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# Genes and Inheritance

(11-12)

# You are a unique combination of your two parents

- We all have two copies of each gene (one maternal and one paternal)
- Gametes produced via **meiosis** contain only one copy of each gene
- Fusion of gametes during **fertilization** produces offspring with two copies of each gene

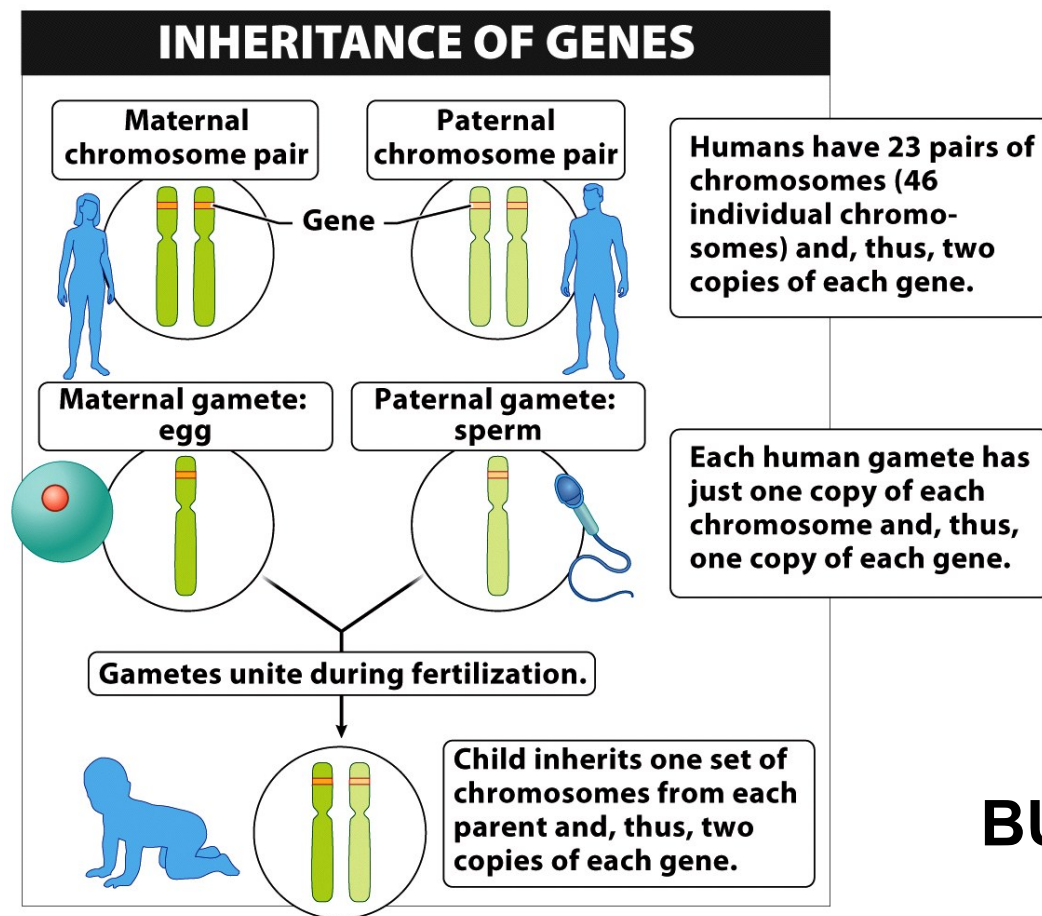
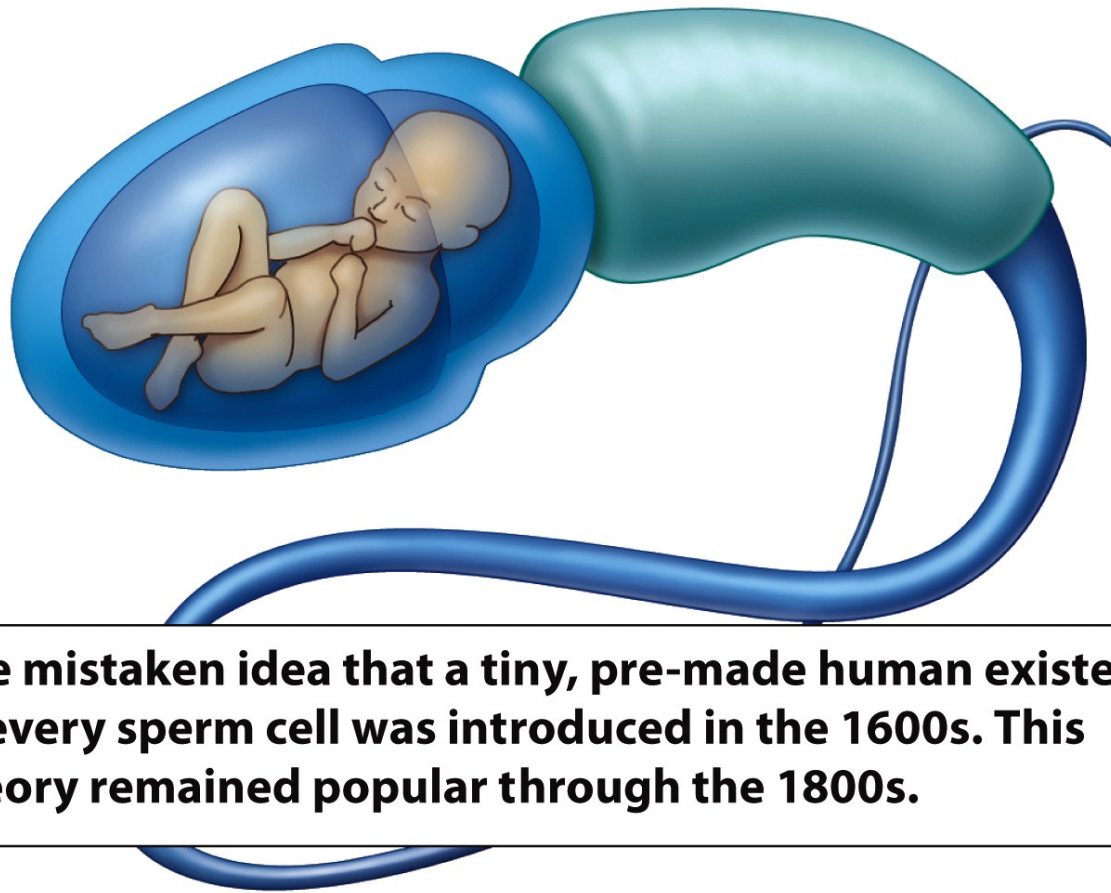


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**BUT WE DIDN'T ALWAYS  
KNOW THIS!!**

People used to think that a tiny baby was transferred via sperm or that traits from both parents were blended through exchange of blood



**The mistaken idea that a tiny, pre-made human existed in every sperm cell was introduced in the 1600s. This theory remained popular through the 1800s.**



# Gregor Mendel (1822-1884)

“Father of Genetics”

(first to understand and define the rules of inheritance)

## MENDEL'S RESEARCH APPROACH



Three features of Mendel's methodical research were critical to its success.

### PEA PLANTS ARE AN IDEAL STUDY ORGANISM

- They are easy to maintain.
- They are easy to breed.
- They reproduce quickly; multiple generations can be observed.



### PEA PLANTS HAVE NUMEROUS EASILY CATEGORIZED TRAITS WITH TWO VARIANTS EACH

TRAIT	VARIANTS	
Flower color:	 Purple	 White
Pea shape:	 Round	 Wrinkled
Pea color:	 Green	 Yellow

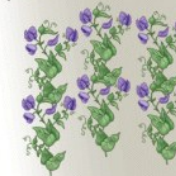
OTHER TRAITS INCLUDE: Pod color, pod shape, flower position, and plant height.

### DISTINCT POPULATIONS WERE ESTABLISHED

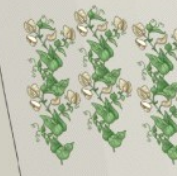
Mendel used true-breeding plants—they always produced offspring with the same variant of the trait as the parents.



True-breeding purple-flower plants



True-breeding white-flower plants



Gregor Mendel (1822–1884)

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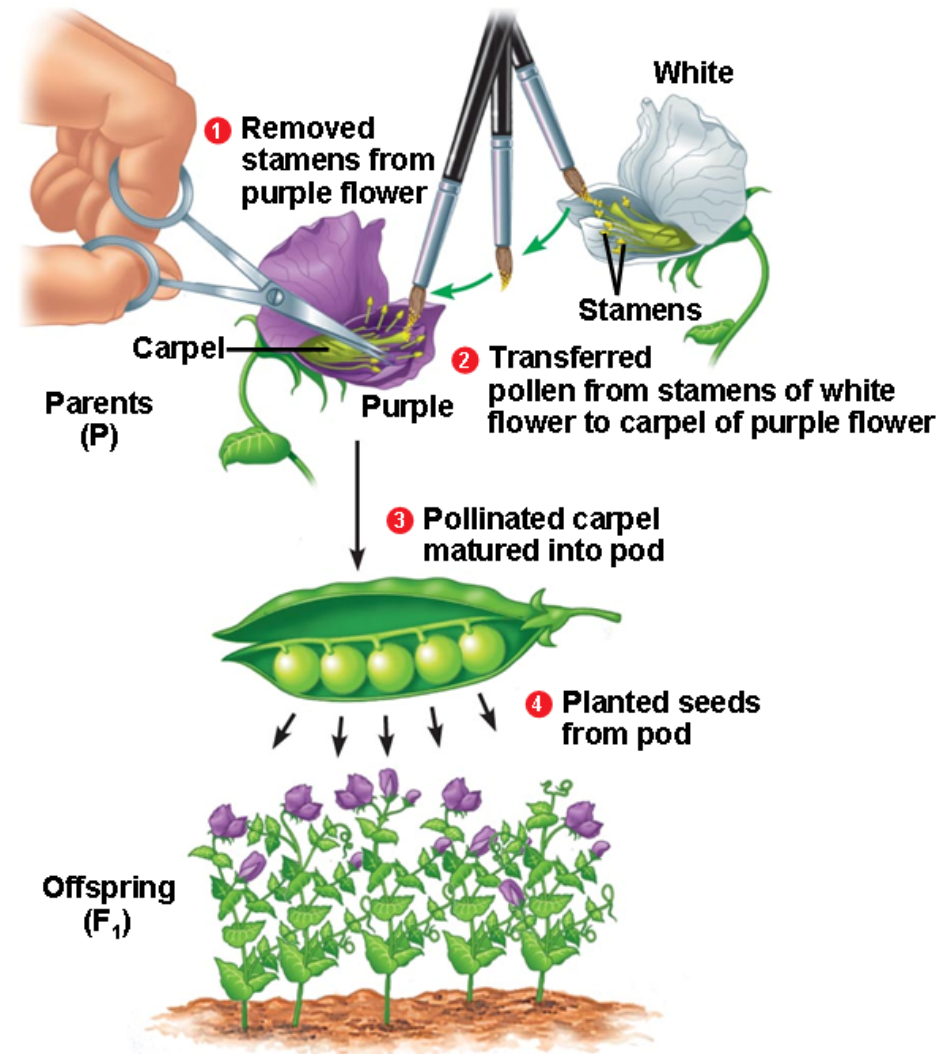
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# Mendel could control mating and produce lots of offspring















Self-fertilization = fertilization of egg and sperm within the same plant

Cross-fertilization = fertilization of one plant by pollen from a different plant



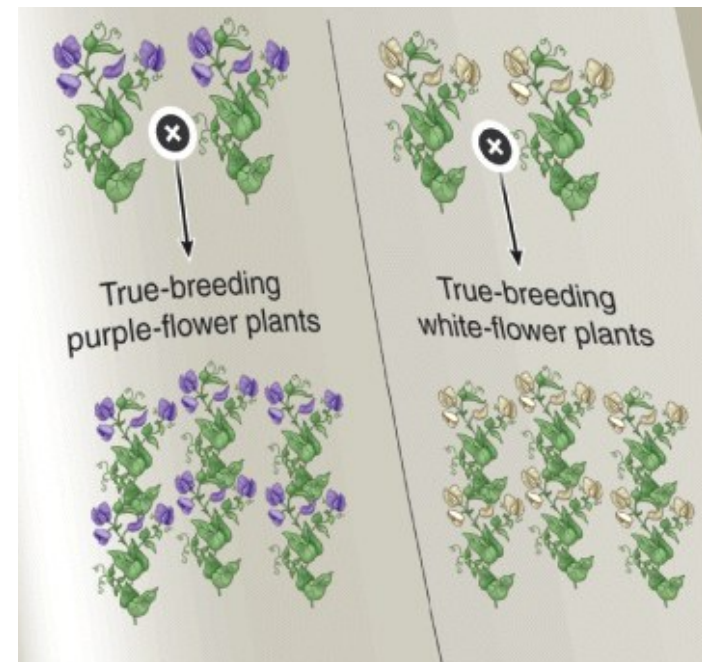
**TRAITS (GENES)**

**VARIANTS (ALLELES)**

	(Dominant)	(Recessive)
Flower color	 <b>Purple</b>	 <b>White</b>
Flower position	 <b>Axial</b>	 <b>Terminal</b>
Seed color	 <b>Yellow</b>	 <b>Green</b>
Seed shape	 <b>Round</b>	 <b>Wrinkled</b>
Pod shape	 <b>Inflated</b>	 <b>Constricted</b>
Pod color	 <b>Green</b>	 <b>Yellow</b>
Stem length	 <b>Tall</b>	 <b>Dwarf</b>

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**True Breeding =**  
varieties for which  
self-fertilization  
produces offspring  
all identical to the  
parent



**P generation  
(true-breeding  
parents)**

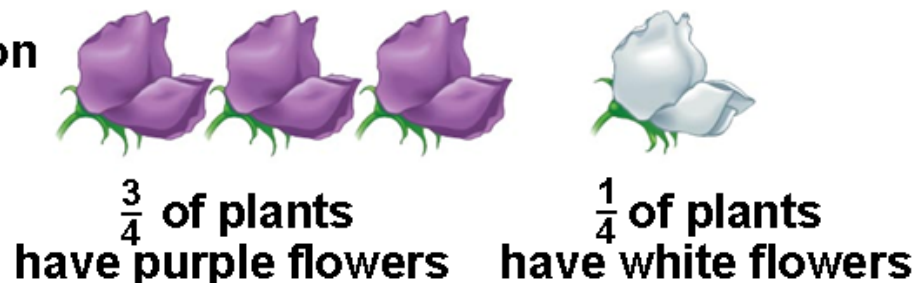


**F<sub>1</sub> generation**



**Fertilization  
among F<sub>1</sub> plants  
(F<sub>1</sub> × F<sub>1</sub>)**

**F<sub>2</sub> generation**



Mendel started  
crossing two  
different  
true-breeding  
plants for one  
trait

**Which flower color  
allele is  
dominant?**



# Mendel's Three Hypotheses of Inheritance

1) Gametes carry only one copy of each gene because homologous chromosomes separate (segregate) during meiosis = **Law of Segregation**

2) An organism inherits one gene from each parent (**Genotype**)

- Homozygous vs. Heterozygous

3) Inherited genes determine the outer appearance of the organism (**Phenotype**)

- Dominant vs. Recessive

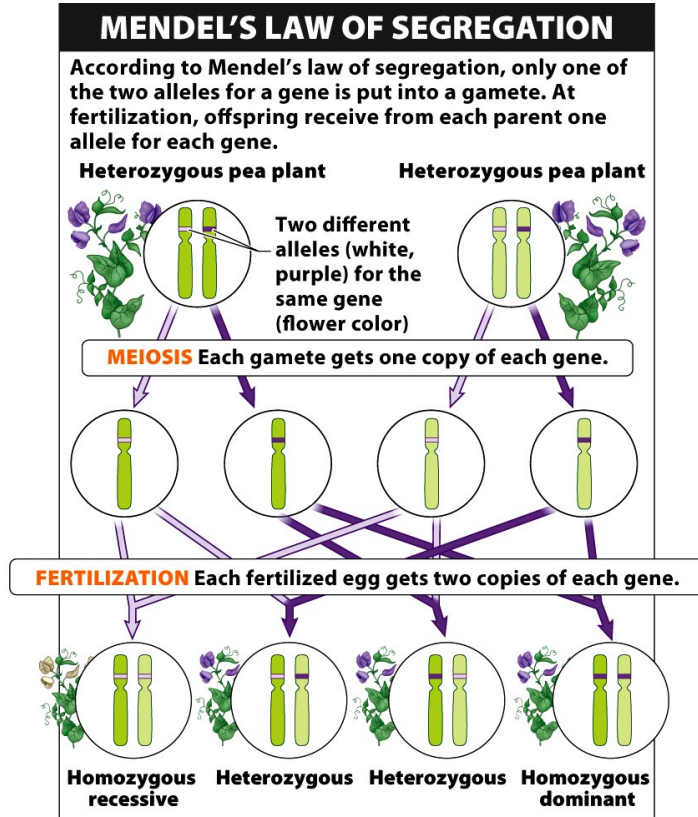
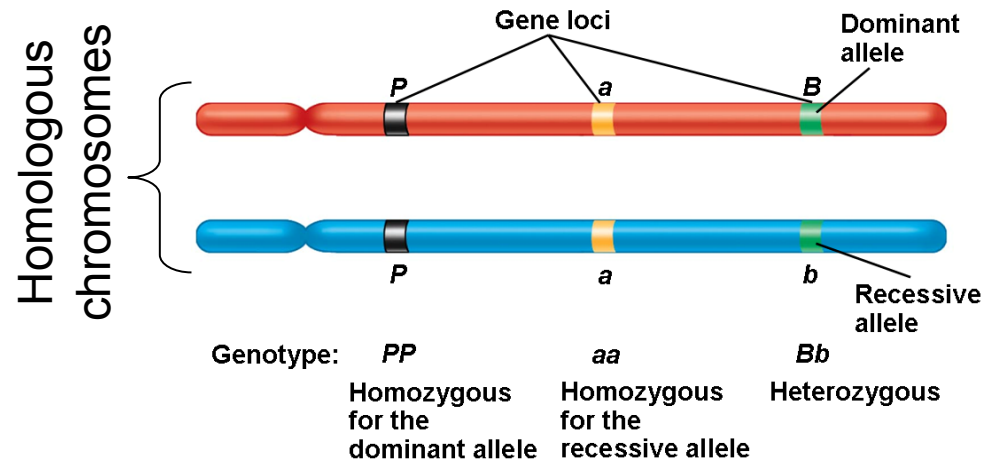
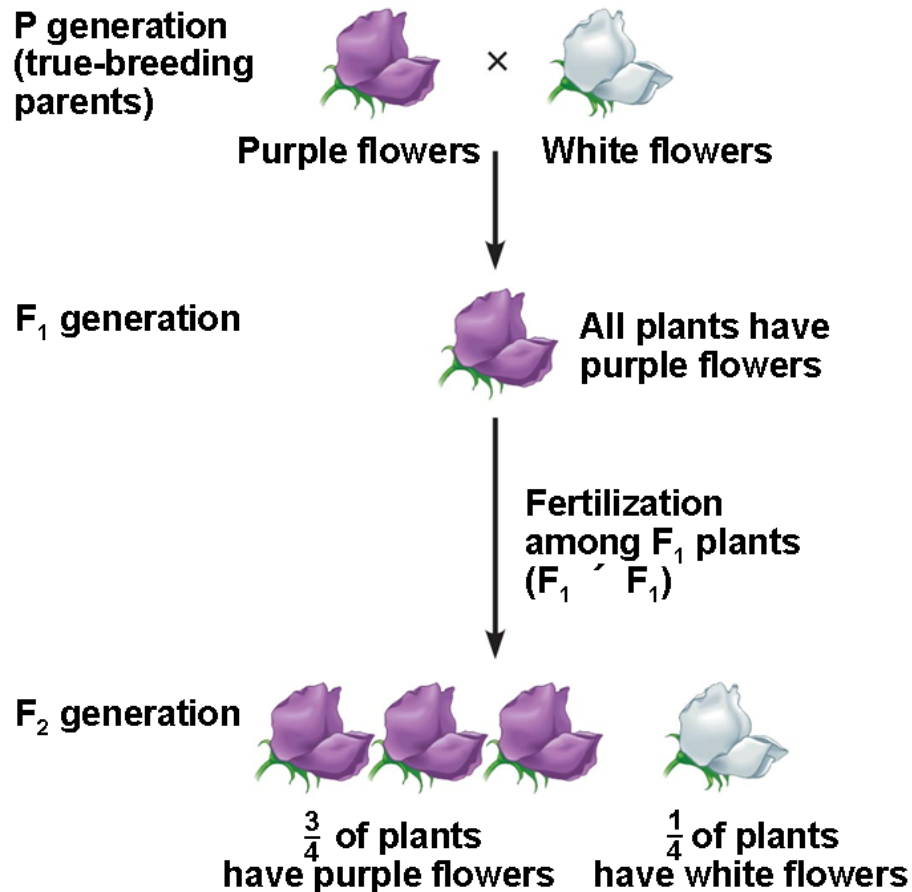


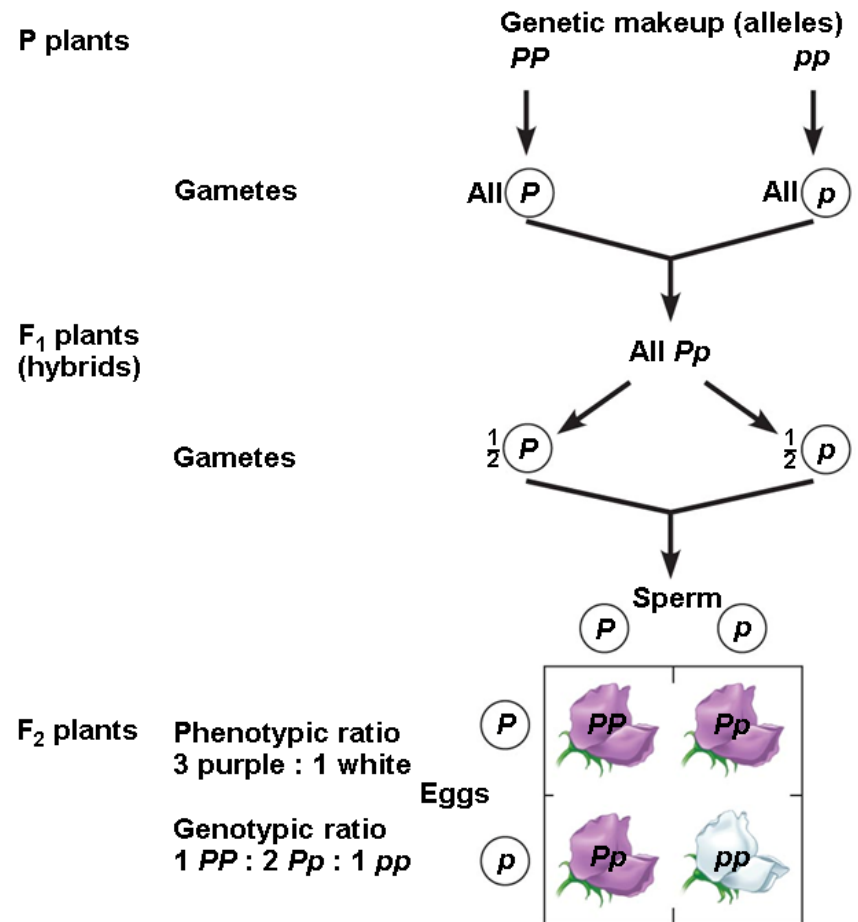
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# Were Mendel's hypotheses correct?



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**PUNNETT SQUARE** = used to predict inheritance of future generation

# New alleles arise through mutation



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Genotypic ratio?

Phenotypic ratio?

The dominant curled ears allele was caused by a mutation.  
Cross a heterozygous and homozygous recessive cat and give the expected genotypic and phenotypic ratios of the offspring.




# Genetics involves **probability** and **chance**

Albinism is caused by a recessive allele for pigmentation.

What are the chances that an **albino female** and a **heterozygous male** will have an albino offspring?



**PHENOTYPE:** Little or no pigment in the eyes, hair, and skin  
**GENOTYPE:** Homozygous for the recessive allele for albinism

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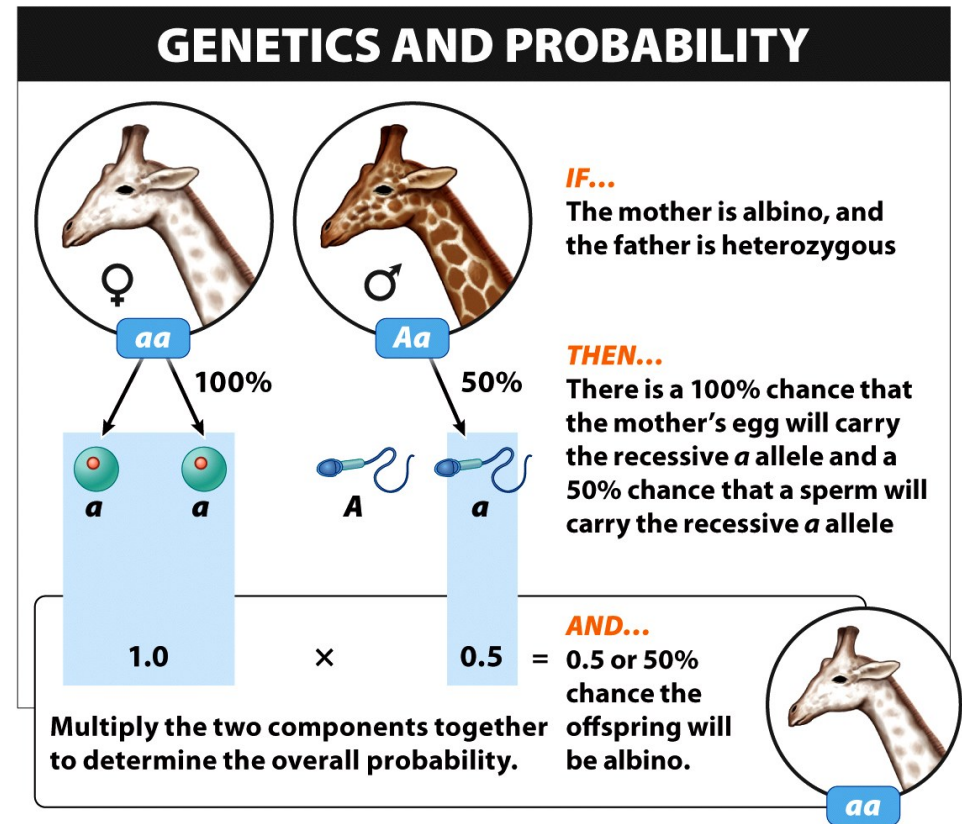


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## MENDEL'S LAW OF INDEPENDENT ASSORTMENT

Mendel's law of independent assortment states that one trait does not influence the inheritance of another trait.

**IF...**

Parents are both homozygous for both traits. (But each parent is homozygous for a different allele than the other parent.)

**MOTHER**  
albino  
homozygous  $aa$   
dimpled chin  
homozygous  $DD$

**GAMETES**

All are  $aD$

**FATHER**  
pigmented  
homozygous  $AA$   
non-dimpled chin  
homozygous  $dd$



**GAMETES**

All are  $Ad$

All are  $Aa Dd$

**THEN...**

All offspring are heterozygous for both traits (i.e., "doubly heterozygous"):  $Aa Dd$  (and exhibit the dominant trait in their phenotype).

OFFSPRING	GENOTYPE	PHENOTYPE
	All heterozygous $Aa$	All pigmented
	All heterozygous $Dd$	All dimpled chin



*In this example, having a dimpled chin does not affect which alleles are inherited for skin pigmentation.*

**IF...**

Parents are both heterozygous for both traits (i.e., "doubly heterozygous"). Four different types of gametes are produced by each:  
 $AD$ ,  $Ad$ ,  $aD$ , and  $ad$ .

**MOTHER**  
pigmented  
heterozygous  $Aa$   
dimpled chin  
heterozygous  $Dd$

**GAMETES**

$AD$   $Ad$   $aD$   $ad$

**FATHER**  
pigmented  
heterozygous  $Aa$   
dimpled chin  
heterozygous  $Dd$

**GAMETES**

$AD$   $Ad$   $aD$   $ad$

	$AD$	$Ad$	$aD$	$ad$
$AD$	 $AA DD$	 $AA Dd$	 $Aa DD$	 $Aa Dd$
$Ad$	 $AA Dd$	 $AA dd$	 $Aa Dd$	 $Aa dd$
$aD$	 $Aa DD$	 $Aa Dd$	 $aa DD$	 $aa Dd$
$ad$	 $Aa Dd$	 $Aa dd$	 $aa Dd$	 $aa dd$

**THEN...**

The genotype proportions for  $A/a$  are still  $\frac{1}{4} AA$ ,  $\frac{1}{2} Aa$ , and  $\frac{1}{4} aa$ . And the genotype proportions for  $D/d$  are still  $\frac{1}{4} DD$ ,  $\frac{1}{2} Dd$ , and  $\frac{1}{4} dd$ .







OFFSPRING	GENOTYPE	PHENOTYPE
	$\frac{1}{4}$ homozygous dominant $AA$	$\frac{3}{4}$ pigmented
	$\frac{2}{4}$ heterozygous $Aa$	
	$\frac{1}{4}$ homozygous recessive $aa$	$\frac{1}{4}$ albino
	$\frac{1}{4}$ homozygous dominant $DD$	$\frac{3}{4}$ dimpled chin
	$\frac{2}{4}$ heterozygous $Dd$	
	$\frac{1}{4}$ homozygous recessive $dd$	$\frac{1}{4}$ non-dimpled chin

Figure 7-27





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Labradors have a gene for coat color and a gene for vision.

What are the chances that a **heterozygous female for coat color and homozygous dominant for vision** and a **heterozygous male for both traits** will have:

- A black-coated and normal vision offspring?
- A black-coated and blind offspring?
- A chocolate-coated and normal vision offspring?
- A chocolate-coated and blind offspring?

Phenotypes Genotypes				
	Black coat, normal vision $B\_N\_$	Black coat, blind (PRA) $B\_nn$	Chocolate coat, normal vision $bbN\_$	Chocolate coat, blind (PRA) $bbnn$



# A **Test cross** can be used to determine genotype in individuals that display the dominant phenotype

- May be homozygous dominant or heterozygous
- Cross with homozygous recessive to find out...



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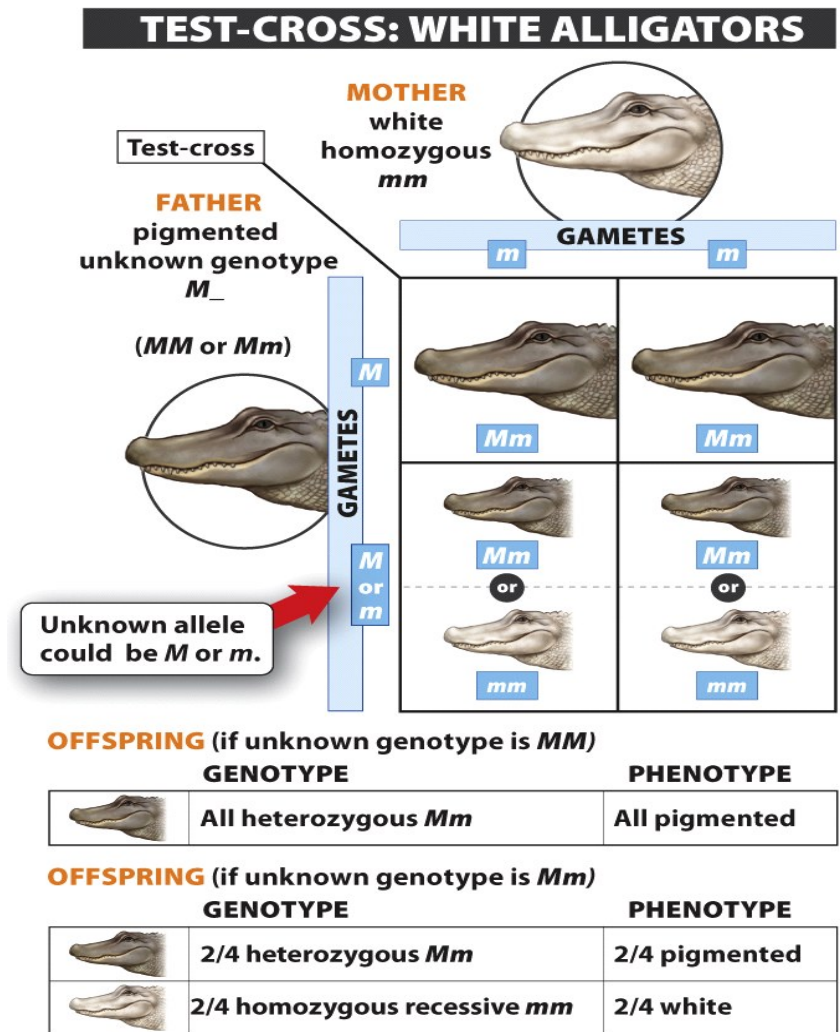
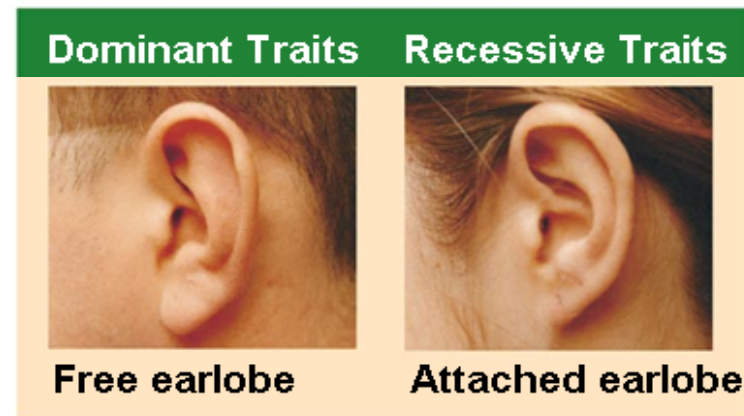


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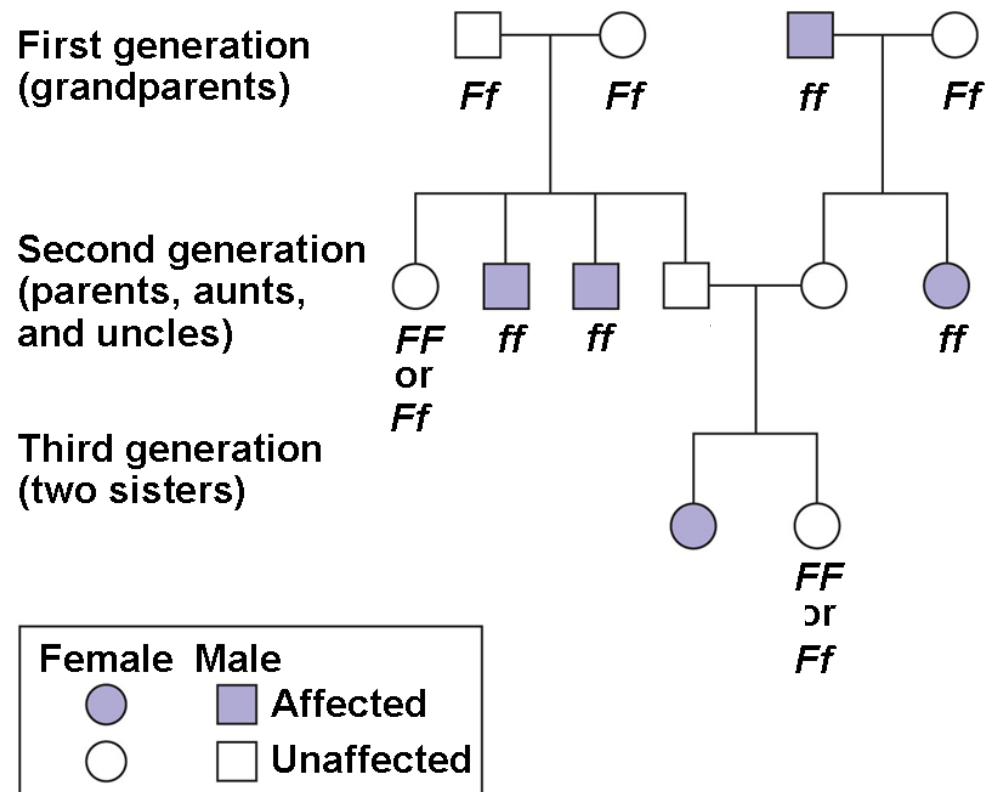
Genetic traits can be tracked through family pedigrees

**Pedigree** = family tree representing the occurrence of heritable traits in parents and offspring across a number of generations

- can determine genotype based on phenotype and family relationships

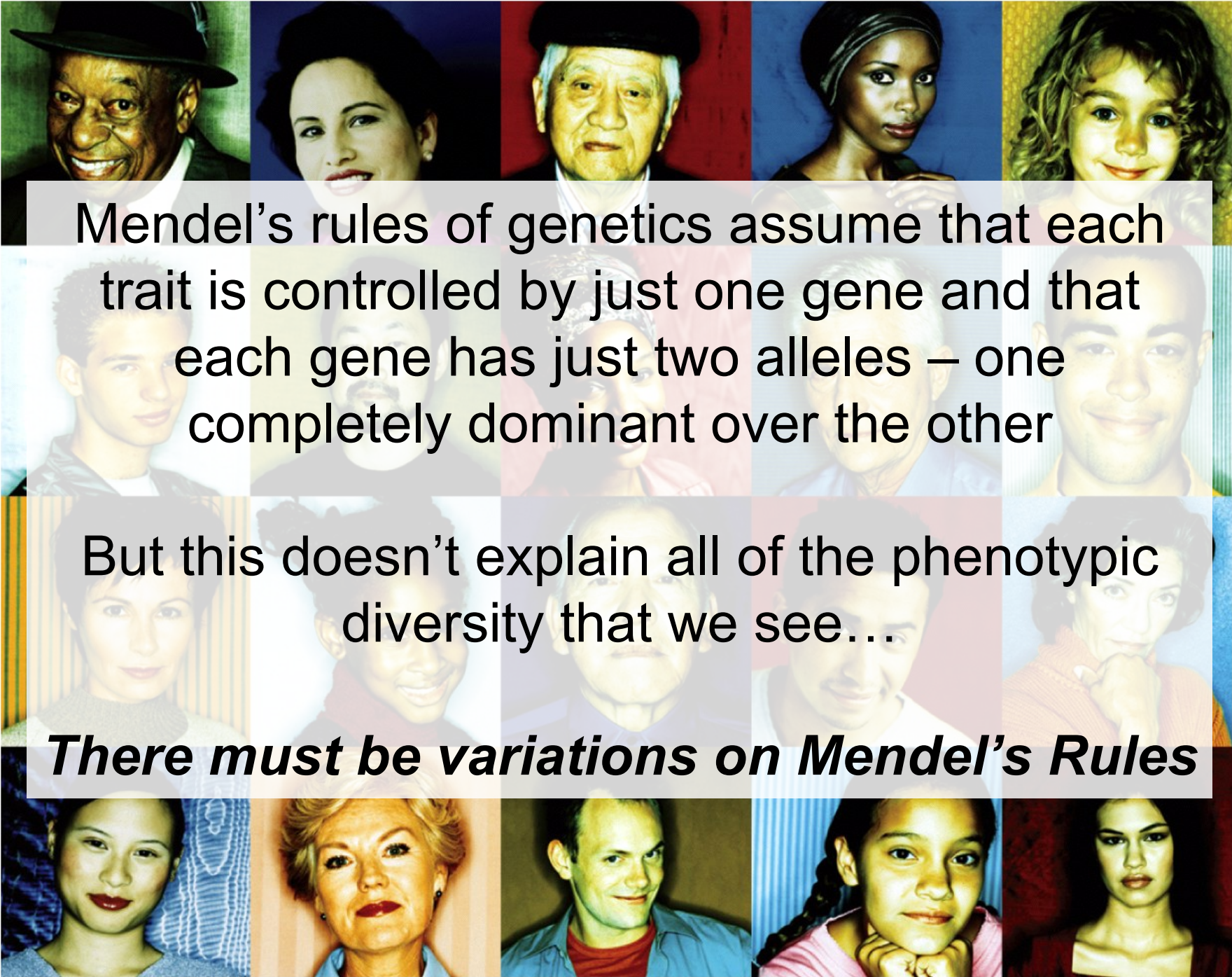


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Mendel's rules of genetics assume that each trait is controlled by just one gene and that each gene has just two alleles – one completely dominant over the other

But this doesn't explain all of the phenotypic diversity that we see...

***There must be variations on Mendel's Rules***



# Variations on Mendel's rules of genetics – Incomplete Dominance

- The phenotype of a heterozygote appears to be **intermediate** between that of the two homozygotes
- No dominant or recessive allele – instead phenotype is intermediate between the two

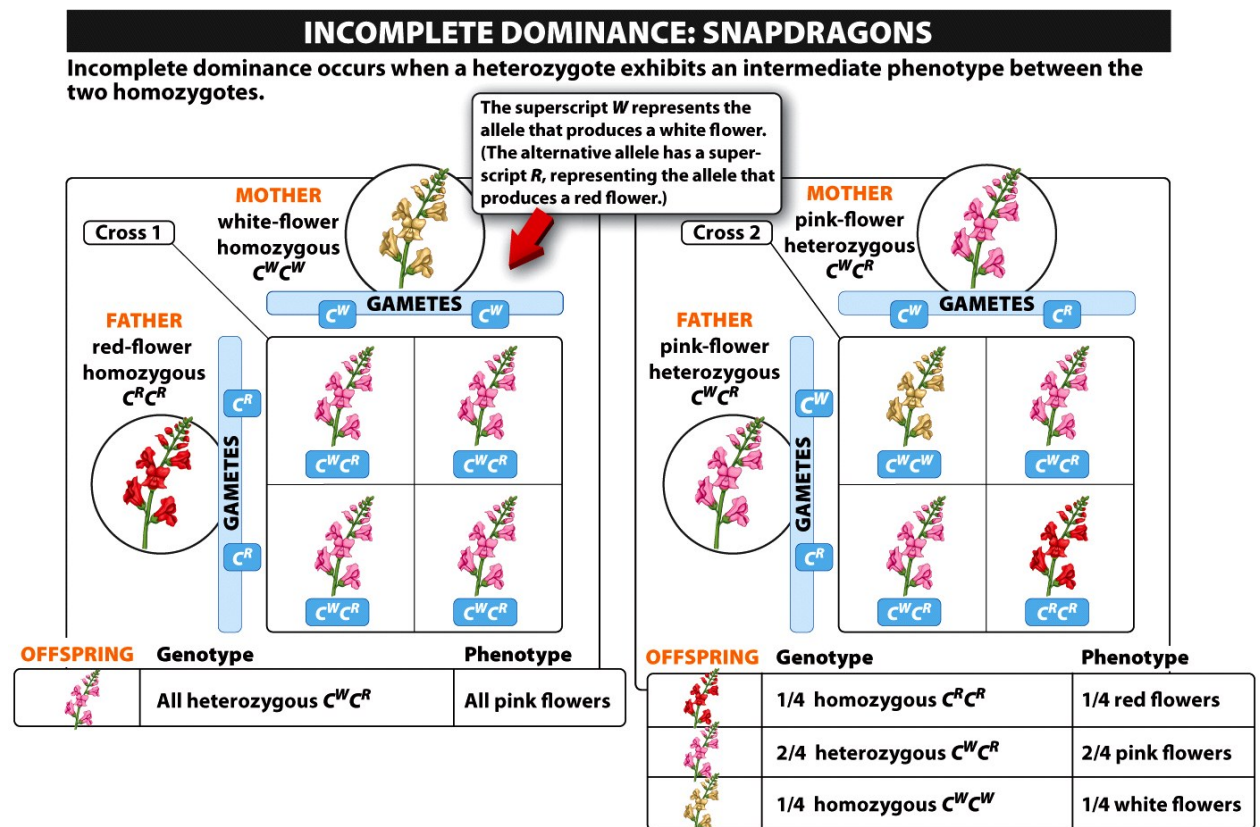
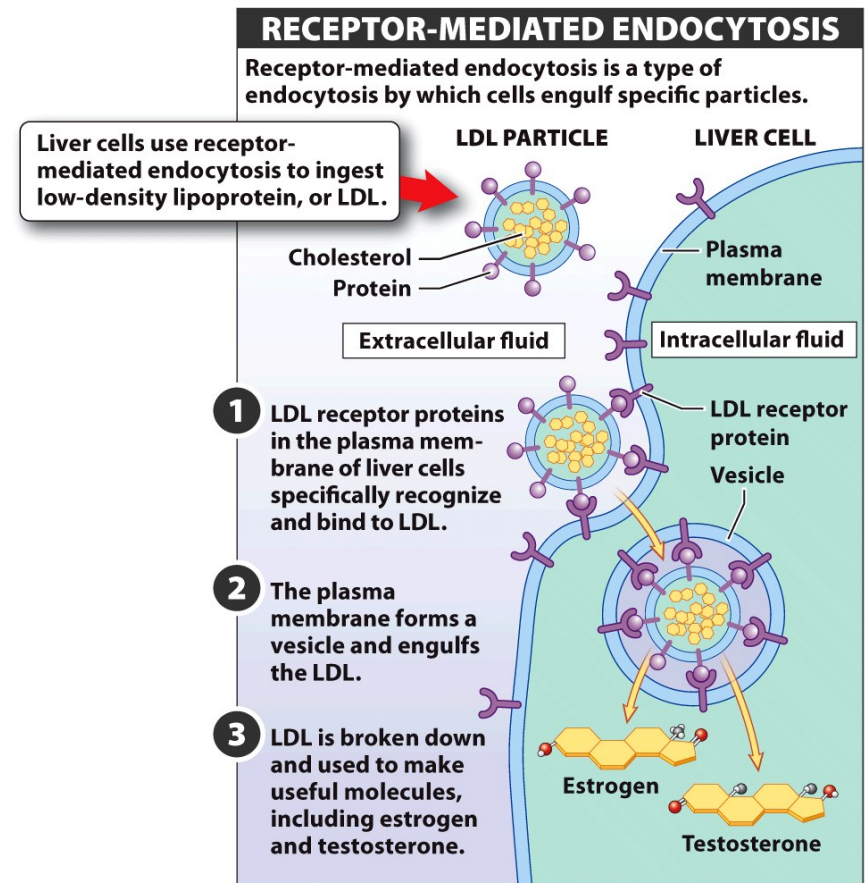
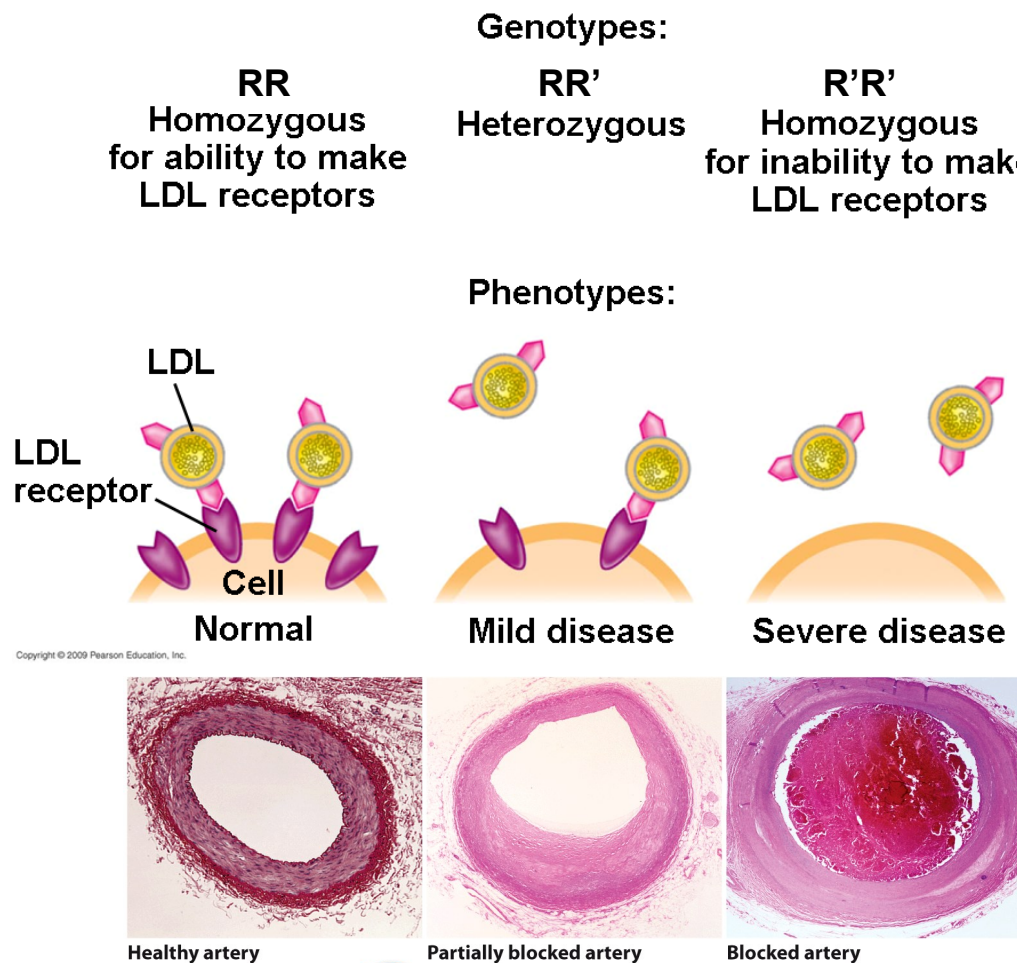


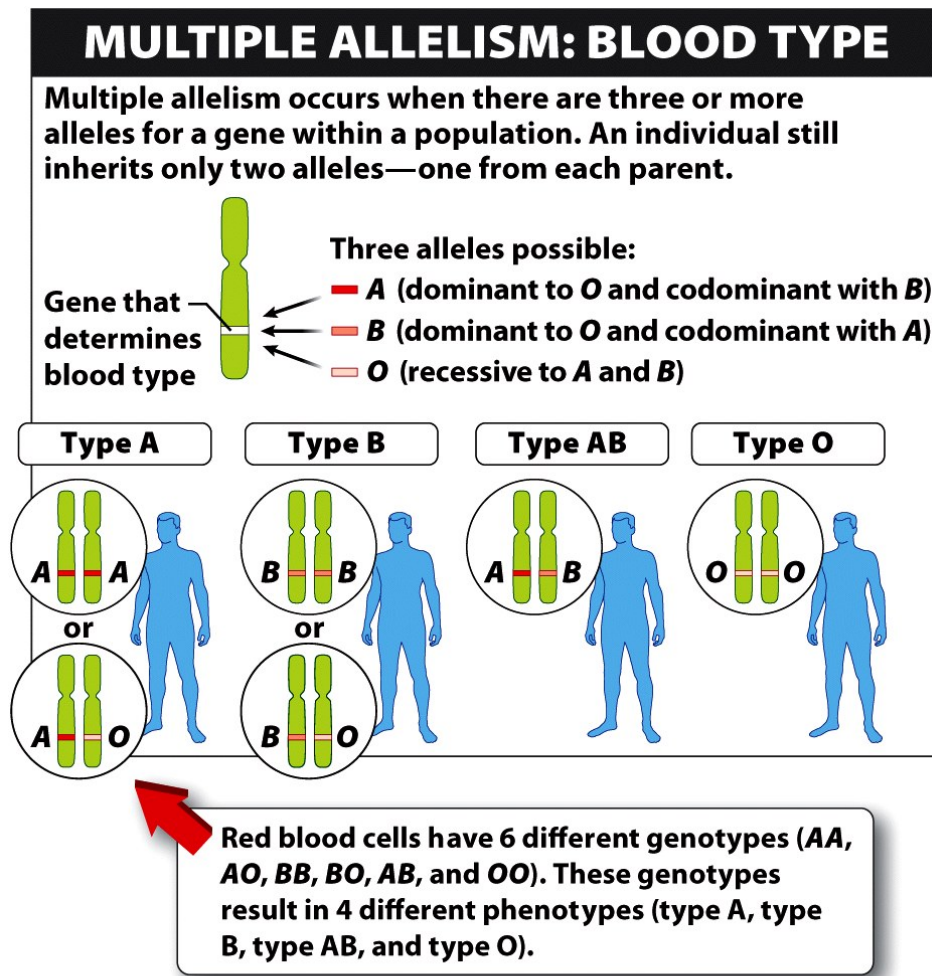
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In humans, the gene to make LDL receptor proteins has two incompletely dominant alleles:  
**R** (protein production) and **R'** (no protein production)



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# Variations on Mendel's rules of genetics – Multiple alleles



- There can be more than two alleles for each gene
- Several phenotypes possible (not just two)



# Variations on Mendel's rules of genetics – Codominance

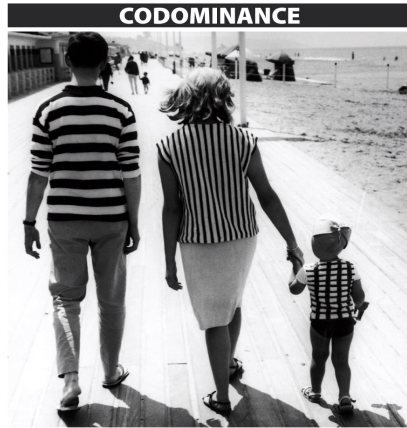


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- Heterozygotes display characteristics of both homozygotes
- No dominant or recessive allele – instead both phenotypes are displayed

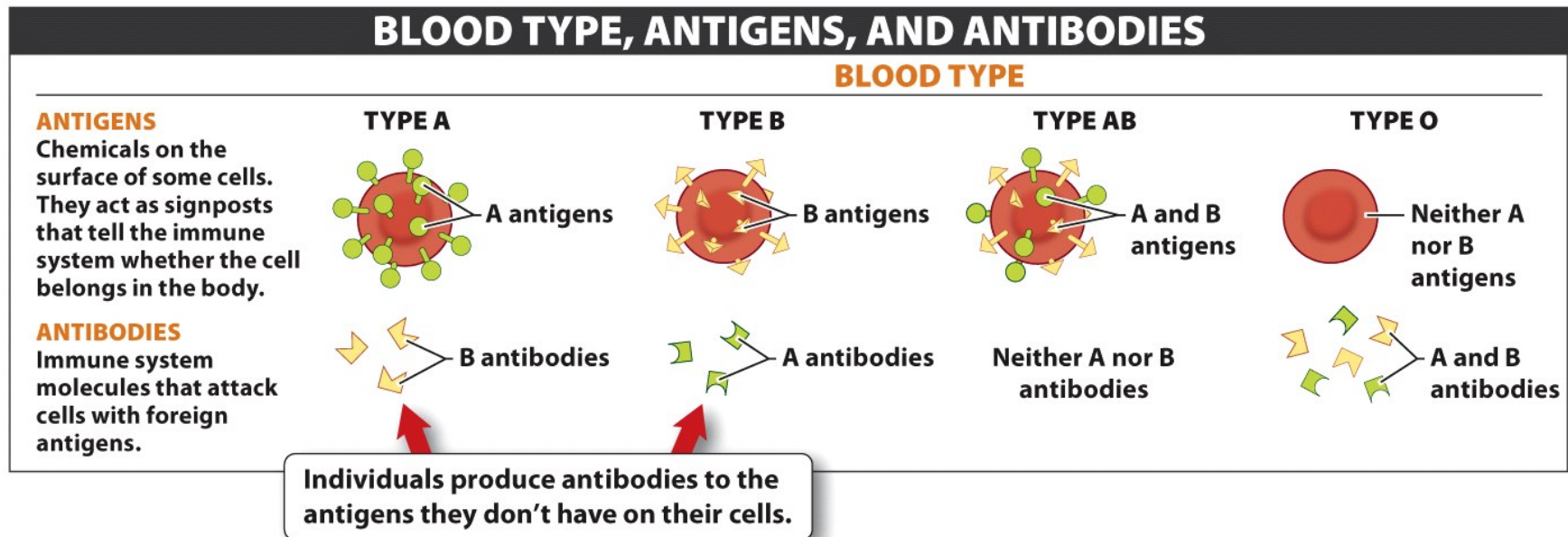


Figure 7-20

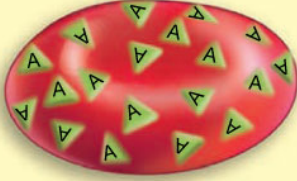
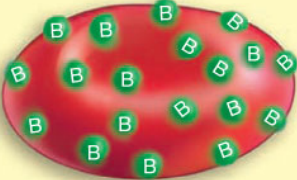
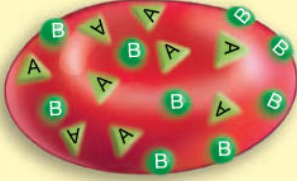

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Table 11.3


**Human Blood Types**

This blood type (phenotype) . . .	. . . has these surface glycolipids . . .	. . . and is produced by these genotypes
A		AA or AO
B		BB or BO
AB		AB
O	 (no surface glycolipids)	OO

The familiar ABO human blood-typing system refers to glycolipid molecules that extend from the surface of red blood cells. People whose blood is “type A” have A extensions on their blood cells. It is also possible to have only B extensions (and be type B); to have both A and B extensions (and be type AB); or to have none of these extensions (and be type O). Note that a person whose genotype is AO is phenotypically type A; likewise, a person whose genotype is BO is phenotypically type B.

Why are people with **type O** blood considered “universal donors”? Why are those with **type AB** considered “universal acceptors”?

THE SCIENCE BEHIND BLOOD DONATION			
BLOOD TYPE	CAN DONATE TO		CAN RECEIVE FROM
 <b>Type A</b> <ul style="list-style-type: none"> <li>• Has A antigens</li> <li>• Produces antibodies that attack B antigens</li> </ul>	<b>Type A</b> <b>Type AB</b>  		<b>Type A</b> <b>Type O</b>  
 <b>Type B</b> <ul style="list-style-type: none"> <li>• Has B antigens</li> <li>• Produces antibodies that attack A antigens</li> </ul>	<b>Type B</b> <b>Type AB</b>  		<b>Type B</b> <b>Type O</b>  
 <b>Type AB</b> <ul style="list-style-type: none"> <li>• Has A and B antigens</li> <li>• Produces neither A nor B antibodies</li> <li>• Universal recipient</li> </ul>	<b>Type AB</b> 		<b>Type A</b> <b>Type B</b> <b>Type AB</b> <b>Type O</b>    
 <b>Type O</b> <ul style="list-style-type: none"> <li>• Has neither A nor B antigens</li> <li>• Produces antibodies that attack A and B antigens</li> <li>• Universal donor</li> </ul>	<b>Type A</b> <b>Type B</b> <b>Type AB</b> <b>Type O</b>    		<b>Type O</b> 



**Individuals with type O blood are universal donors.  
Individuals with type AB are universal recipients.**

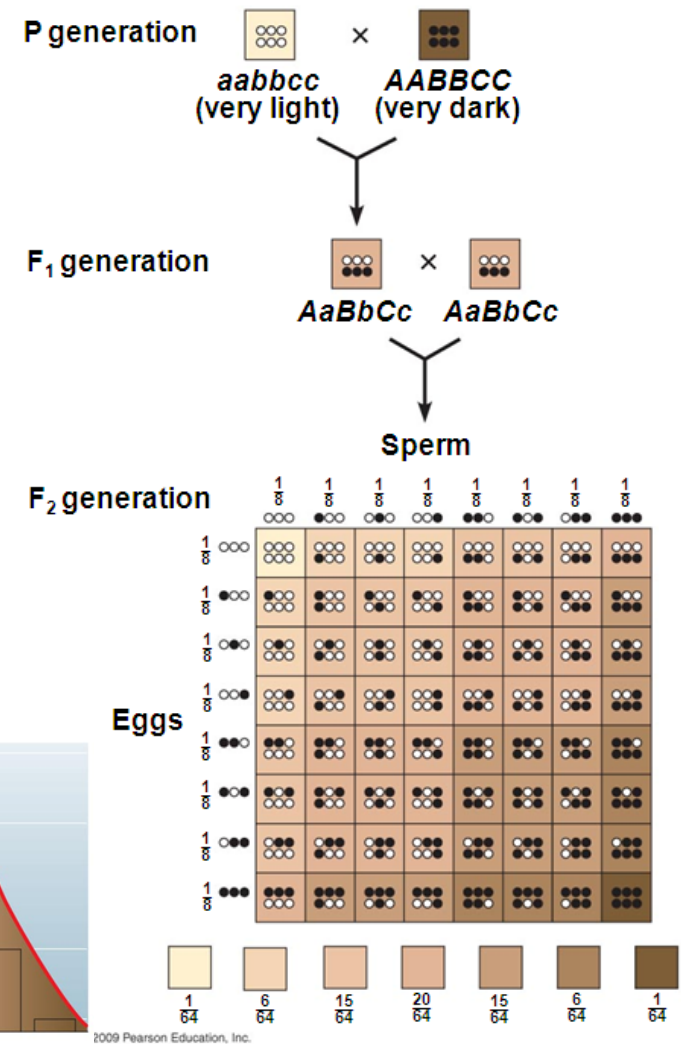
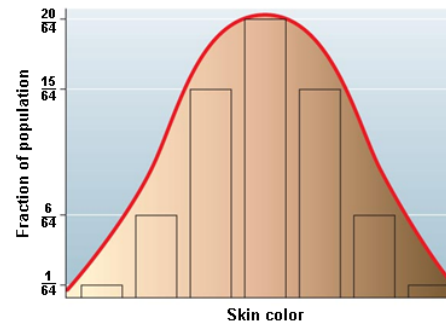
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# Variations on Mendel's rules of genetics – Polygenic traits

- Traits that are controlled by more than one gene
- Additive effects of all the genes together produce a continuum of phenotypes



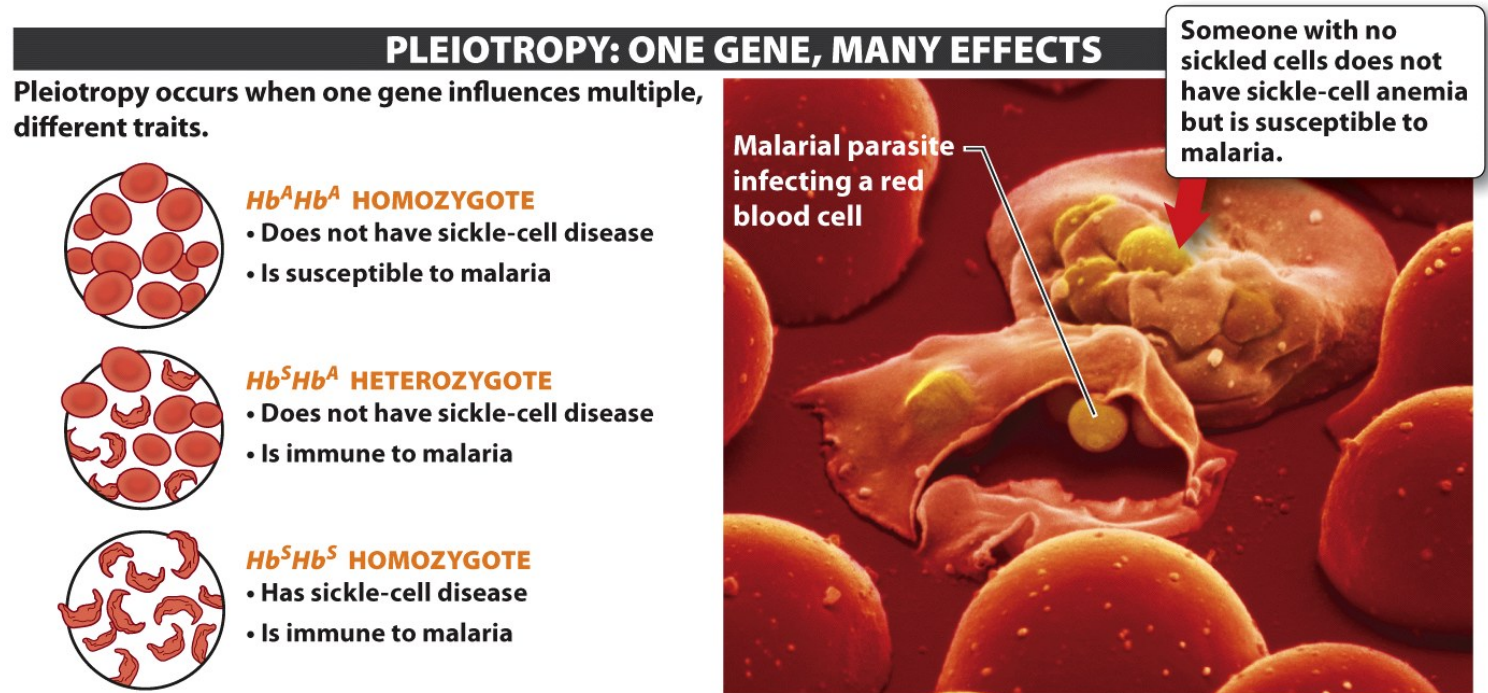
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# Variations on Mendel's rules of genetics – **Pleiotropy**

- Individual genes can influence multiple traits (phenotypes)
- Ex: Hemoglobin gene in red blood cells has multiple effects on health and function

What is  
the benefit of  
“almost”  
having sickle  
cell disease?



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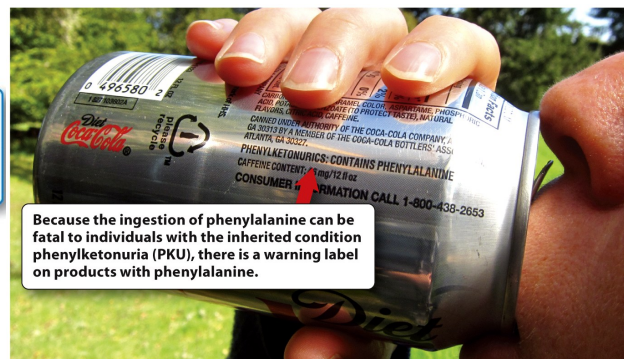


# Variations on Mendel's rules of genetics – The role of the Environment



**Some pigment genes produce dark pigment only under cold conditions—such as on the tail, nose, ears, and feet of these animals.**

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**Figure 7-25**  
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- The environment can influence how and if genes are expressed
- Environment can be changed to offset the effect of genes
- More the rule than an exception

# Variations on Mendel's rules of genetics – Linked genes



- Genes on the same chromosome are usually inherited together (chromosomes are packaged into gametes)
- Exception: Crossing over

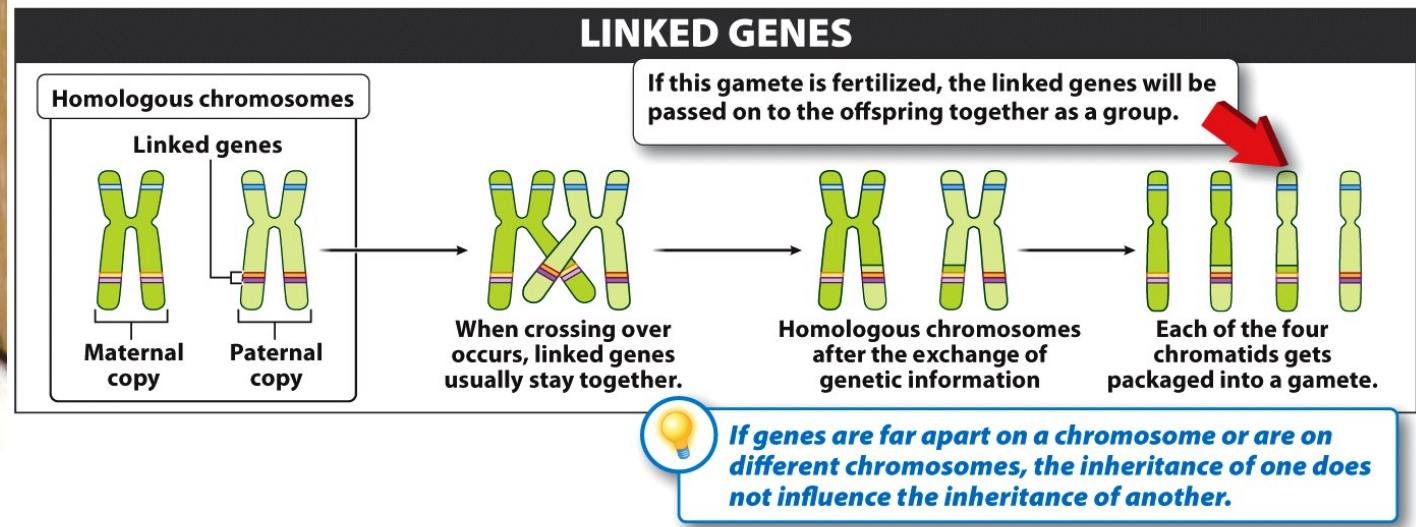


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# Variations on Mendel's rules of genetics – Sex-linked traits

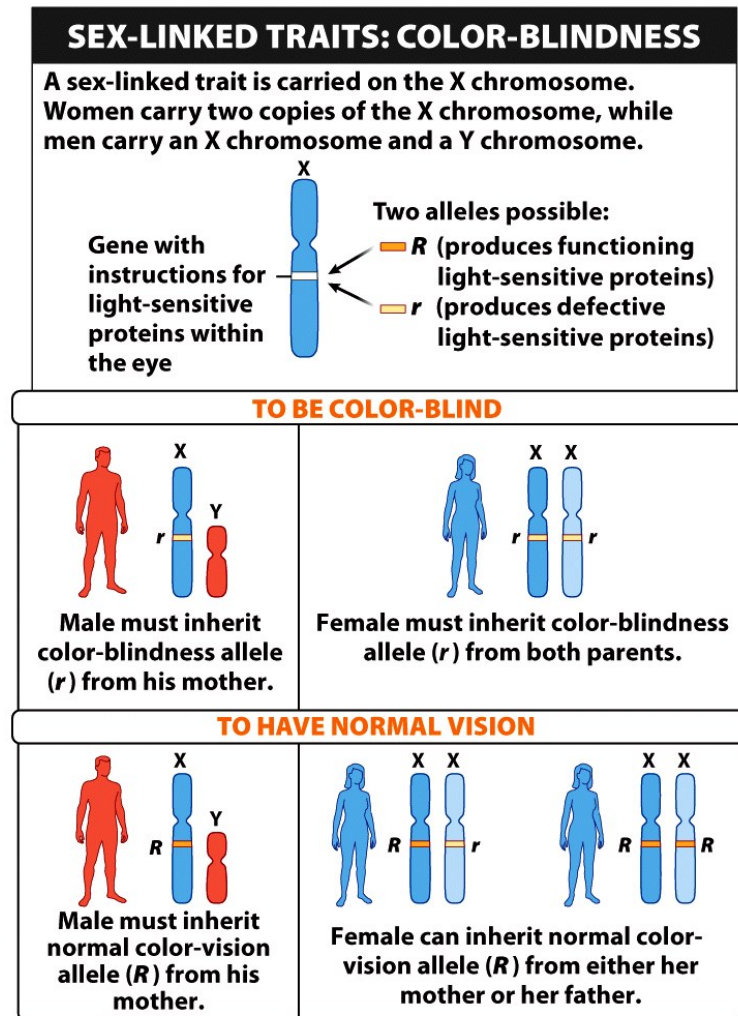
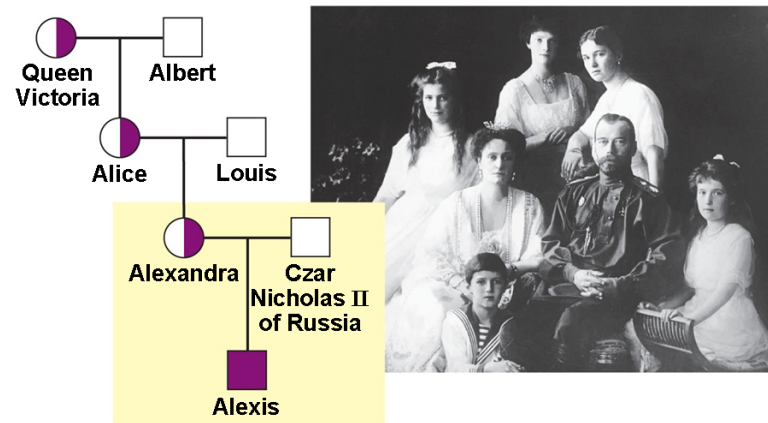
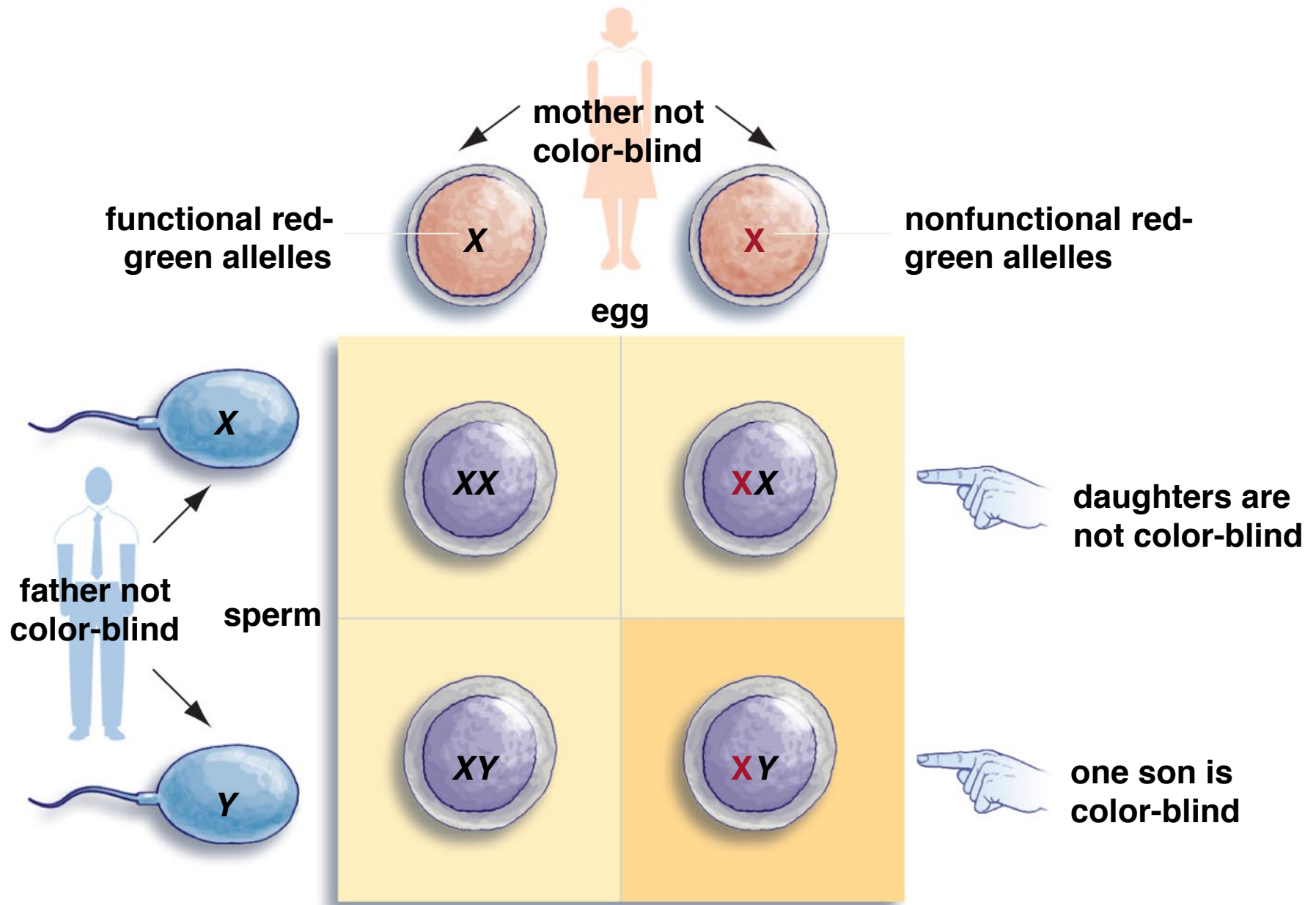


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- X chromosome has many more genes than the Y chromosome
- Females have two Xs and males only have one X
- Sex-linked disorders usually affect males and not females (however, females can be **carriers**)



# X-linked Inheritance





Hemophilia is a blood-clotting disorder caused by a defective recessive gene on the X chromosome.

If a mother is a carrier for hemophilia ( $X^N X^n$ ), and a father is not affected ( $Y X^N$ ), what is the chance that they will have a baby with hemophilia?


# Summary

- Modern genetics began with Gregor Mendel's pea plant experiments
- Alleles are different forms of a gene, and are generally either dominant or recessive
- Offspring have two alleles for every gene – one from each parent (homozygous vs. heterozygous)
- Mendel's rules include the Law of segregation and the Law of independent assortment
- Variation on Mendel's rules include incomplete dominance, multiple alleles, codominance, polygenic traits, pleiotropy the role of the environment, linked genes, and sex-linked traits

## **By the end of this chapter, you should be able to:**

- 1) Describe Gregor Mendel's inheritance experiments involving pea plants
- 2) List and describe Mendel's three hypotheses of inheritance
- 3) Define the Law of segregation and the Law of independent assortment
- 4) Utilize pedigrees and punnett squares to identify individuals and to determine genotypic and phenotypic probabilities
- 5) Describe the role of carriers in autosomal and sex-linked disorders
- 6) Describe the variations on Mendel's laws and be able to utilize these concepts in genetics problems