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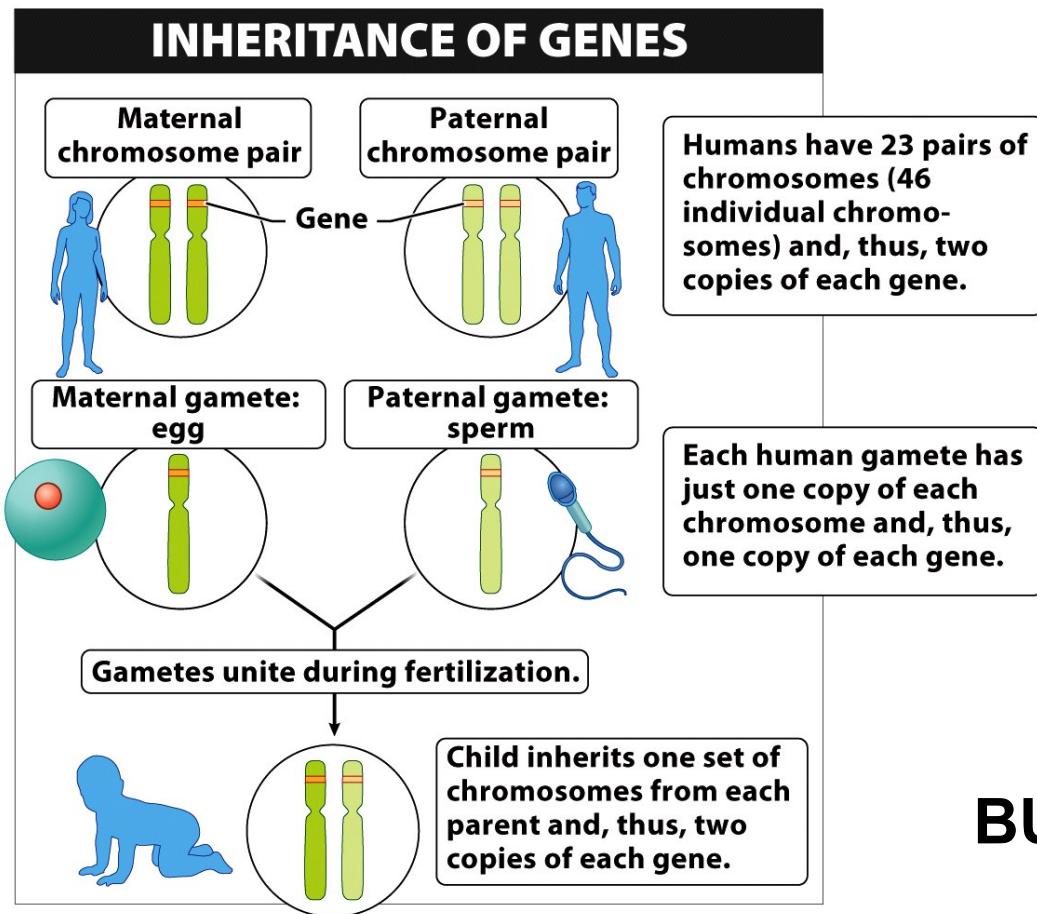
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Genes and Inheritance

(11-12)

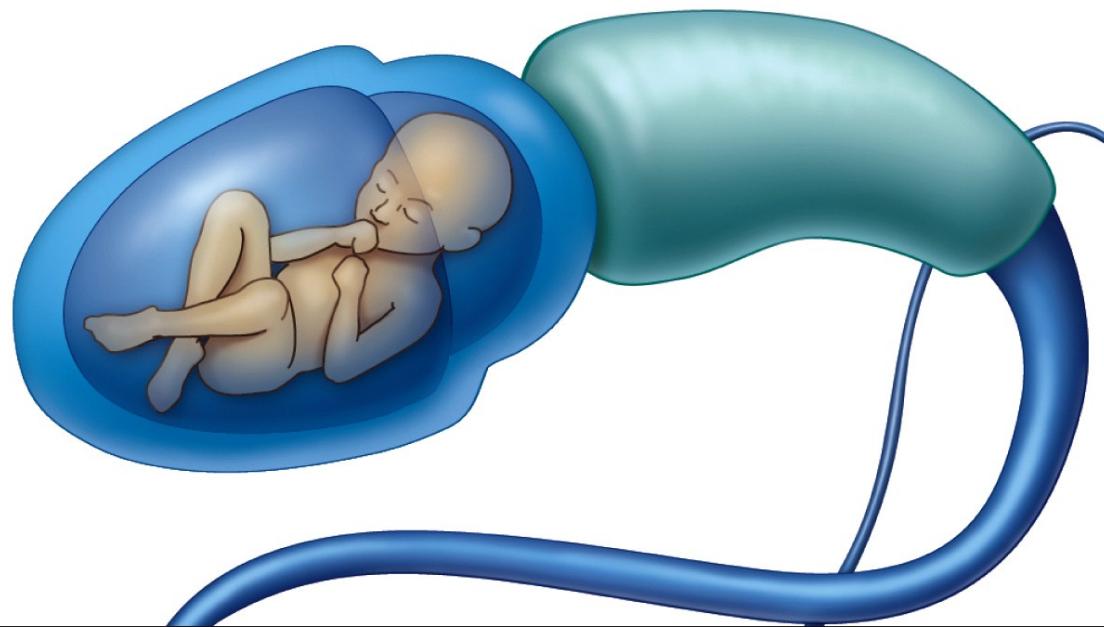
You are a unique combination of your two parents



- 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

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.

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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.

Gregor Mendel (1822-1884)

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

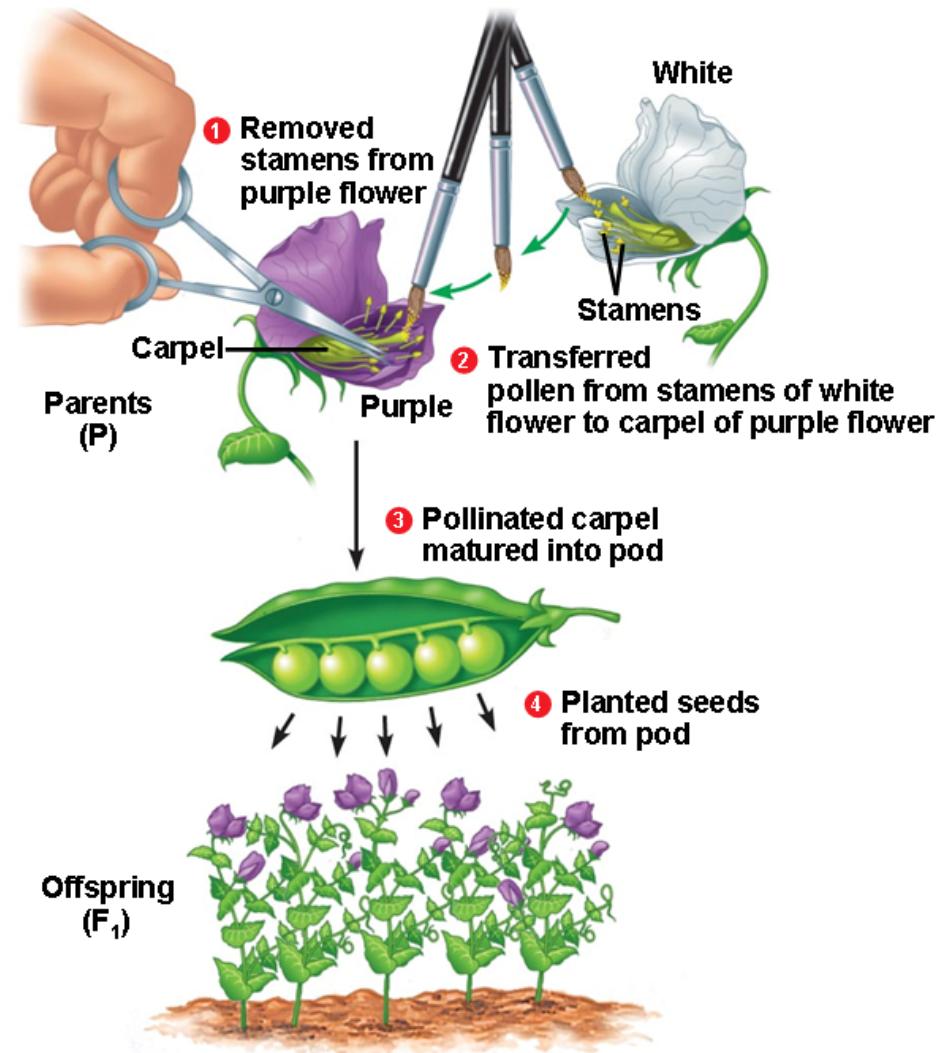
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Mendel could control mating and produce lots of offspring

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

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



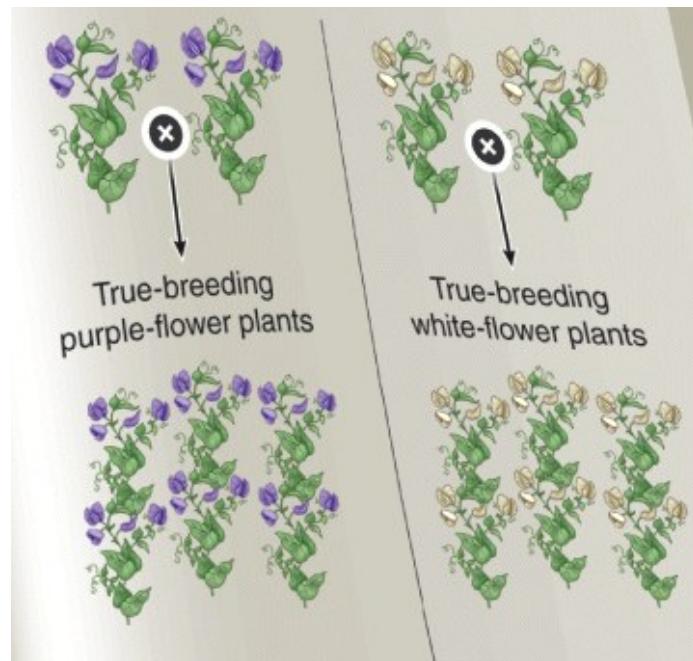
TRAITS (GENES)

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

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VARIANTS (ALLELES)

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



P generation
(true-breeding
parents)



×



Purple flowers

White flowers

F₁ generation



All plants have
purple flowers

↓

Fertilization
among F₁ plants
(F₁ × F₁)

F₂ generation



$\frac{3}{4}$ of plants
have purple flowers $\frac{1}{4}$ of plants
have white flowers

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

Which flower color
allele is
dominant?

Mendel's Three Hypotheses of Inheritance

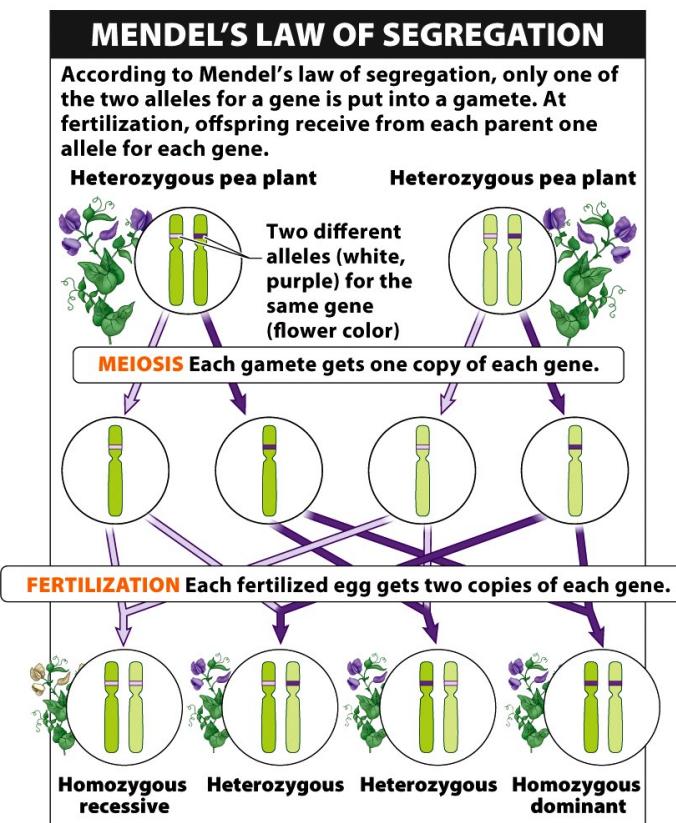
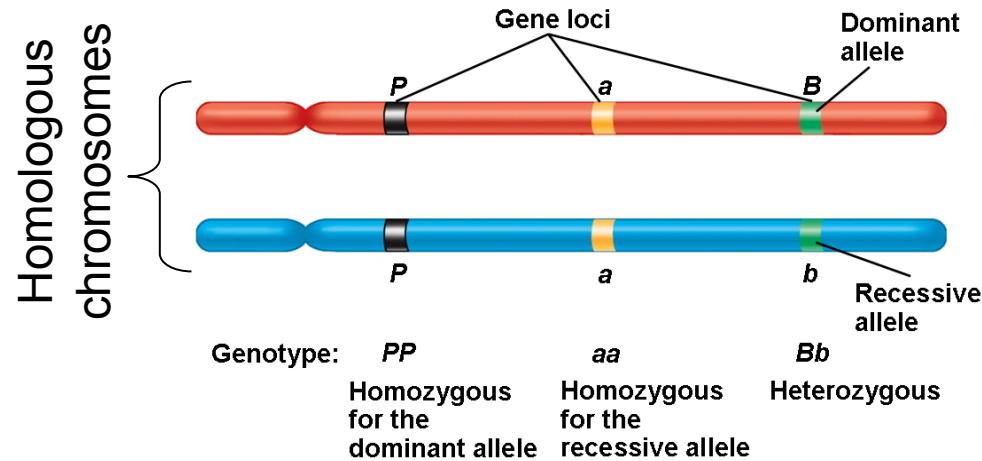
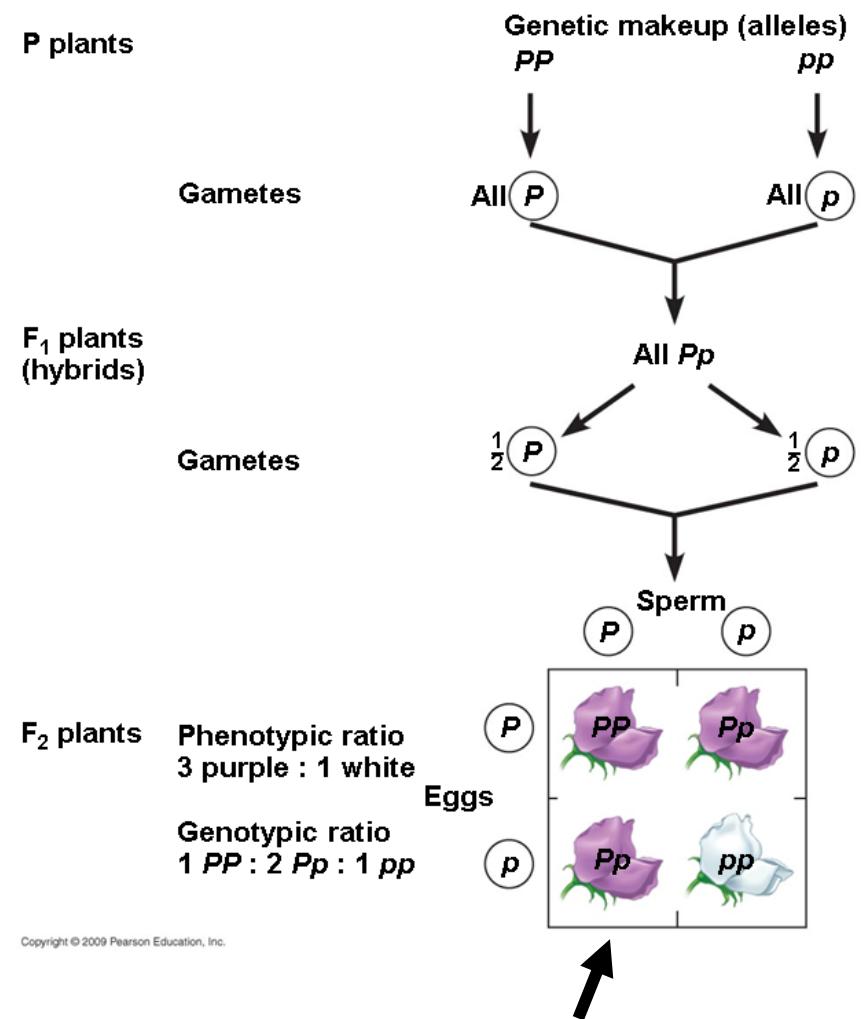
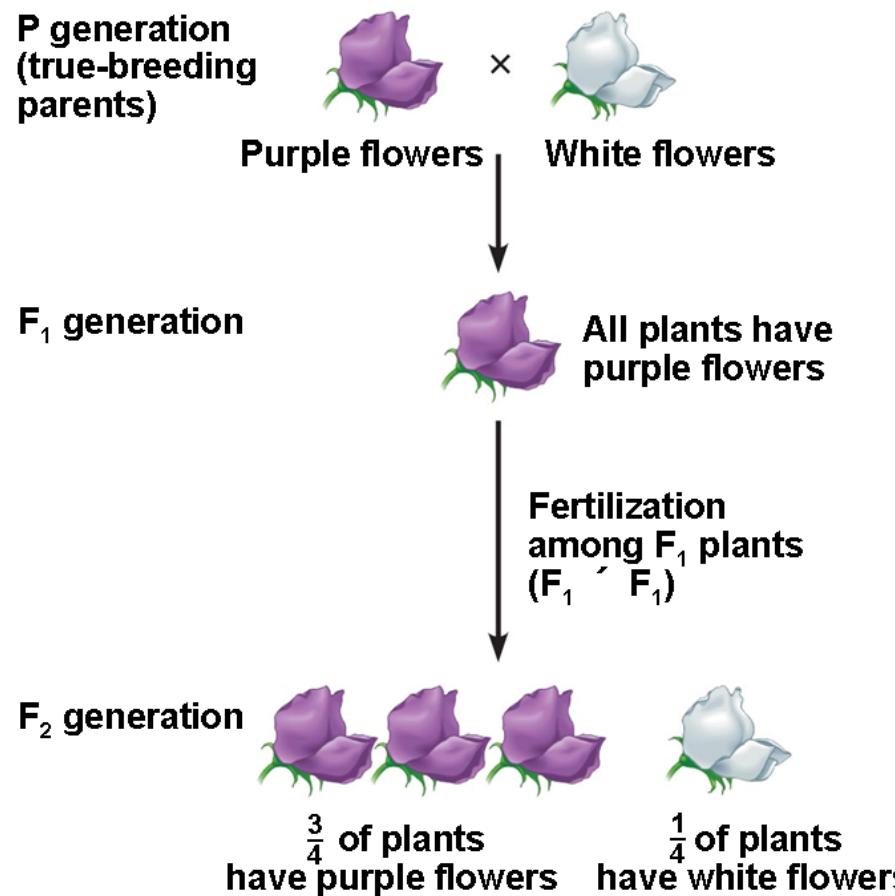


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- 1) Gametes carry only one copy of each gene because homologous chromosomes separate (segregate) during meiosis = **Law of Segregation**
- 2) An organism inherits one gene from each parent (**Genotype**)
 - Homozygous vs. Heterozygous
- 3) Inherited genes determine the outer appearance of the organism (**Phenotype**)
 - Dominant vs. Recessive



Were Mendel's hypotheses correct?



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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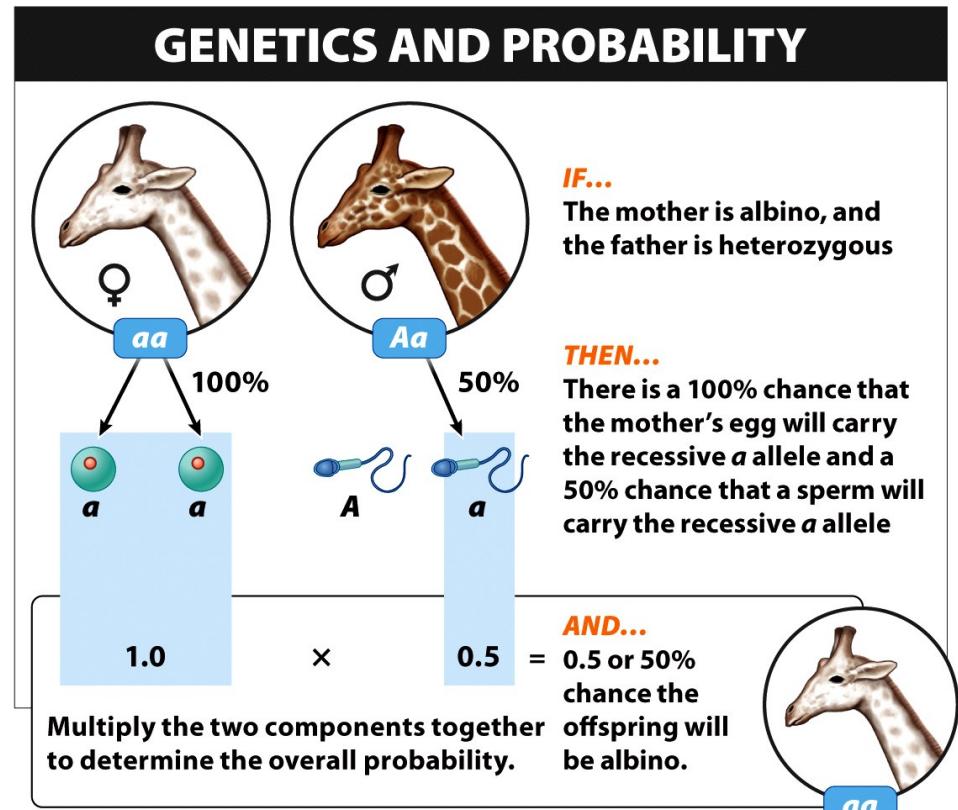


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MENDEL'S LAW OF INDEPENDENT ASSORTMENT

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

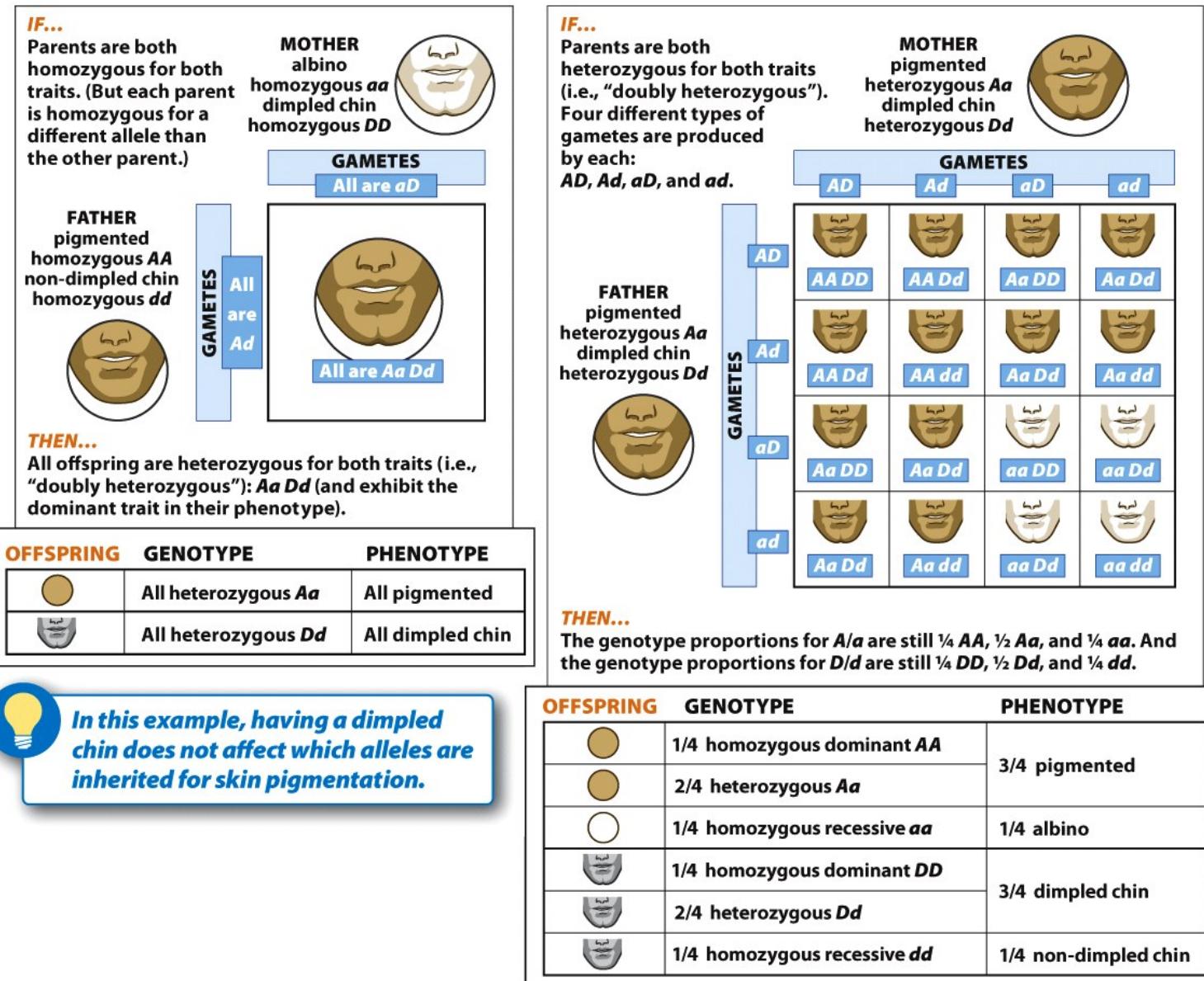


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Labradors have a gene for coat color and a gene for vision.

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) $bbnn$

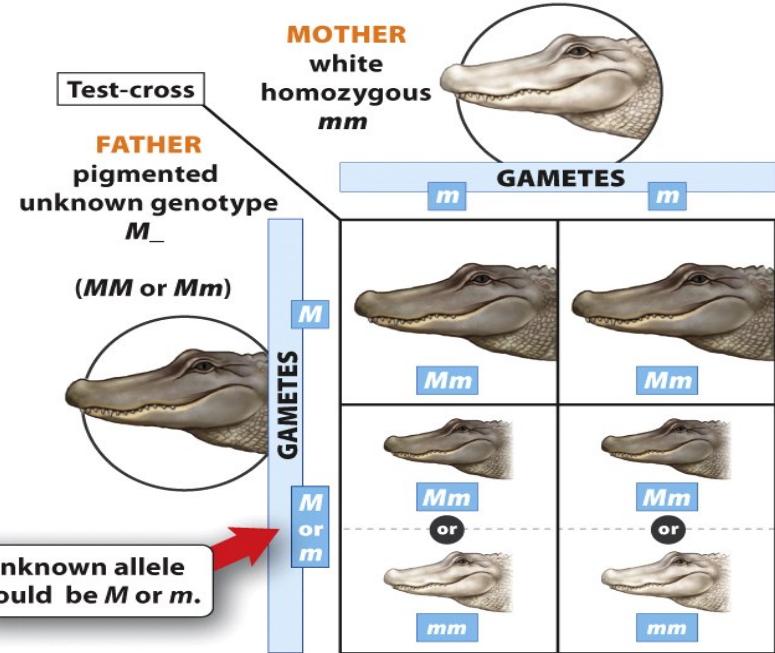
A Test cross can be used to determine genotype in individuals that display the dominant phenotype

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



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TEST-CROSS: WHITE ALLIGATORS



OFFSPRING (if unknown genotype is MM)

GENOTYPE	PHENOTYPE
All heterozygous Mm	All pigmented

OFFSPRING (if unknown genotype is Mm)

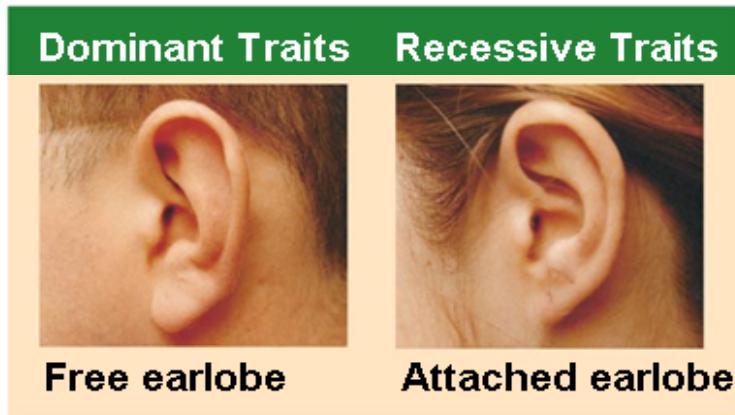
GENOTYPE	PHENOTYPE
2/4 heterozygous Mm	2/4 pigmented
2/4 homozygous recessive mm	2/4 white

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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



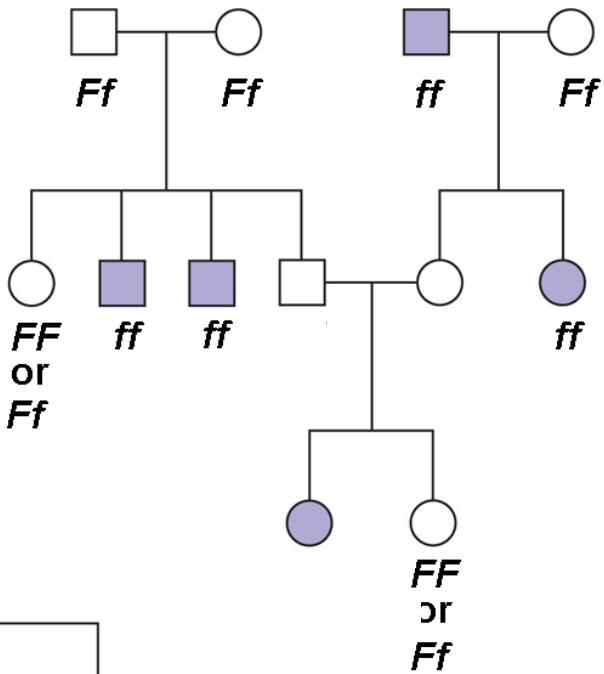
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First generation
(grandparents)

Second generation
(parents, aunts,
and uncles)

Third generation
(two sisters)

Female	Male
●	■ Affected
○	□ Unaffected



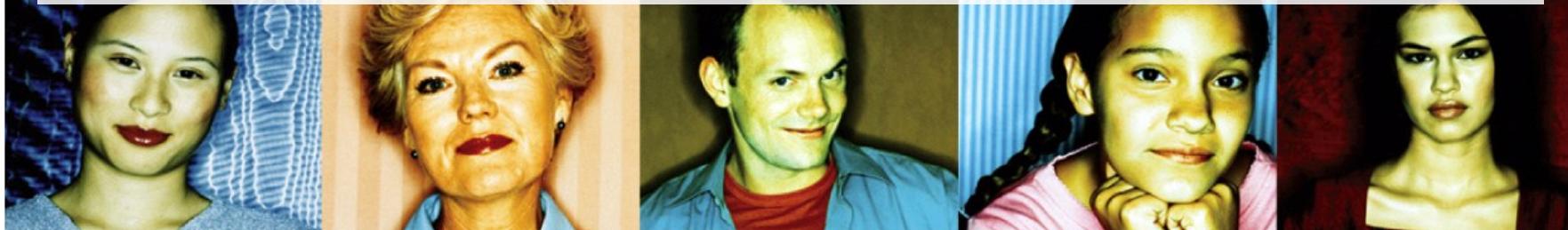
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Mendel's rules of genetics assume that each trait is controlled by just one gene and that each gene has just two alleles – one completely dominant over the other

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

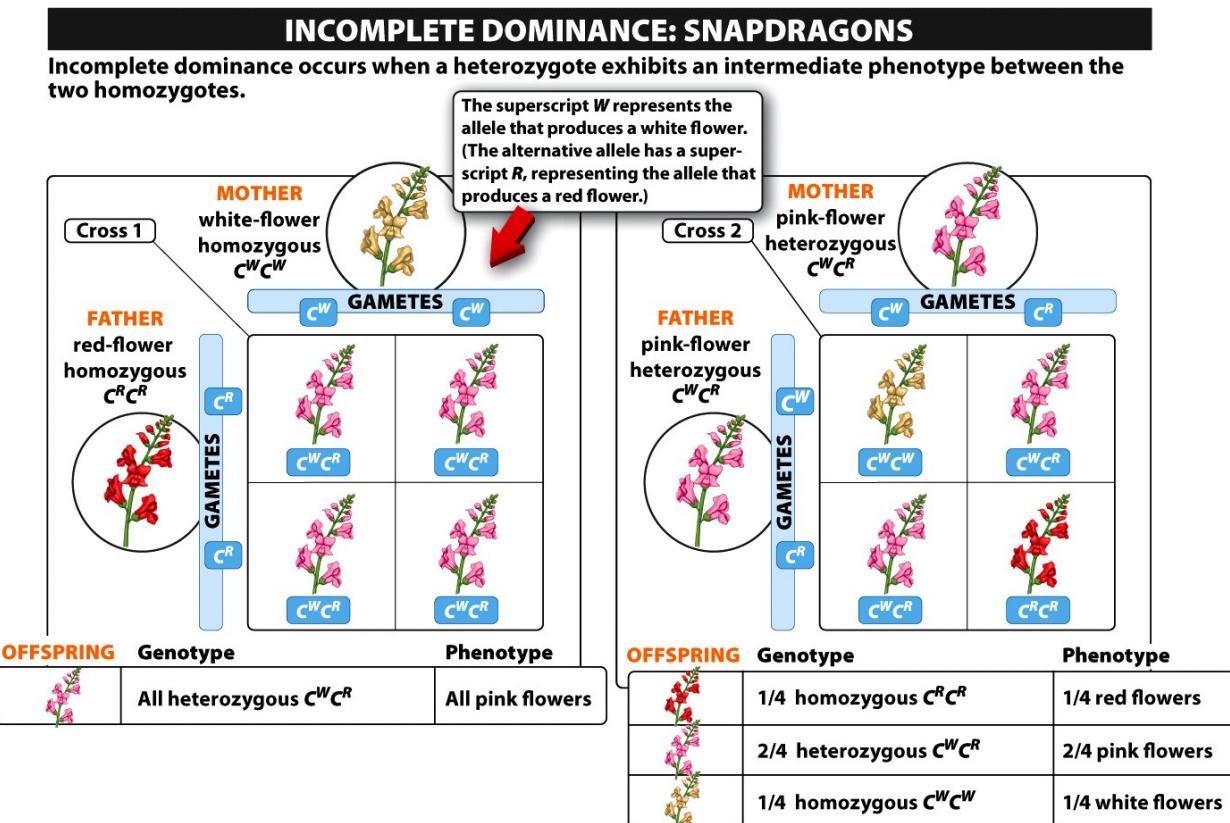


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In humans, the gene to make LDL receptor proteins has two incompletely dominant alleles: **R** (protein production) and **R'** (no protein production)

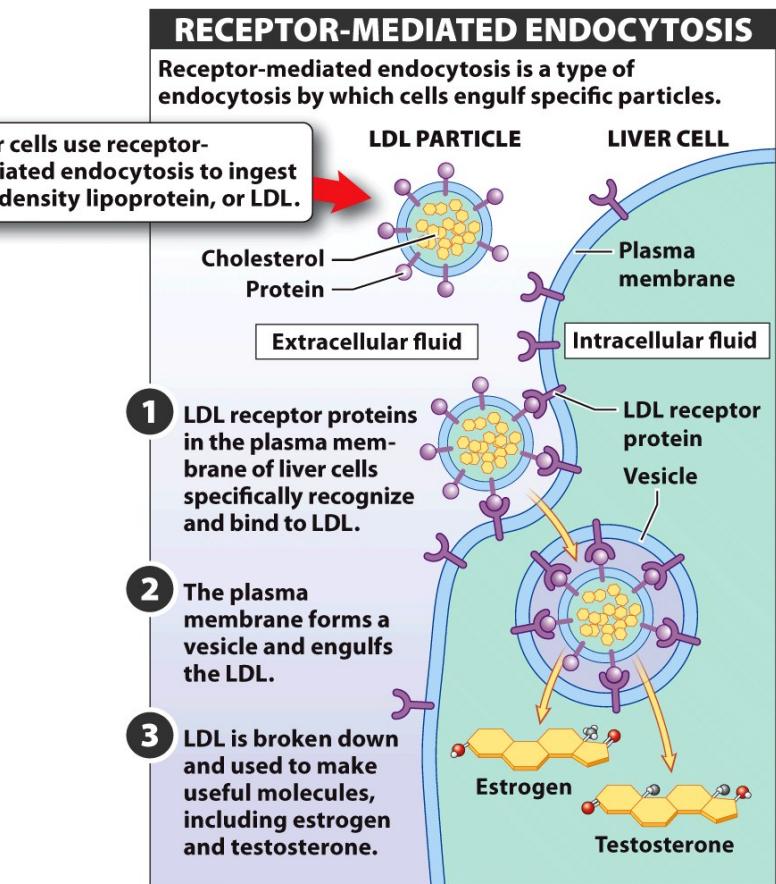
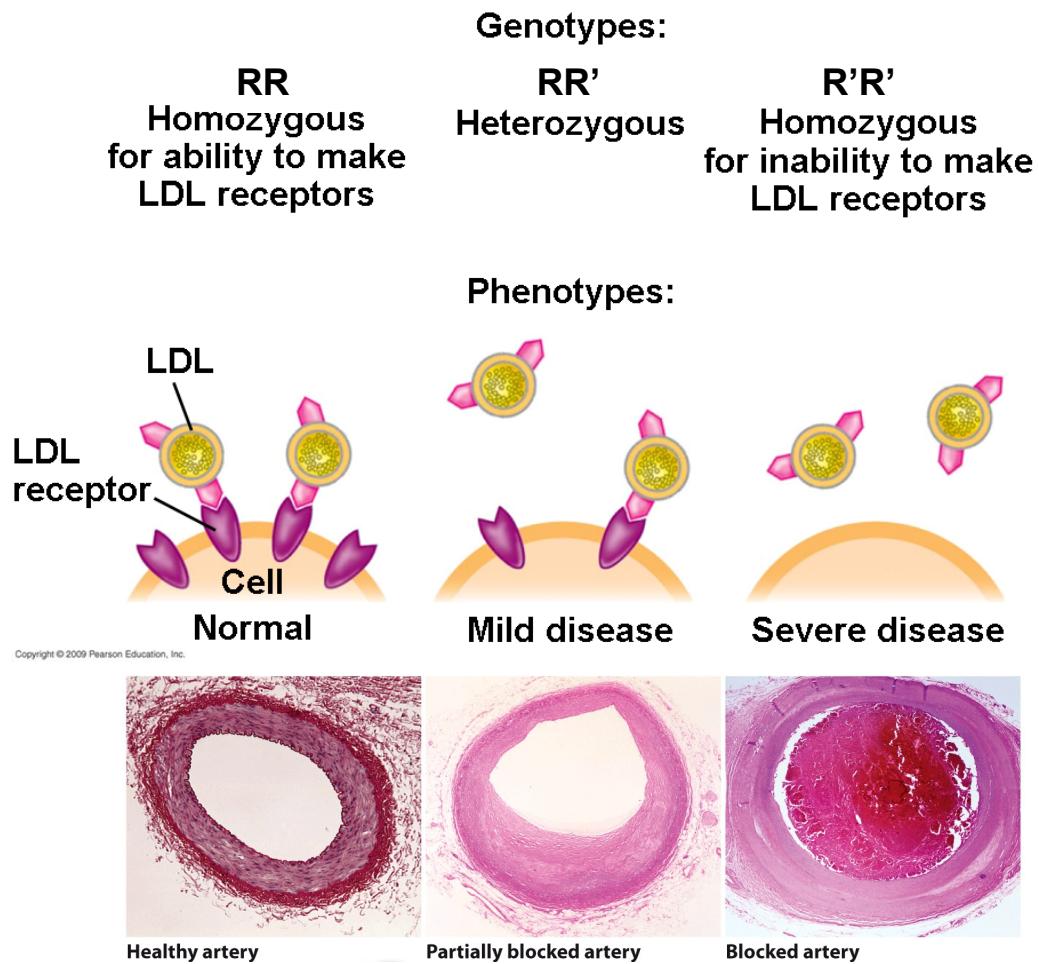
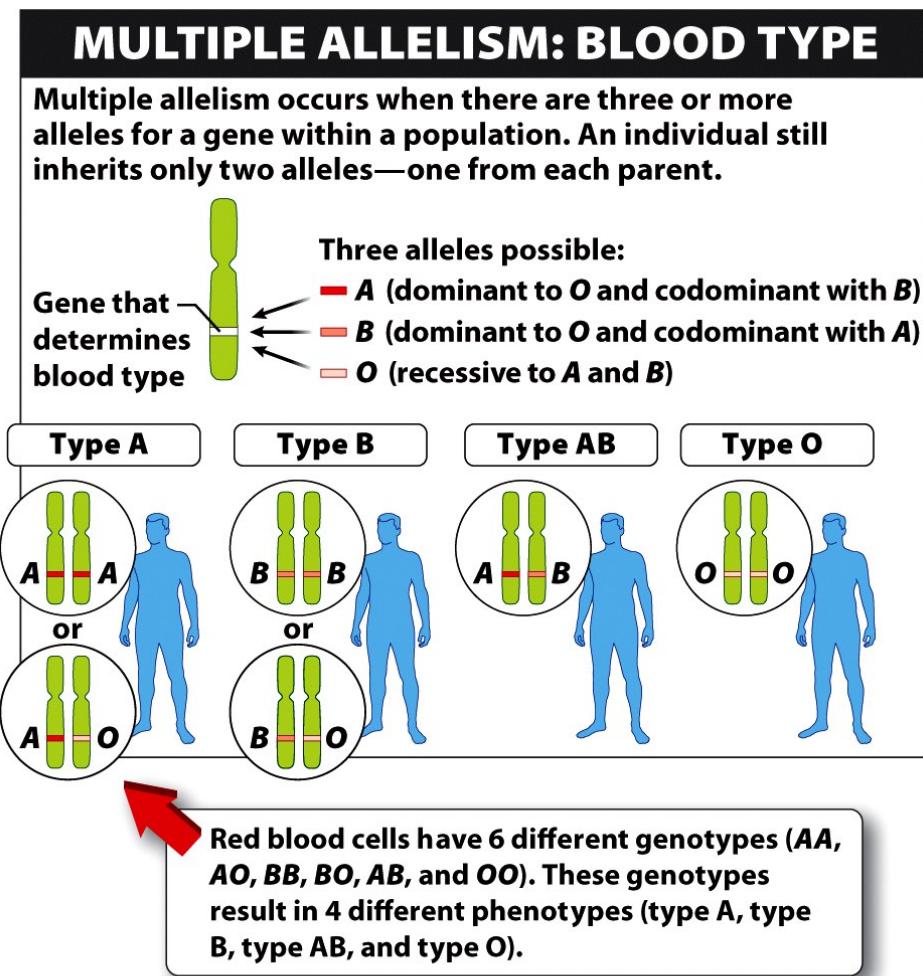


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Variations on Mendel's rules of genetics – Multiple alleles



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

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Variations on Mendel's rules of genetics – Codominance

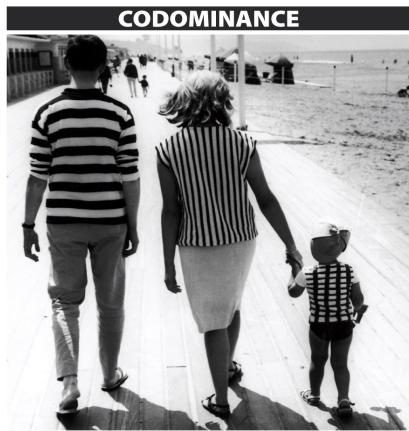


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- Heterozygotes display characteristics of both homozygotes
- No dominant or recessive allele – instead both phenotypes are displayed

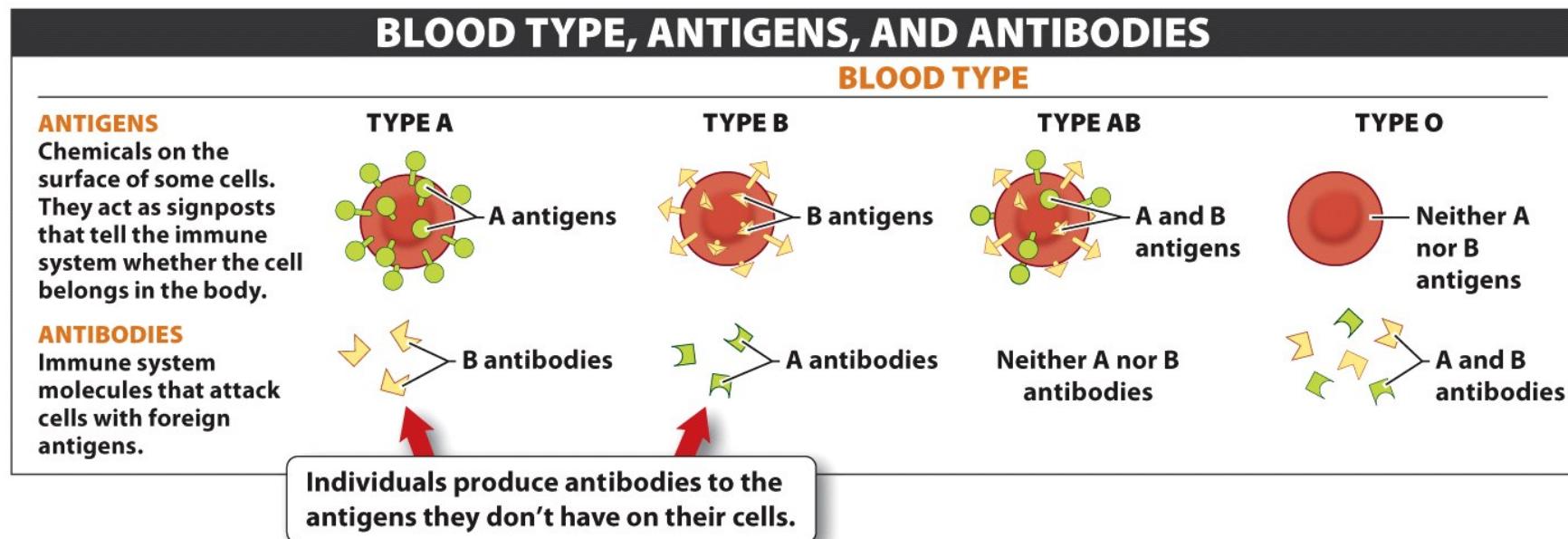


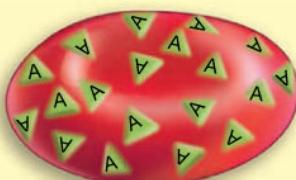
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Table 11.3

Human Blood Types

This blood type . . . has these surface glycolipids . . . and is produced by these genotypes

A



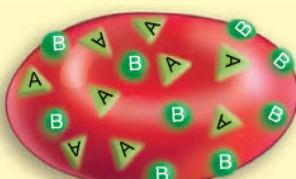
AA or AO

B



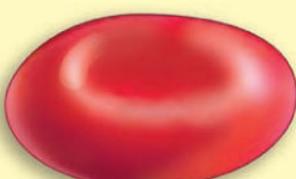
BB or BO

AB



AB

O



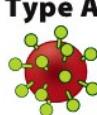
OO

(no surface glycolipids)

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
 <p>Type A • Has A antigens • Produces antibodies that attack B antigens</p>	 	 
 <p>Type B • Has B antigens • Produces antibodies that attack A antigens</p>	 	 
 <p>Type AB • Has A and B antigens • Produces neither A nor B antibodies • Universal recipient</p>		   
 <p>Type O • Has neither A nor B antigens • Produces antibodies that attack A and B antigens • Universal donor</p>	   	



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

Figure 7-21

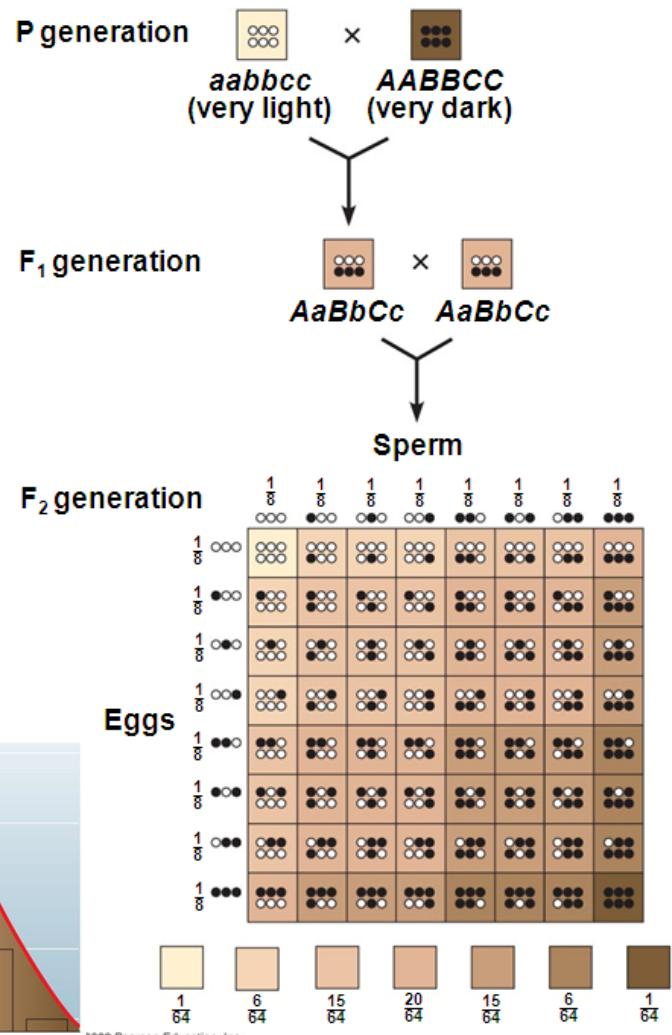
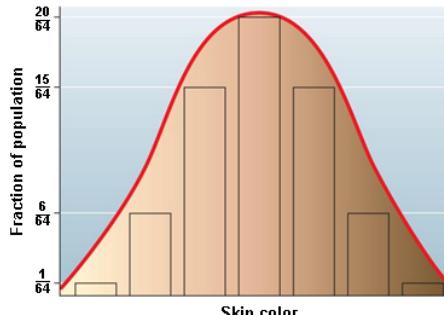
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Variations on Mendel's rules of genetics – **Polygenic traits**

- 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?

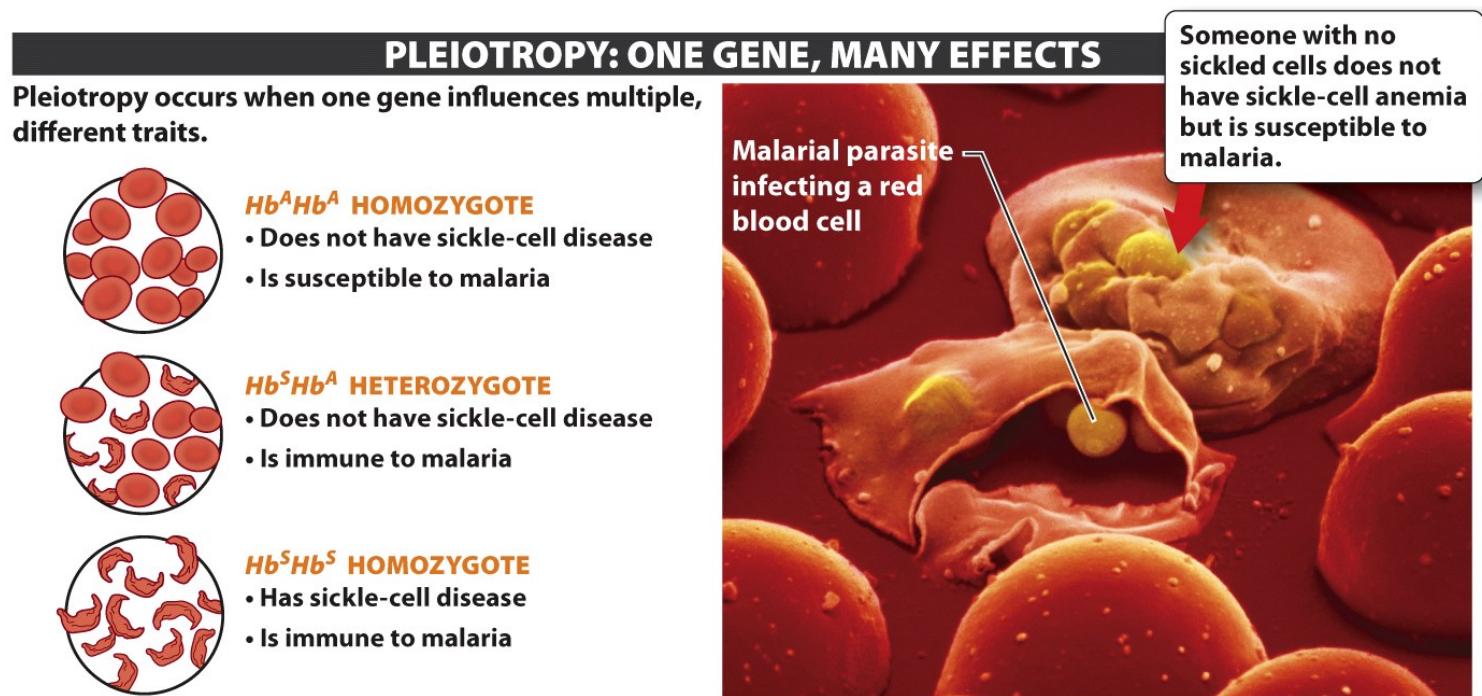


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Variations on Mendel's rules of genetics – The role of the Environment



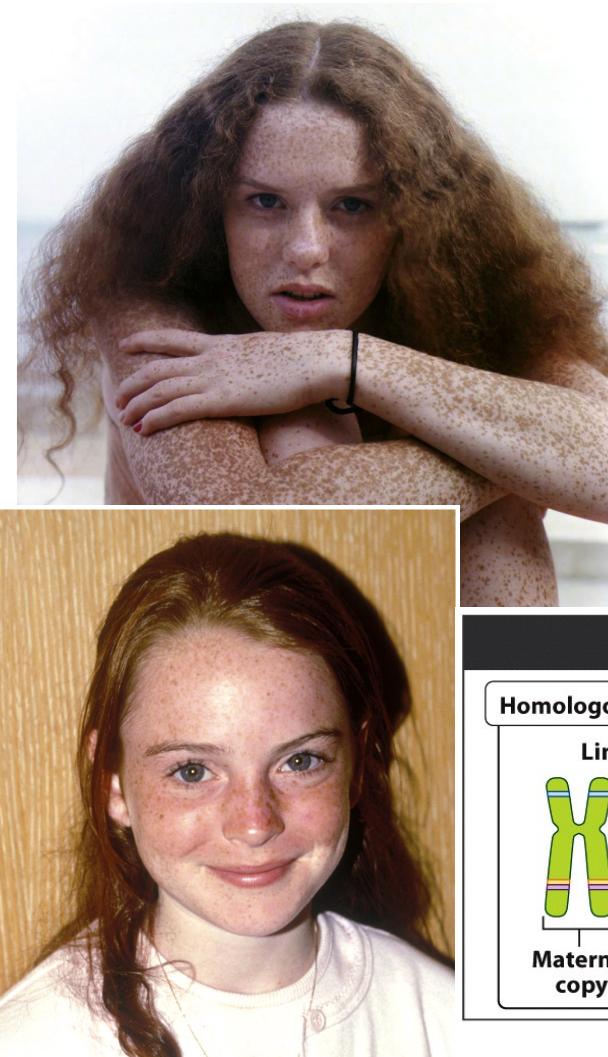
Some pigment genes produce dark pigment only under cold conditions—such as on the tail, nose, ears, and feet of these animals.



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- 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

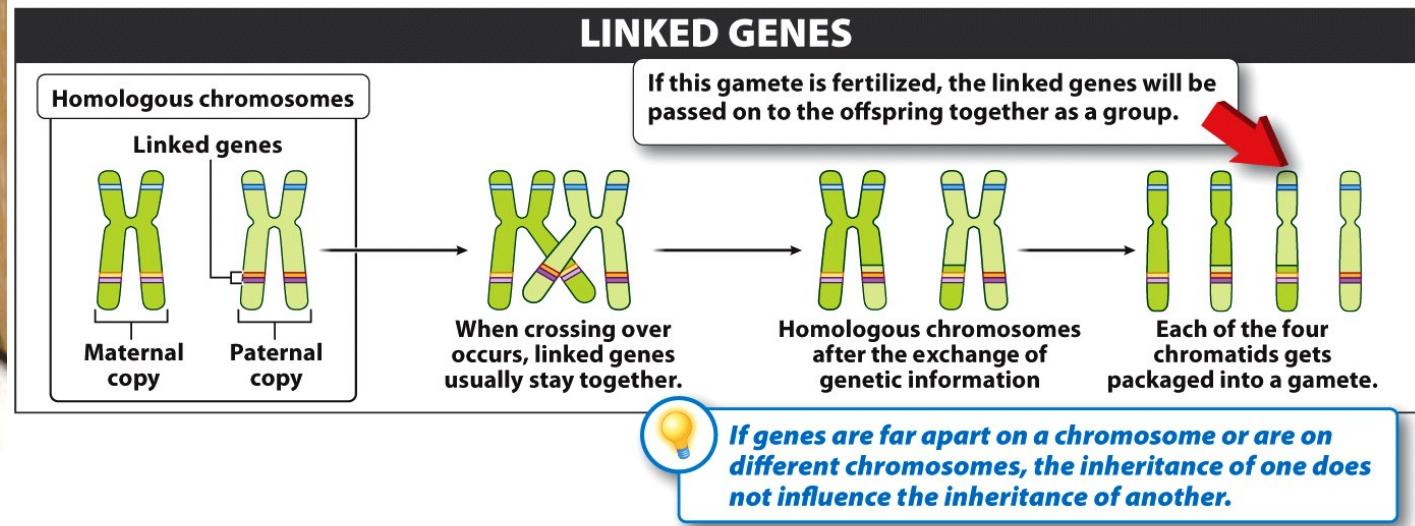
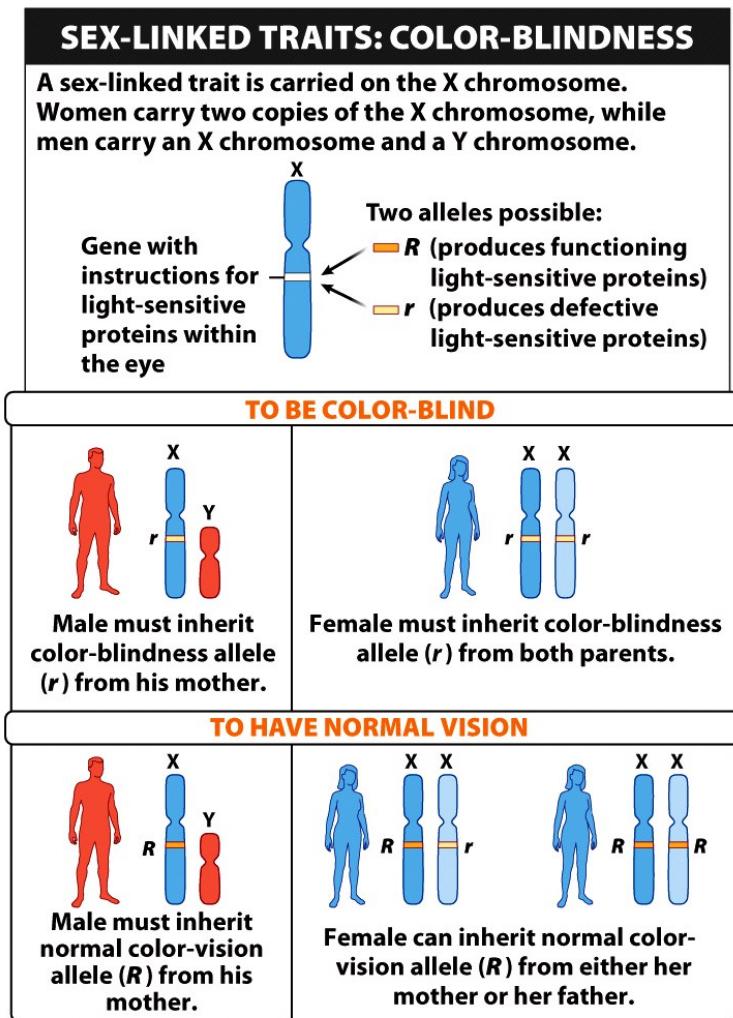


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Variations on Mendel's rules of genetics – Sex-linked traits



- 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**)

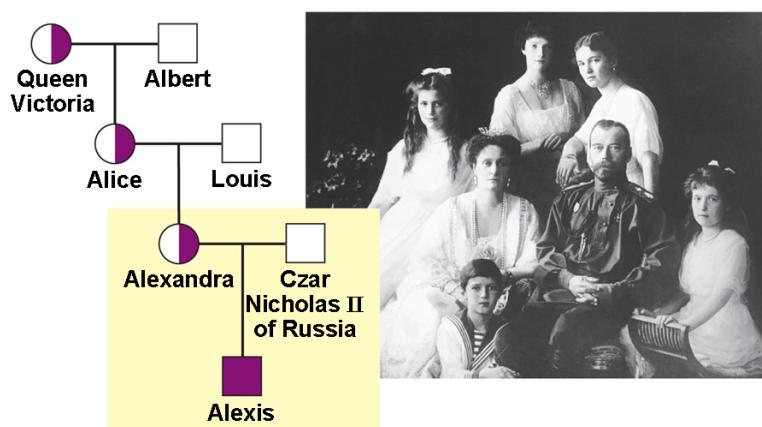
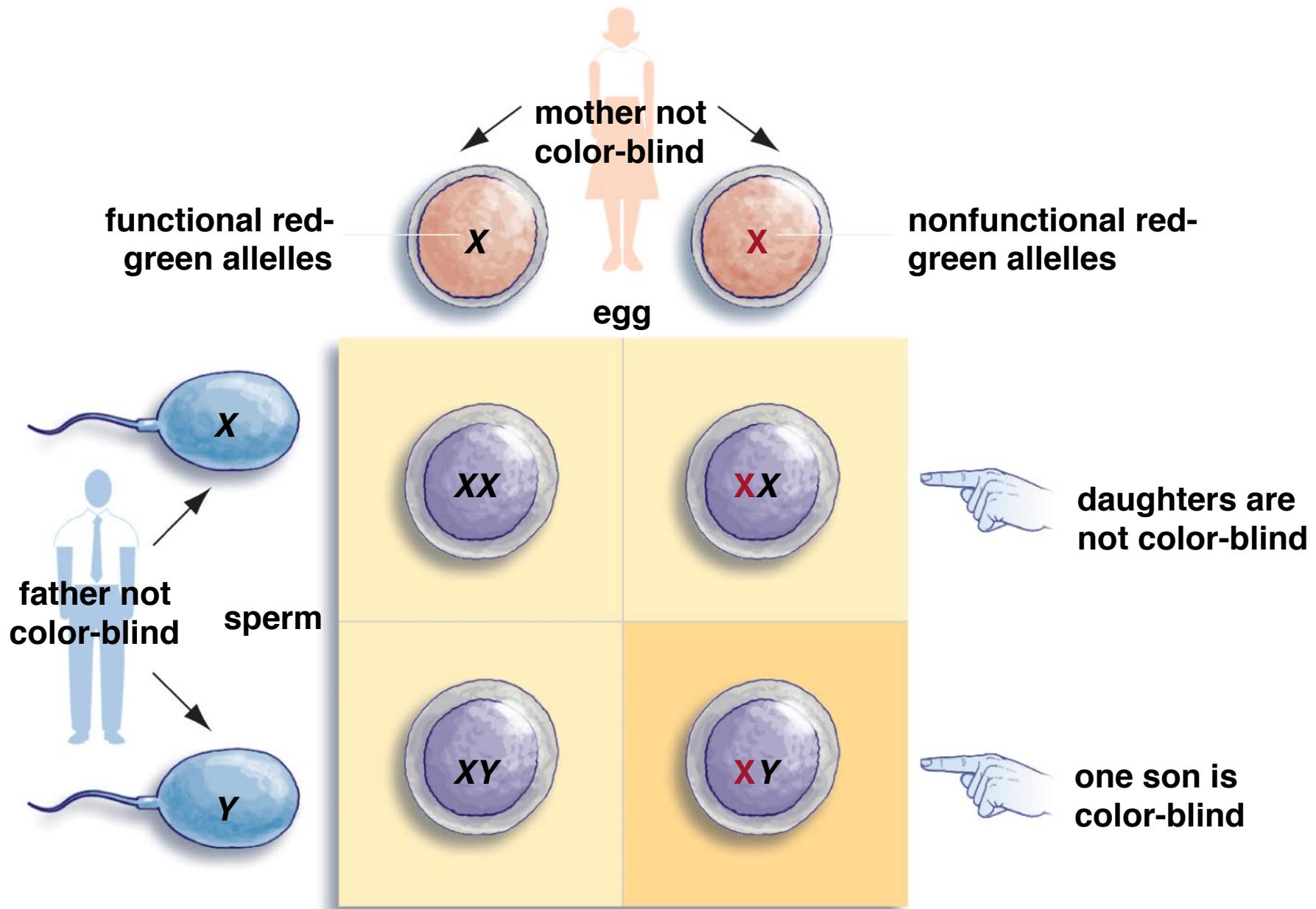


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X-linked Inheritance



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 (YX^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