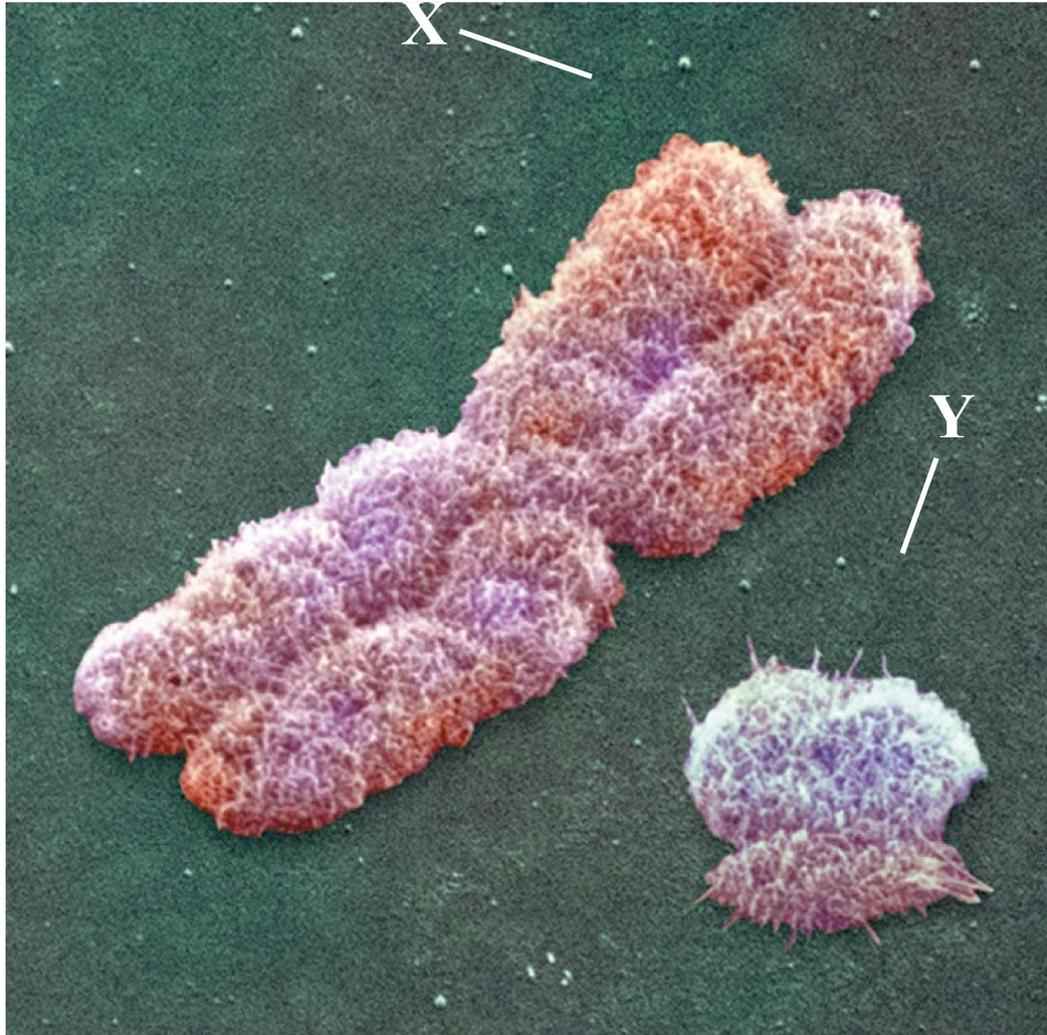


ROAN horses and cows are another example of **Codominance** ... 2 colors are each expressed at the same time in a “salt-n-pepper” effect

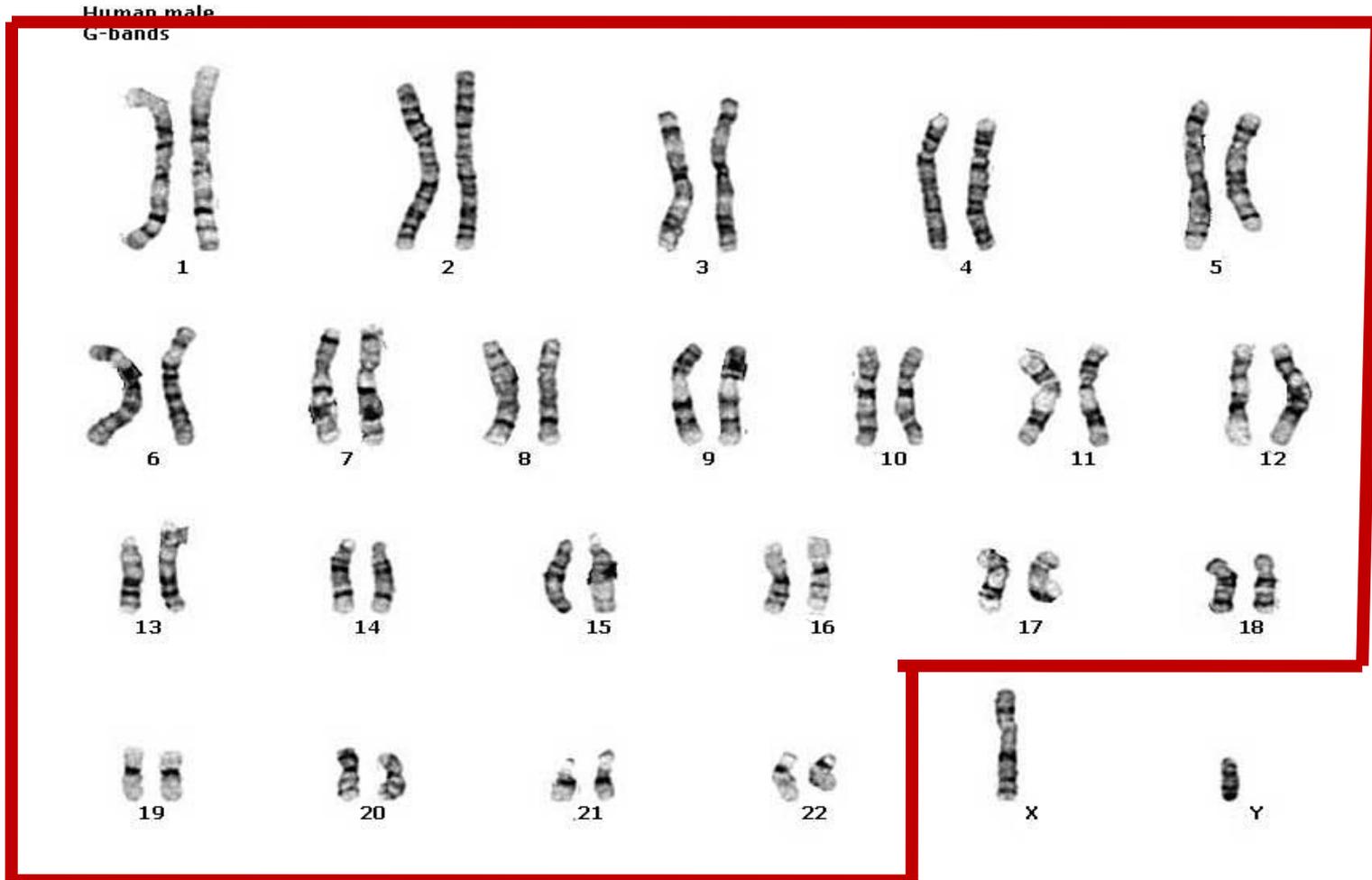


# Topic 6: Sex-Linked Traits

# Boy or Girl?

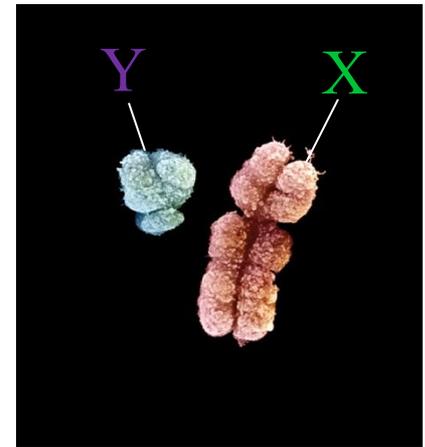
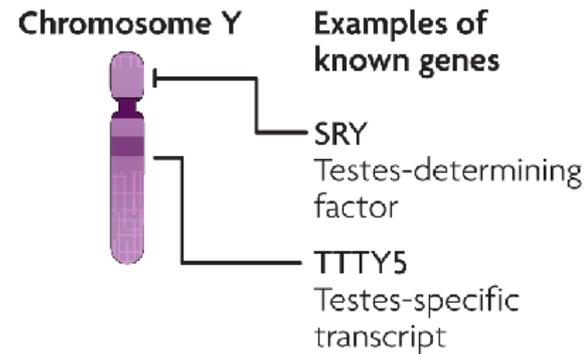
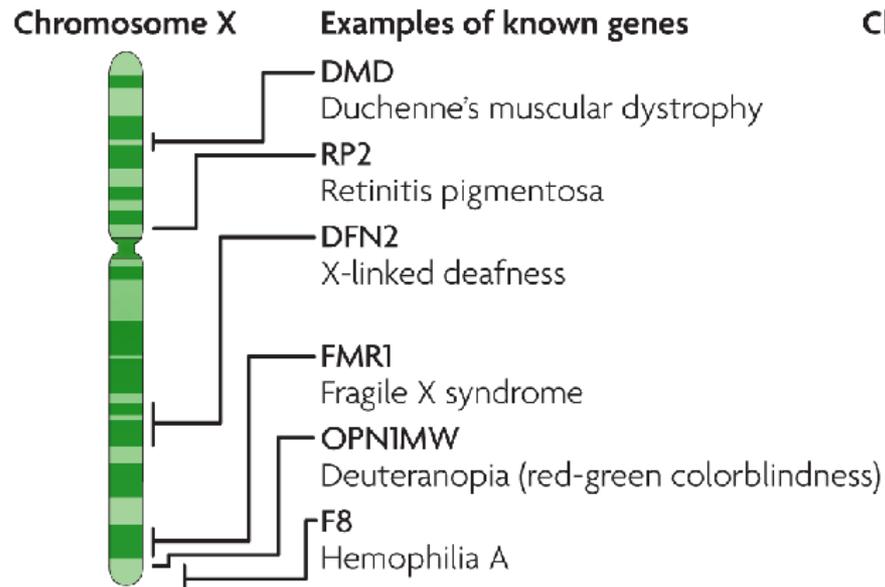


- Most traits for this boy are determined by genes that are located on his **autosomes** = (chromosomes 1-22)



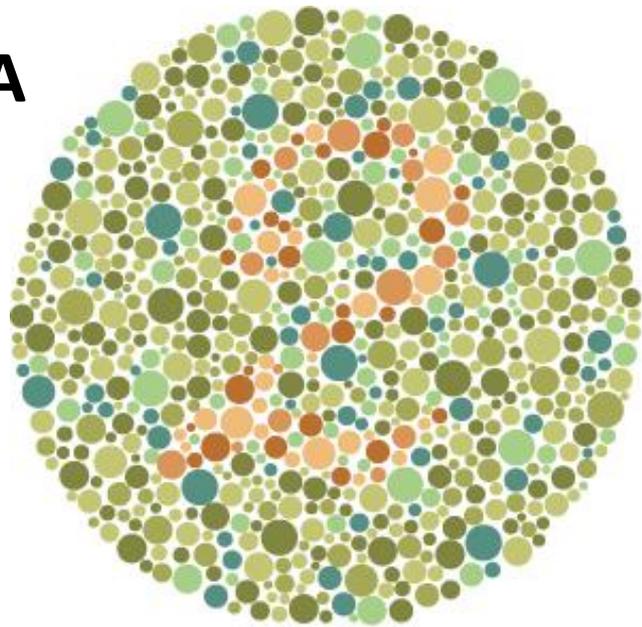
A few traits are **SEX-LINKED**. This means the gene for the trait is located on the **X** chromosome

- Compare the **X** and **Y** chromosomes below

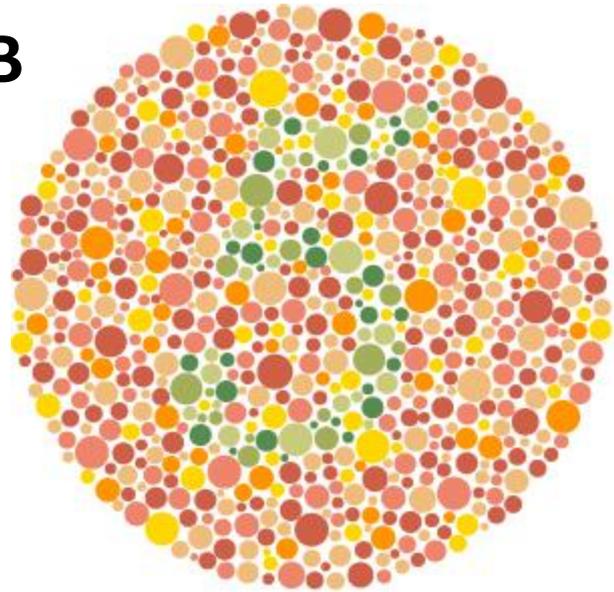


- If females are **XX** and males are **XY**, how do you think recessive disease mutations affect girls vs boys ???
- **Let's check the class for a common sex-linked mutation**

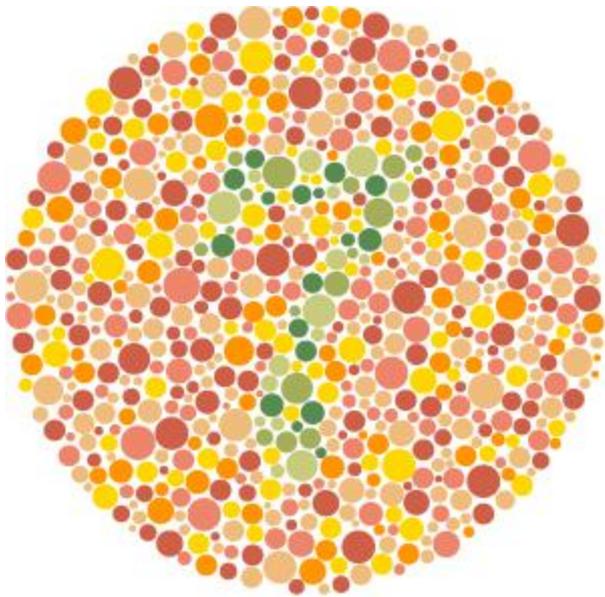
**A**



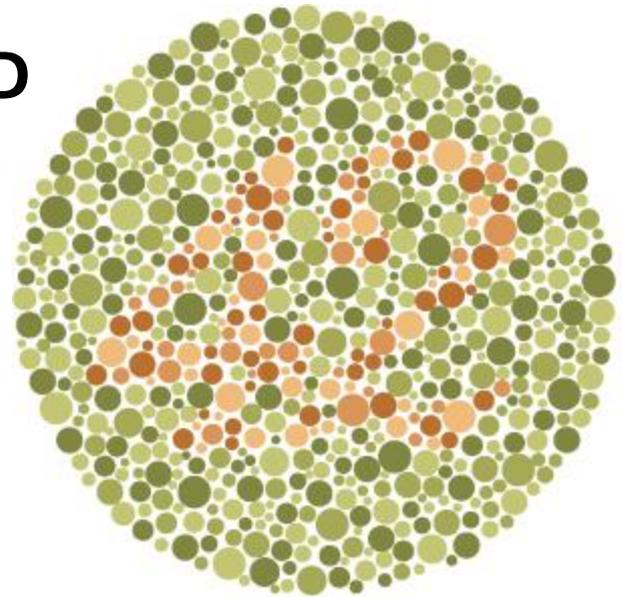
**B**



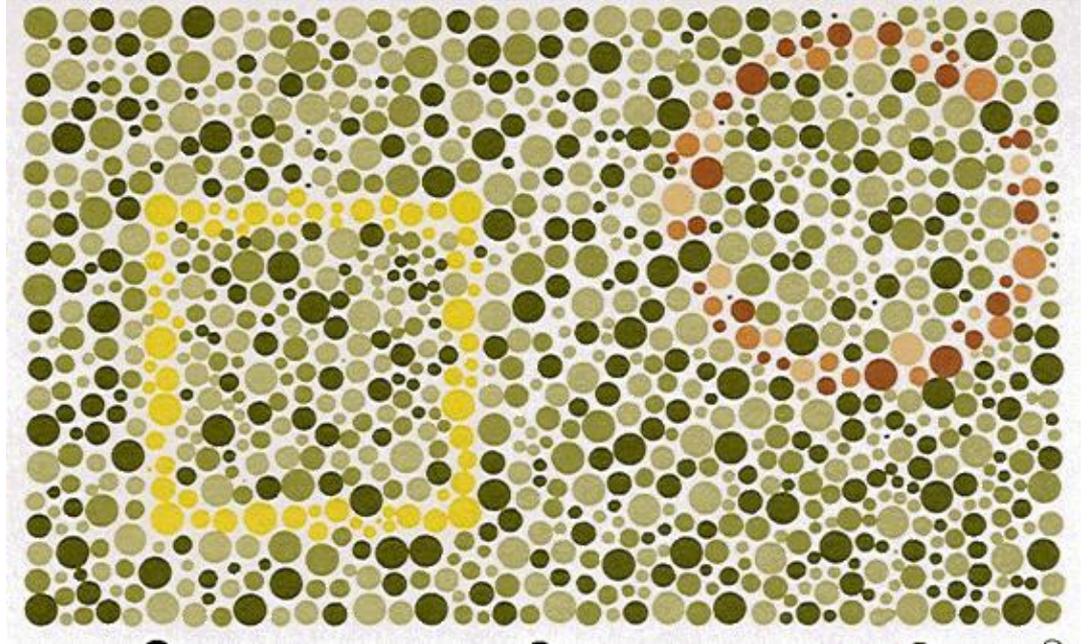
**C**



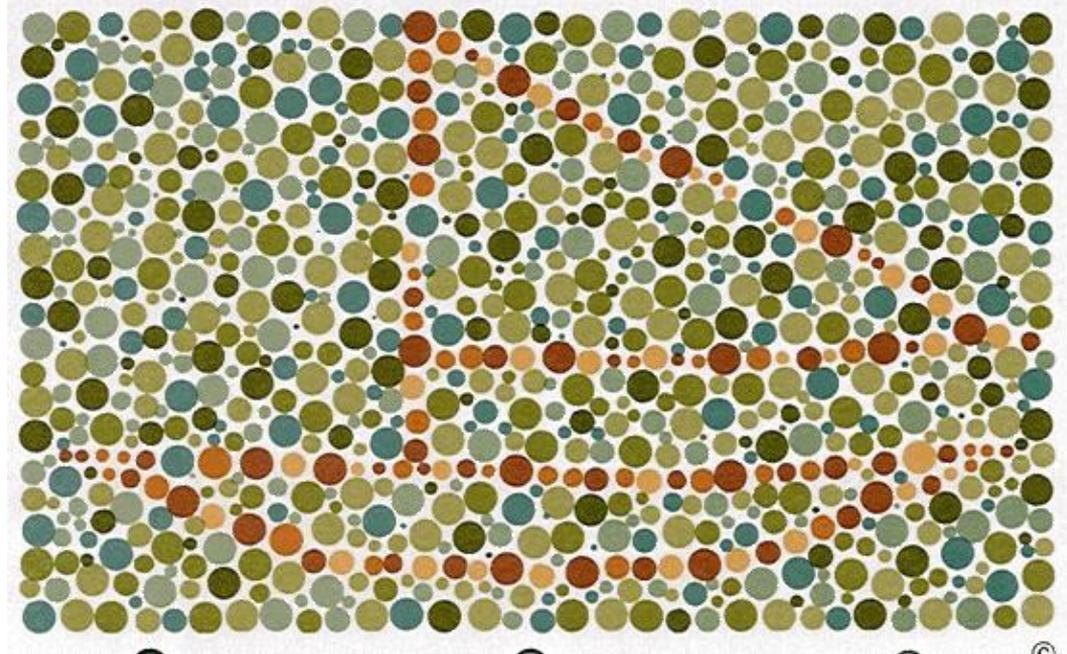
**D**



**E**



**F**

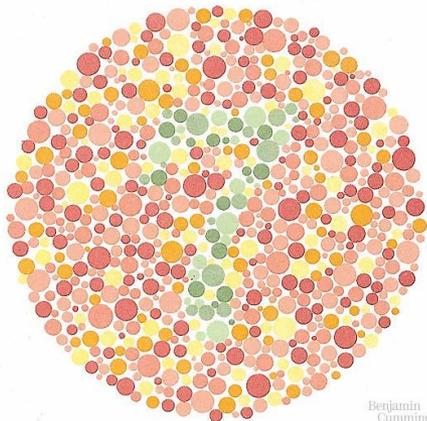


**Notice any difference?**



# Sex-linked genes affect males and females in different ways:

- **Color blindness** is a sex-linked trait caused by a recessive mutation (**b**= colorblind allele)
- Males → Always show the sex-linked trait if inherited (  $X^bY$  )
- Females → Rarely show CB, only when **homozygous** (  $X^b X^b$  )
  - Most females are healthy but could be:
    - **Heterozygous** carrier (  $X^B X^b$  )
    - **Homozygous** (  $X^B X^B$  )





# Duchenne Muscular Dystrophy is a Sex-Linked Disease



- *Extraordinary Measures* is a 2010 movie about a family's quest to find a cure for their kids who have a form of MD called Pompe Disease

# Let's try a sex-linked Punnett Square

Jim is not colorblind and neither is his wife, Mary. However, because Mary's dad WAS colorblind, she is heterozygous for red-green colorblindness.

	Genotypes
Jim	$X^B Y$
Mary	$X^B X^b$

	$X^B$	$X^b$
$X^B$	$X^B X^B$	$X^B X^b$
Y	$X^B Y$	$X^b Y$

Will any children be colorblind?

# Topic 7: Epistasis

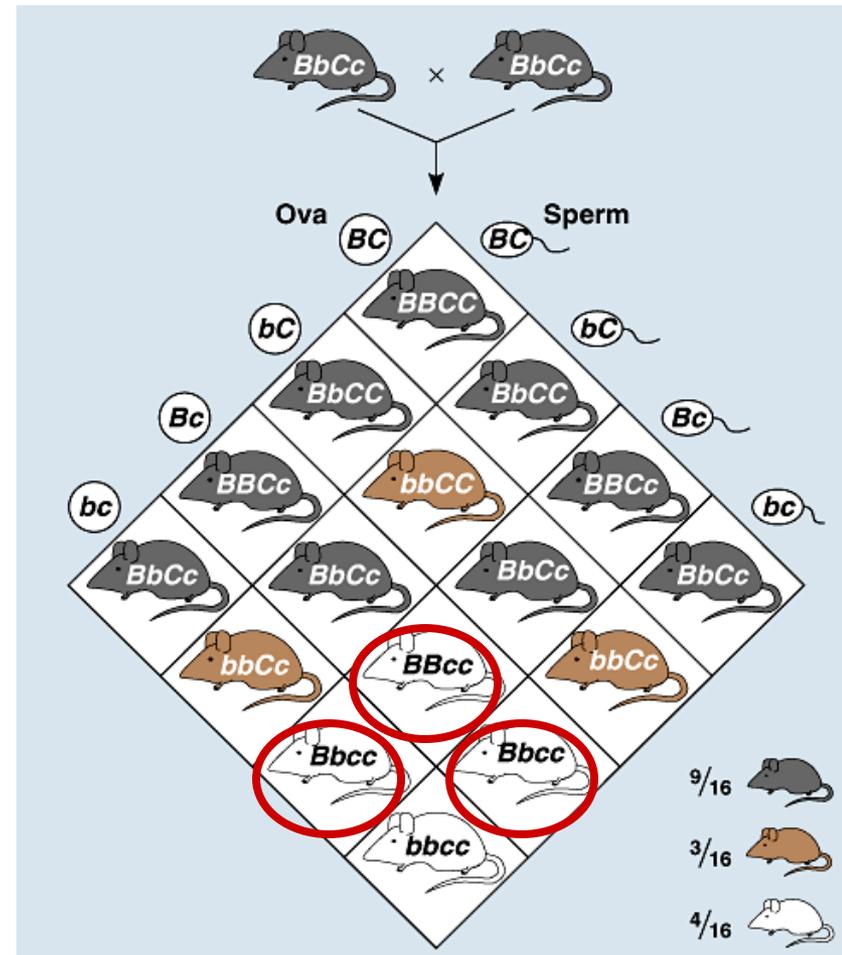
# Epistasis

One gene masks another (i.e., one gene can block the expression of genetic information at another gene)

• Examples = coat color in mice and Labrador dogs

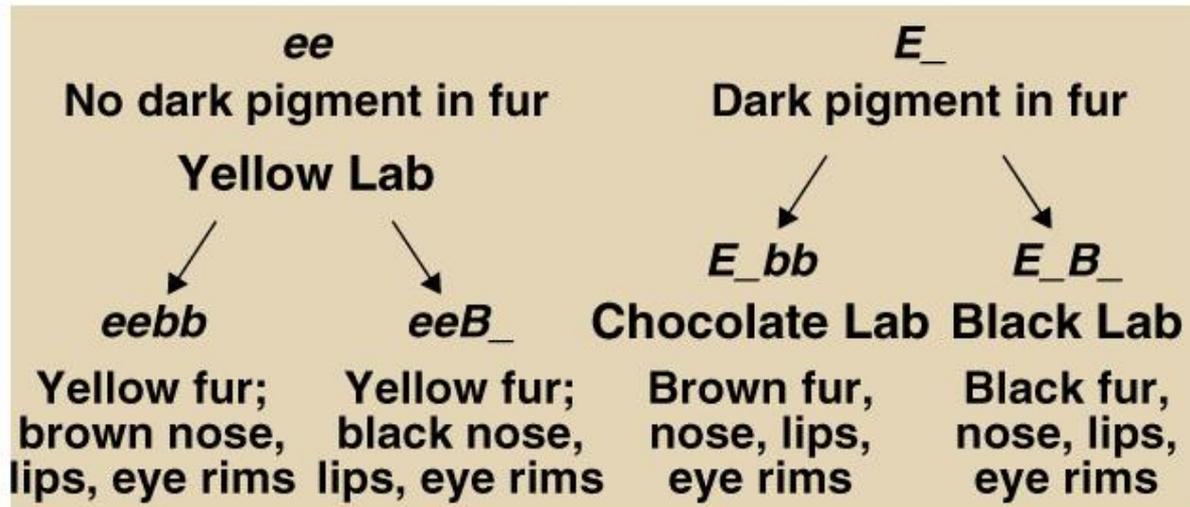
• coat color in mice = 2 genes

- Pigment yes (C) or no pigment (c)
- more pigment (black=B) or less (brown=b)
- cc = albino, no matter B allele
- 9:3:3:1 becomes 9:3:4



# Epistasis in Labrador retrievers

- 2 genes: **E** & **B**
  - pigment (**E**) or no pigment (**e**)
  - how dark pigment will be: black (**B**) to brown (**b**)



# Determine the litter color ratio for the following cross: Black Lab (BbCc) x Chocolate Lab (bbCc)

Mom's genotype: BbCc (Black fur)

possible egg varieties: BC Bc bC bc

Dad's genotype: bbCc (Chocolate fur)

possible sperm varieties: bC ~~bC~~ bc ~~bc~~

	BC	Bc	bC	bc
bC	 BbCC	 BbCc	 bbCC	 bbCc
bc	 BbCc	 Bbcc	 bbCc	 bbcc

Phenotype Probabilities:

-  Black fur, Running:  $\frac{3}{8}$
-  Choc fur, waltzing:  $\frac{3}{8}$
-  Yellow fur, Running:  $\frac{2}{8}$

B = black fur b = chocolate fur C = YES pigment c = NO pigment

# Topic 8: Polygenic Traits

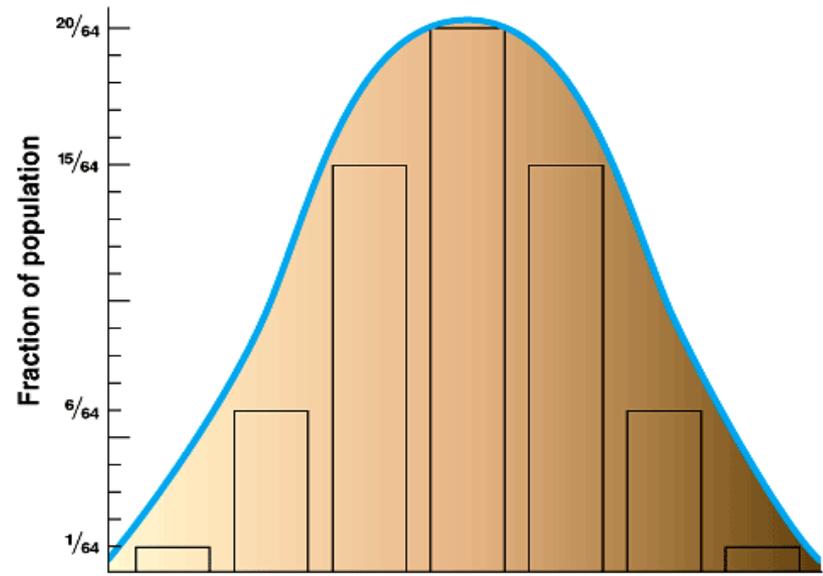
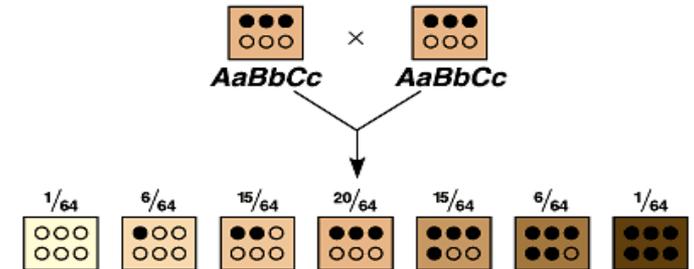
# Polygenic inheritance

- The alleles from “many genes” combine to determine the phenotype for one trait

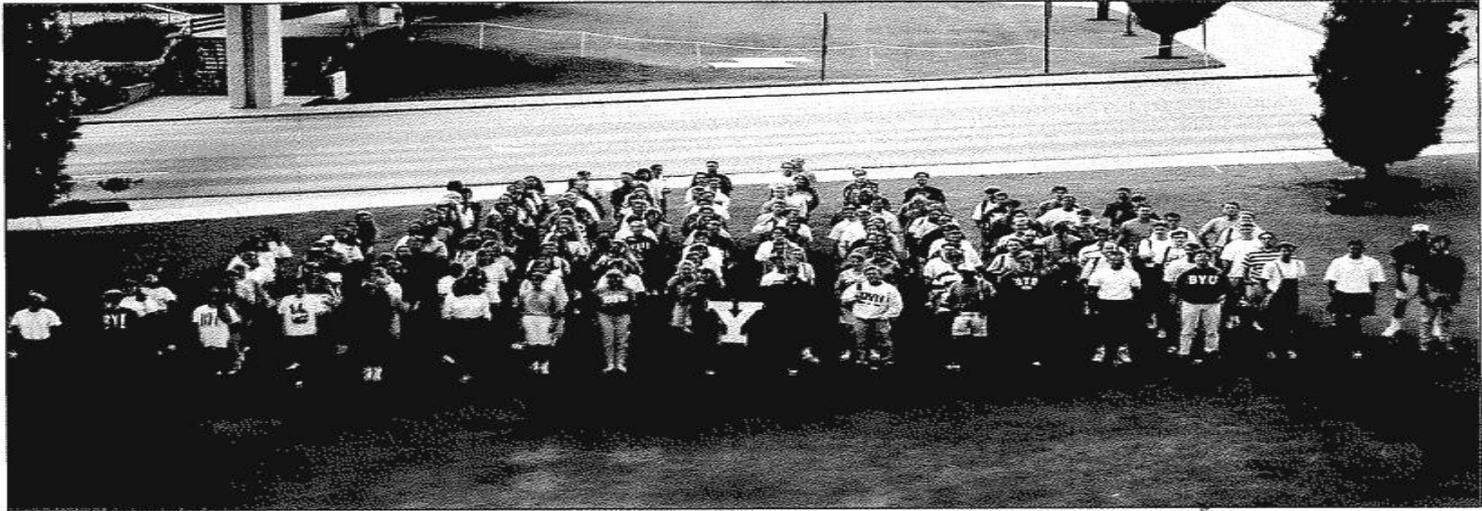
– phenotypes range from extreme to extreme along a “bell-shaped” continuum

– human trait examples:

- skin color
- height
- weight
- eye color
- intelligence
- behaviors

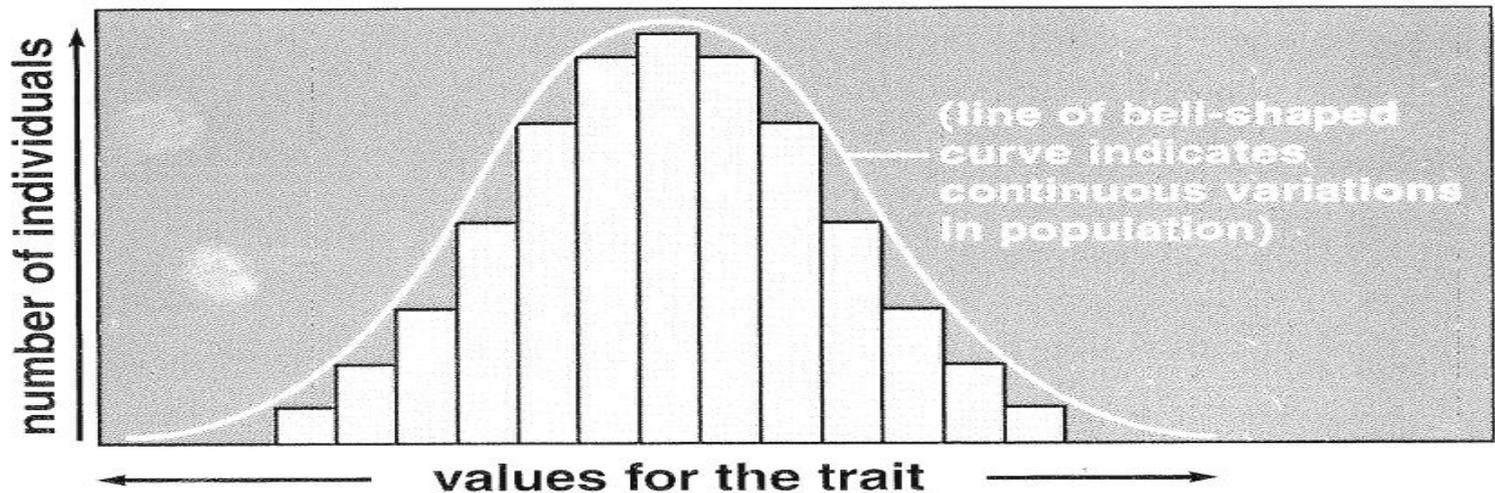


# Human height is a polygenic Trait



a

————— Height —————→



Many genes may interact to produce one trait.

- Polygenic traits are produced by two or more genes.

Order of dominance:  
brown > green > blue.



GENE NAME	DOMINANT ALLELE	RECESSIVE ALLELE
BEY1	brown	blue
BEY2	brown	blue
GEY	green	blue

# Topic 9: Wrap Up

What factor determines if the fox is white or brown?

**Coat color in arctic fox is influenced by heat sensitive alleles**

What factor determines if the flower is purple or magenta?

**Color of Hydrangea flowers is influenced by soil pH**



(a)



(b)



# What factor determines the color of human skin tone?

**Human skin color is influenced by both genetics & environmental conditions**



## **Conclusion:**

- Phenotype is controlled by both
- **genes** (Nature) and **environment** (Nurture)

# Sometimes the environment directs how a gene is used

- Phenotype is a combination of genotype and environment.
- The sex of sea turtles depends on both genes and the nest temperature
- Height is an example of a phenotype strongly affected by the environment.



## 9.15 Connection: Genetic testing can detect disease-causing alleles

- Genetic testing can be of value to those at risk of developing a genetic disorder or of passing it on to offspring
- Pros vs Cons of gene testing?



Figure 9.15B

Figure 9.15A

# 9.10 Connection: Fetal testing can spot many inherited disorders early in pregnancy

- Karyotyping and gene tests of fetal cells can help people make reproductive decisions
  - Fetal cells can be obtained through \_\_\_\_\_  
**amniocentesis**

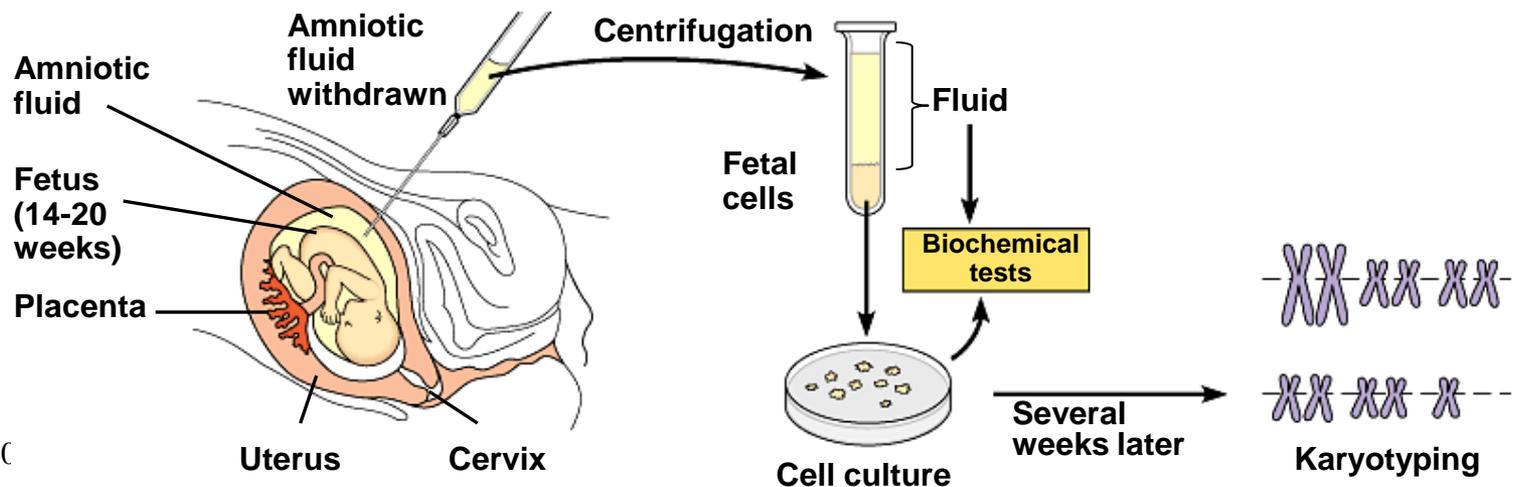


Figure 9.1C

- **Chorionic villus sampling** is another procedure that obtains fetal cells for karyotyping

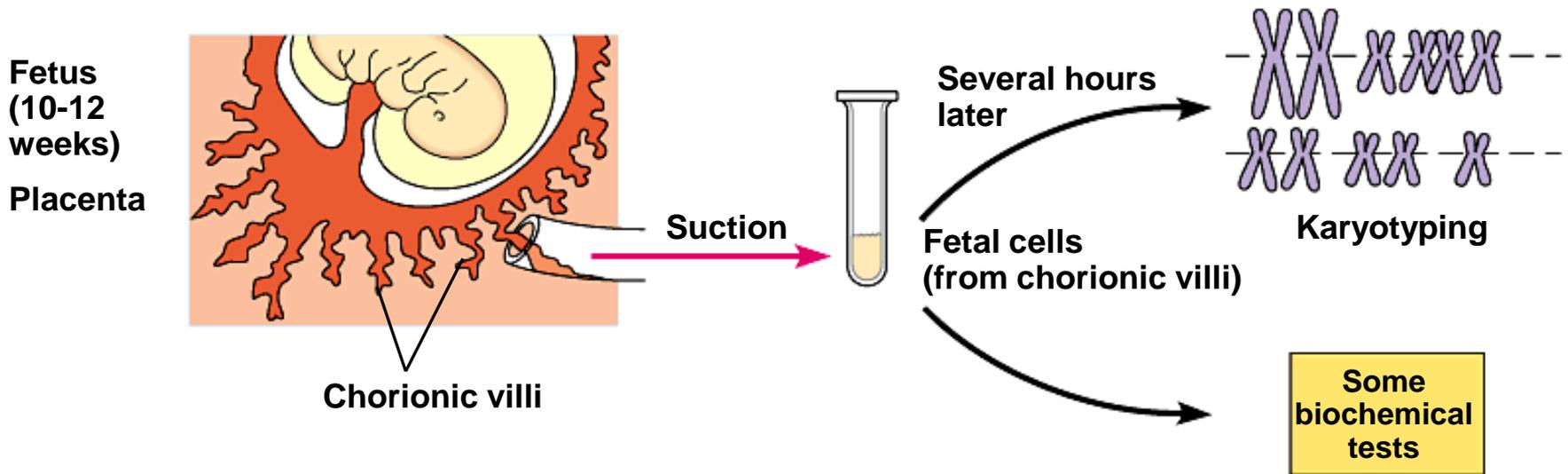


Figure 9.10B

- Examination of the fetus with ultrasound is another helpful technique

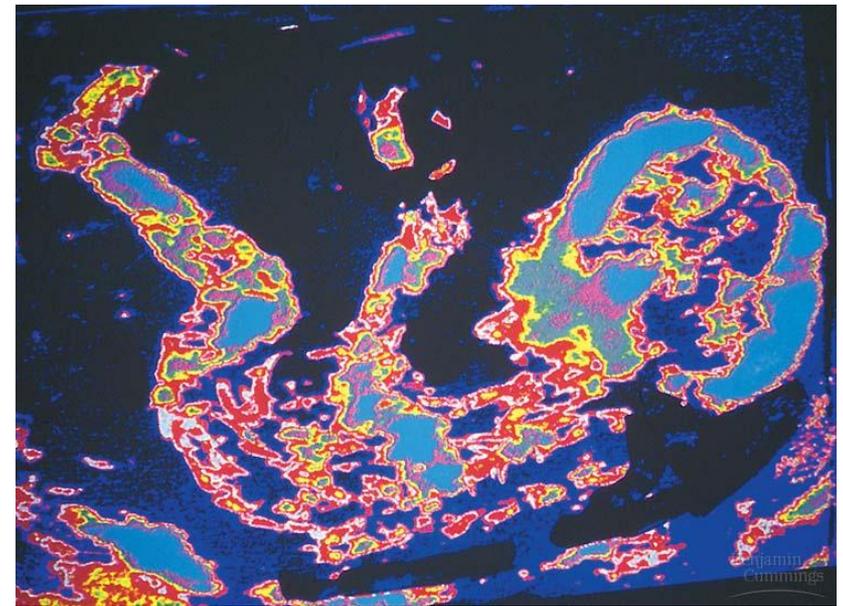
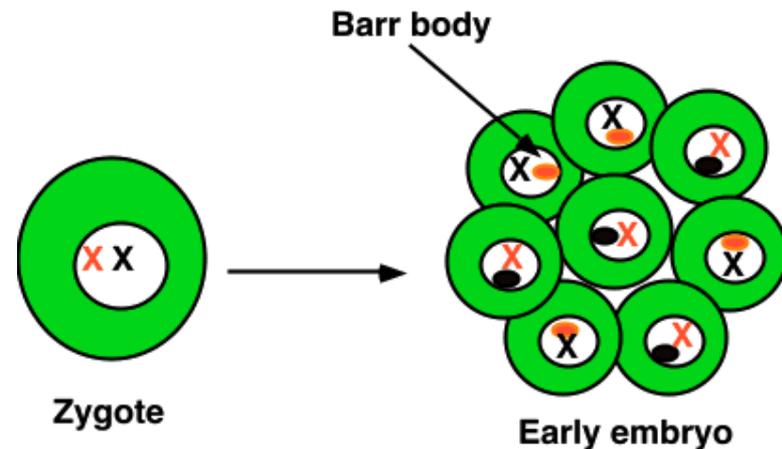
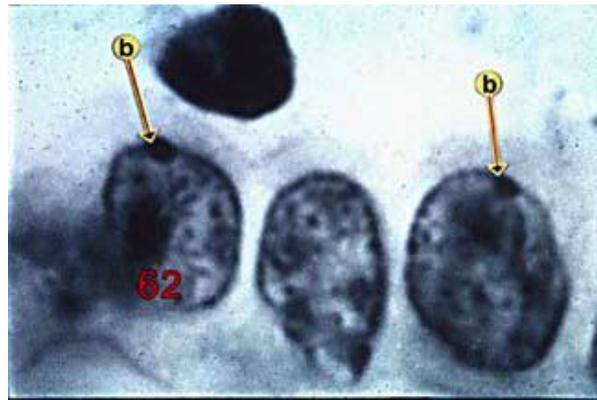


Figure 9.10C, D

# X-inactivation

- Female mammals inherit two X chromosomes
  - one X becomes inactivated during embryonic development
    - condenses into compact object = Barr body

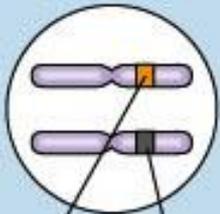


# X-inactivation & tortoise shell cat

- 2 different cell lines in cat

**Early embryo**

X chromosomes



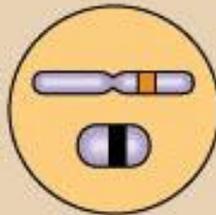
Allele for orange fur

Allele for black fur

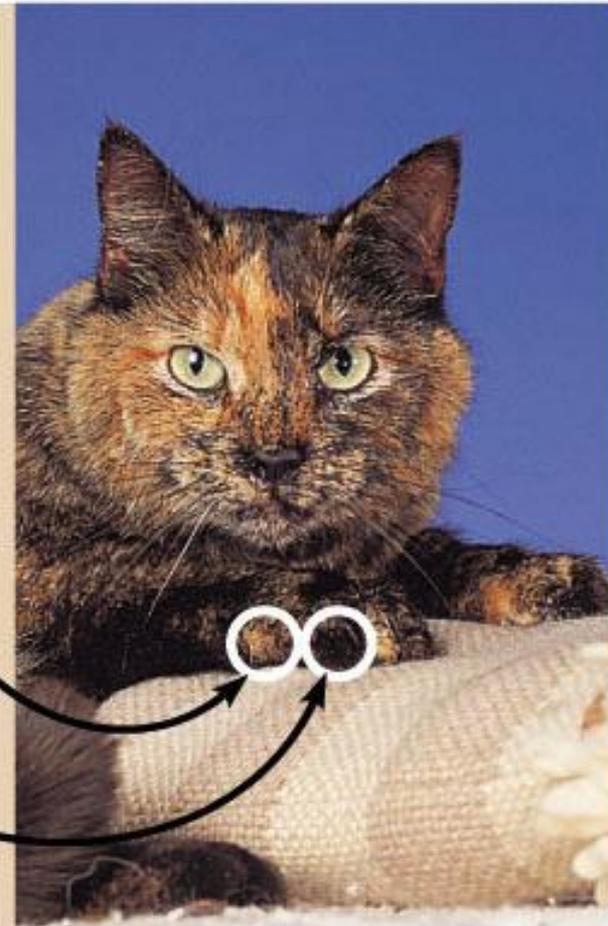
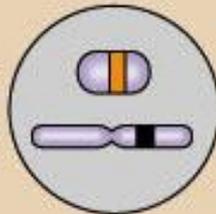
Cell division  
and  
X chromosome  
inactivation

**Two cell populations in adult**

Active X Orange fur  
Inactive X



Inactive X  
Active X Black fur



# Pleiotropy

- Most genes are pleiotropic
  - one gene affects more than one phenotypic characteristic
    - wide-ranging effects due to a single gene:
    - dwarfism (achondroplasia)
    - gigantism (acromegaly)



# Acromegaly: André the Giant

