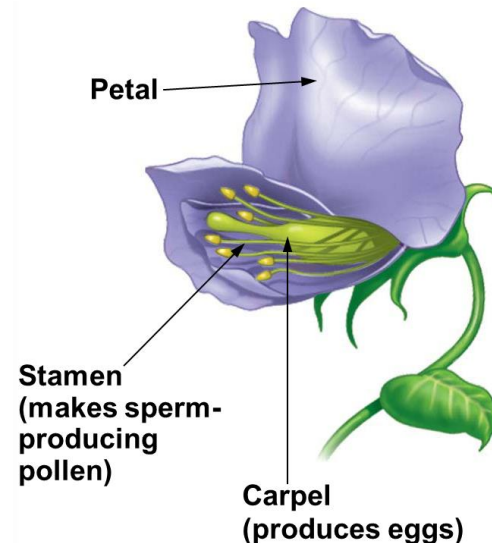


Chapter 5: Chromosomes and Inheritance

Part 2: Genetics

Mendel and Genetics

- Gregor Mendel
 - Began studying genetics in pea plants in the 1860s
- **Genetics** – study of heredity
- **Heredity** – passing of traits from one generation to the next



Pea Plant Genetics

- **Character** – a feature that is heritable
- **Trait** – different versions or variations of a character

Mendel's Studies

7 characters

Each character has two traits

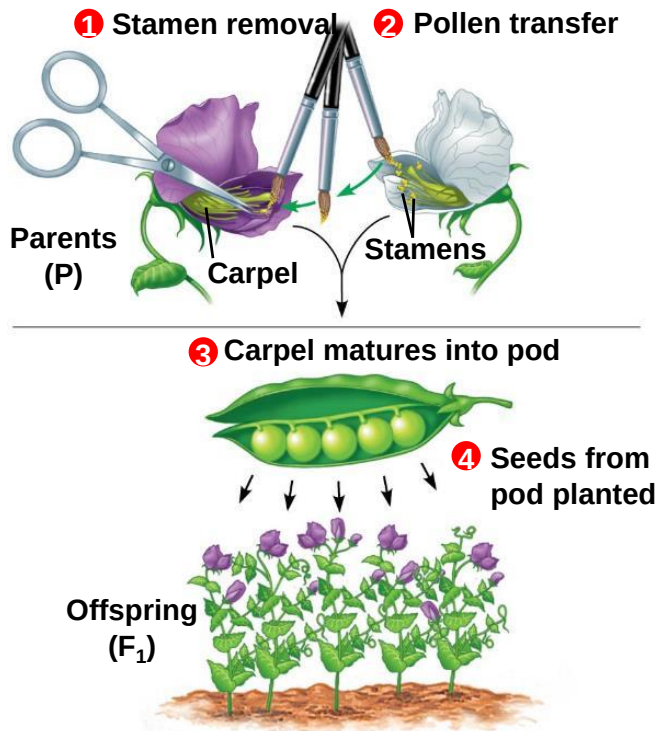
Flower color	 Purple	 White
Flower position	 Axial	 Terminal
Seed color	 Yellow	 Green
Seed shape	 Round	 Wrinkled
Pod shape	 Inflated	 Constricted
Pod color	 Green	 Yellow
Stem length	 Tall	 Dwarf

Dominant Traits and Recessive Traits

Mendel crossed:

Purple flower x White flower
Got all purple flower plants!

Purple flower trait is dominant to white flower trait. (Purple covers up white). The white flower trait is recessive.



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

Dominant Traits and Recessive Traits

- Different letters are used to encode different traits
 - **Capital letters** are used for **dominant traits**
 - **Lowercase letters** are used for **recessive traits**

CHARACTER	PEA FLOWER COLOR	PEA SEED COLOR	LABRADOR COAT COLOR	HUMAN FRECKLES
Dominant trait	<i>P</i> (purple)	<i>Y</i> (yellow)	<i>B</i> (black)	<i>F</i> (freckled)
Recessive trait	<i>p</i> (white)	<i>y</i> (green)	<i>b</i> (chocolate)	<i>f</i> (not freckled)

Mendel's Hypotheses

1. **Alleles** are alternative versions of genes

ex.) Flower color gene is the “P gene”.

Alleles are:

P = purple flowers and
p = white flowers

2. An organism inherits one allele from each parent (two total)

ex.) Plants can have: PP, Pp, or pp

3. Alleles have dominant/recessive relationships

ex.) P (purple) is dominant to p (white)



Purple flower trait



White flower trait

Purple flower plant:
PP or Pp

CHARACTER:
PEA PLANT
FLOWER COLOR

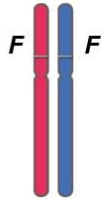
White flower plant:
pp

Genotype and Phenotype

- **Genotype** – genetic makeup of a trait
 - **Homozygous dominant** (PP)
 - **Heterozygous** (Pp)
 - **Homozygous recessive** (pp)
- **Phenotype** – appearance or expression of a trait
 - Purple flowers (from PP or Pp genotypes)
 - White flowers (from pp genotype)

Genotype Determines Phenotype

GENOTYPE



Homozygous dominant
for freckles

A homozygous dominant individual—an individual with two copies of the dominant allele—will express the dominant trait.

PHENOTYPE



Freckled trait

HUMAN FRECKLES

F (freckled)

f (not freckled)

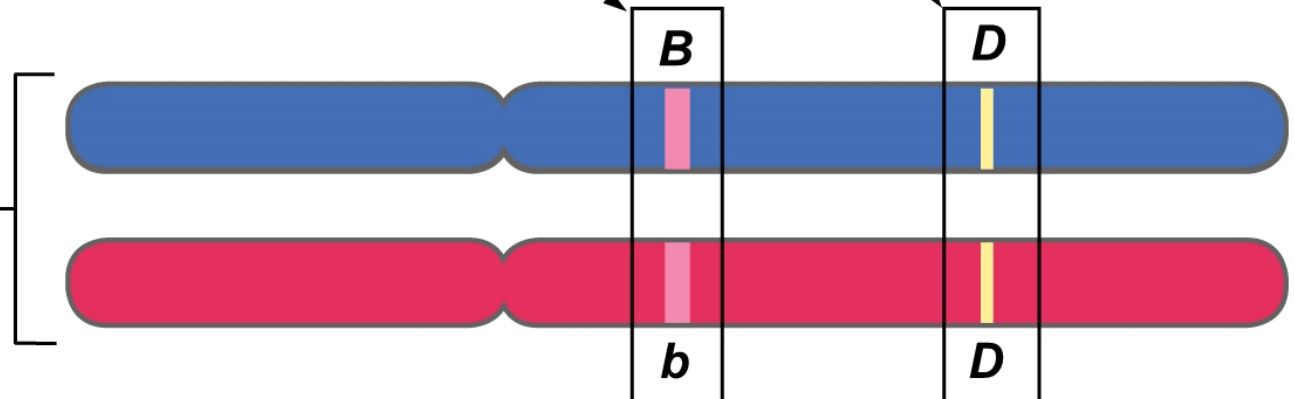
Alleles are Inherited on Chromosomes

Homologous chromosomes contain the same genes in the same order but can have different alleles

Heterozygous
(two different
alleles) for this
gene

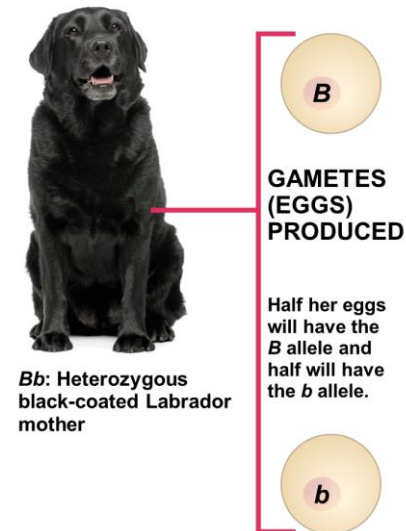
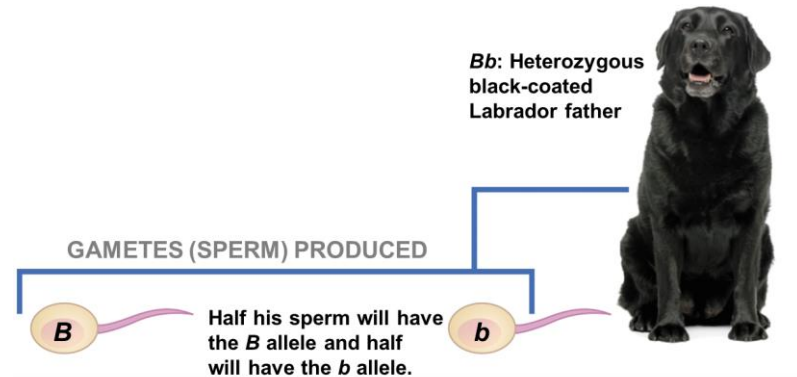
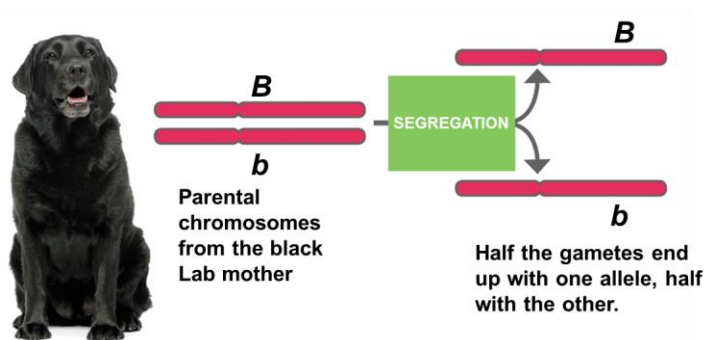
Homozygous
(two identical
alleles) for
this gene

Matched set of
chromosomes,
one derived
from the father
(blue) and one
derived from the
mother (red)



The Law of Segregation

- Each sperm or egg cell contains one allele for each gene
- Fusion of gametes (when sperm and egg come together) creates a diploid cell with two alleles



Punnett Square

- Shows all the possible gamete combinations from a mating

KEY
B = black allele (dominant)
b = chocolate allele (recessive)



Bb: Heterozygous black-coated Labrador mother



GAMETES (SPERM) PRODUCED

Half his sperm will have the *B* allele and half will have the *b* allele.

B

b



Bb: Heterozygous black-coated Labrador father

Offspring made from fertilization of *B* egg and *B* sperm



***BB* (BLACK)**

Offspring made from fertilization of *B* egg and *b* sperm



***Bb* (BLACK)**

Offspring made from fertilization of *B* egg and *b* sperm



***Bb* (BLACK)**

Offspring made from fertilization of *b* egg and *b* sperm



***bb* (CHOCOLATE)**

RESULTS = 1 *BB* (BLACK) : 2 *Bb* (Black) : 1 *bb* (CHOCOLATE)

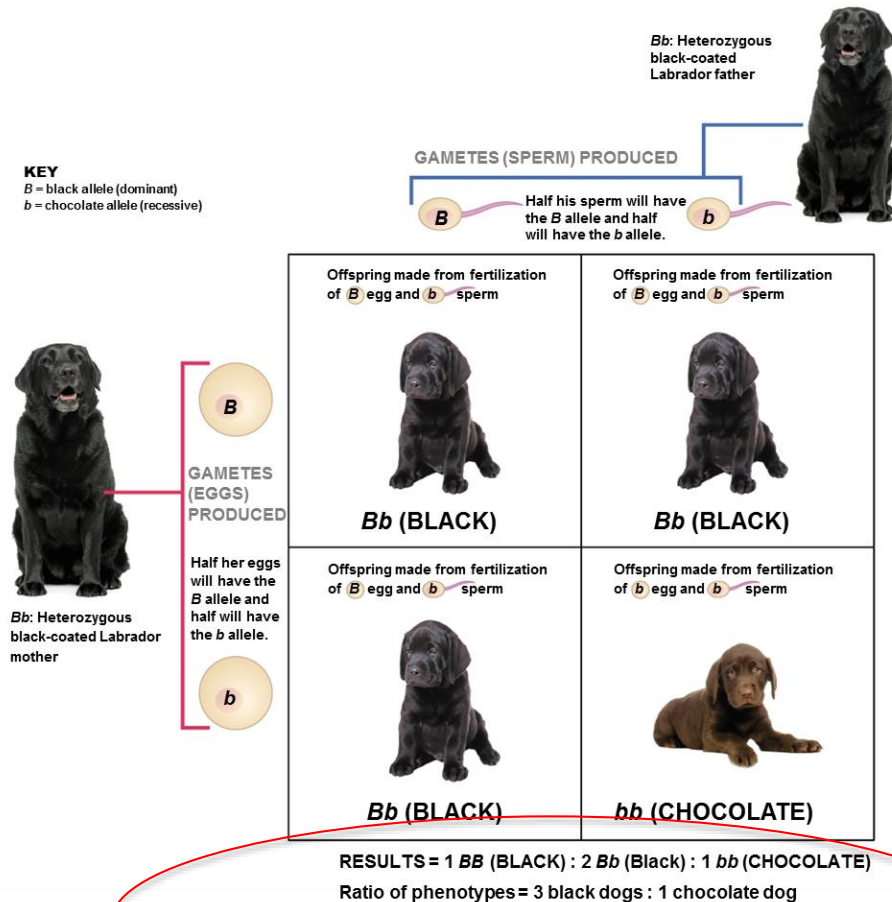
Ratio of phenotypes = 3 black dogs : 1 chocolate dog

1. Write the possible alleles in gametes from dad

3. Fill in the boxes of the Punnett square to determine the possible combinations of alleles in the offspring

Genotypic and Phenotypic Ratios

- What are the number of each possible offspring inside the Punnett Square?
- Genotypic ratio (the possible allele combinations)
 - Ex. BB, Bb, or bb
- Phenotypic ratio (the possible phenotypes/physical appearances)
 - Ex. Black or chocolate



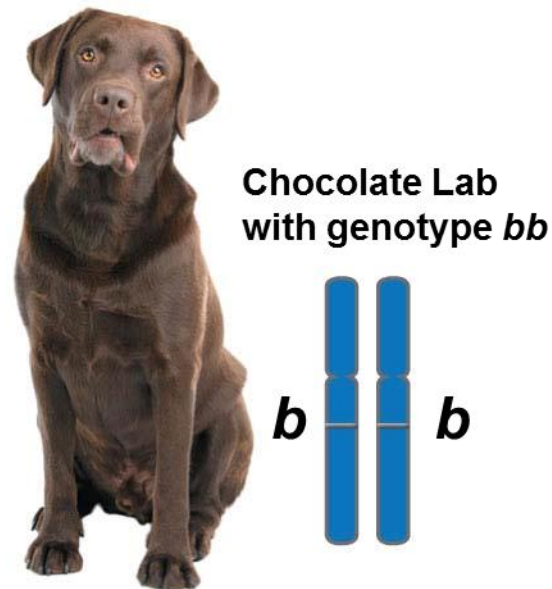
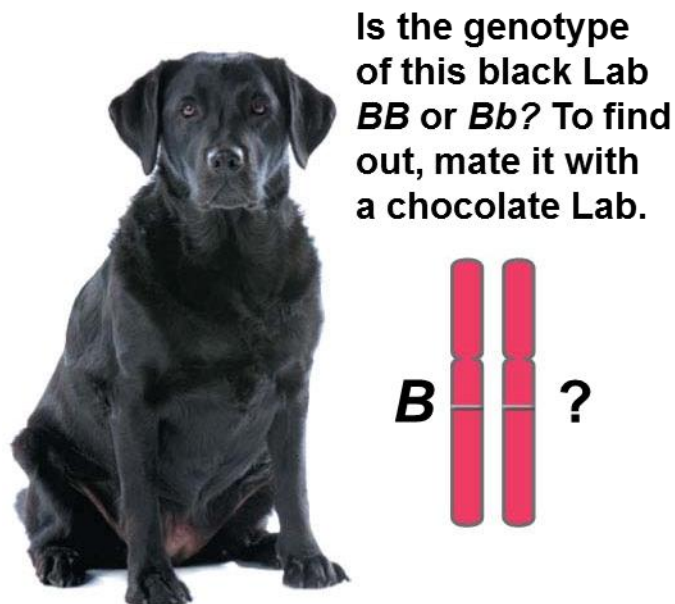
Punnett Square Example

- A pea plant heterozygous for purple flowers is crossed with a pea plant homozygous for purple flowers. Draw a Punnett Square to represent this cross:

- What is the genotypic ratio from this cross?
- What is the phenotypic ratio from this cross?

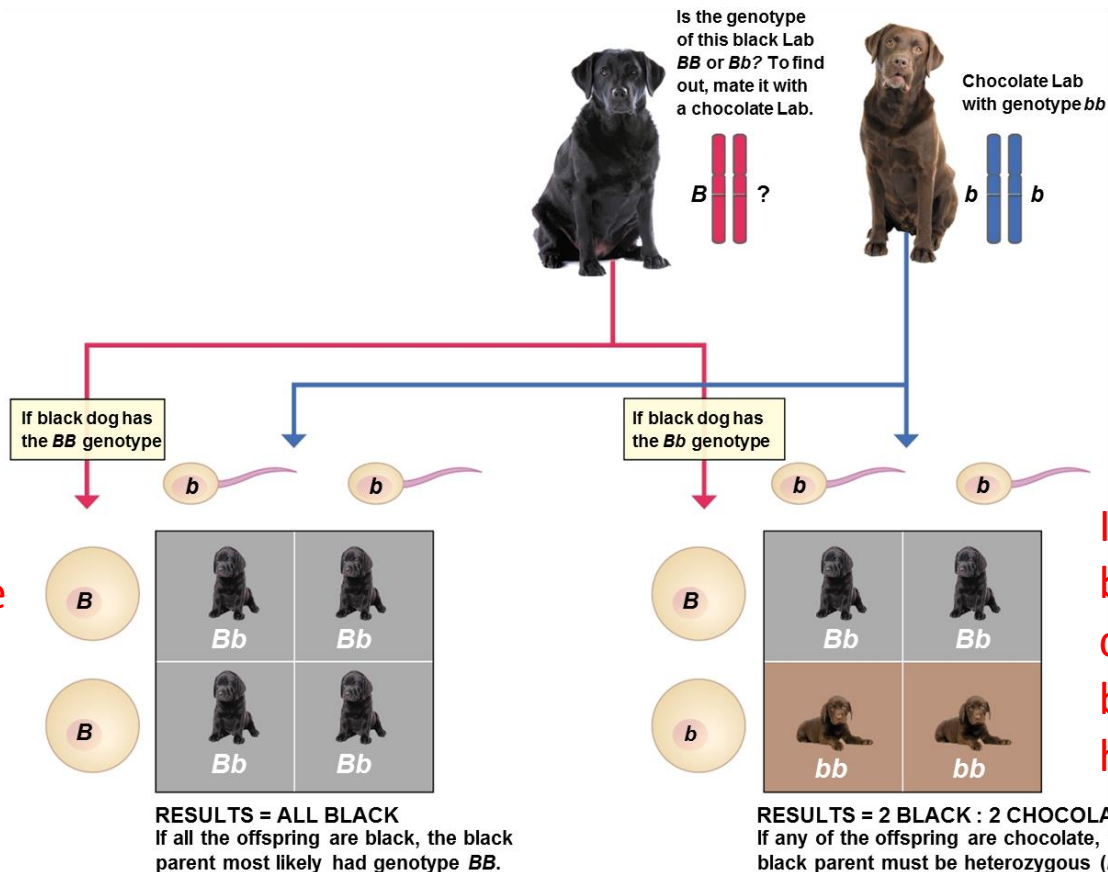
Testcross: Determine an Unknown Genotype

- What is the genotype of a black lab?
 - Mate with chocolate lab (recessive phenotype)



Testcross: Determine an Unknown Genotype

- What is the genotype of a black lab?



Testcross: Determine an Unknown Genotype

- What is the genotype of a white flower pea plant?

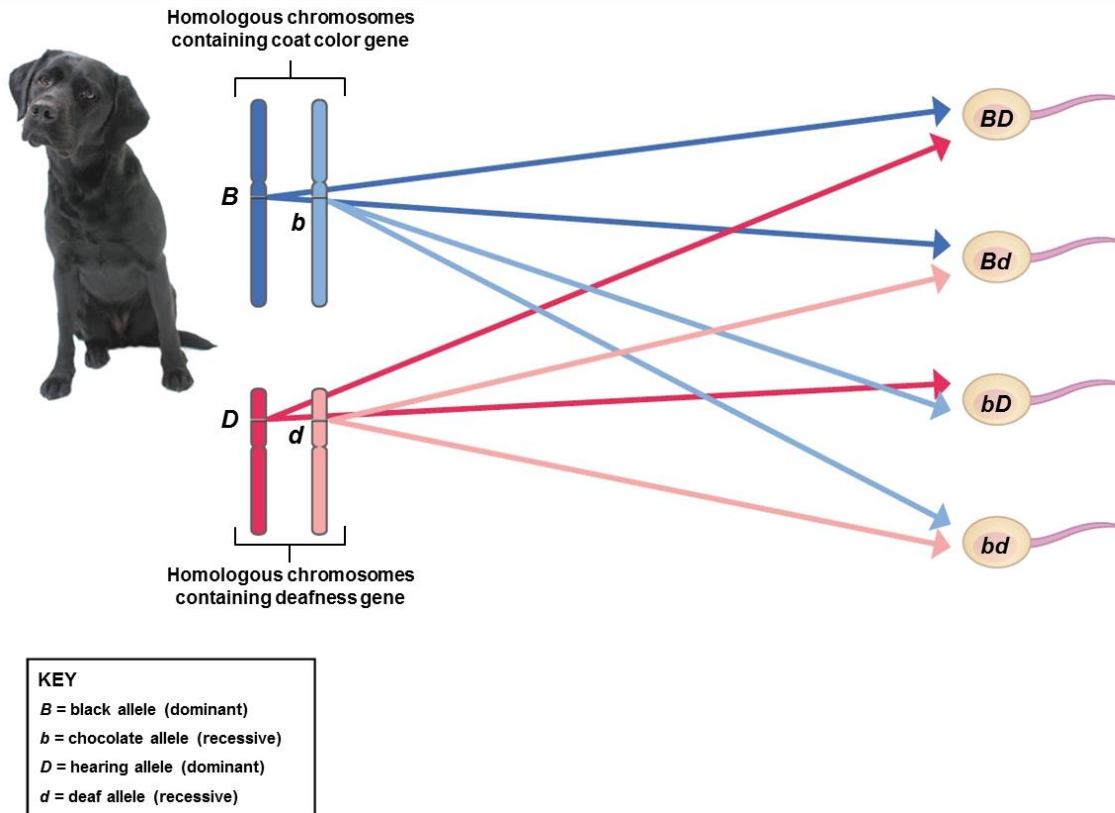


- What is the genotype of a purple flower pea plant?



- A **testcross** helps determine the genotype of the individual displaying the dominant phenotype
 - Unknown purple flower plant x White flower plant
 - Practice: draw Punnett squares for this testcross

The Law of Independent Assortment

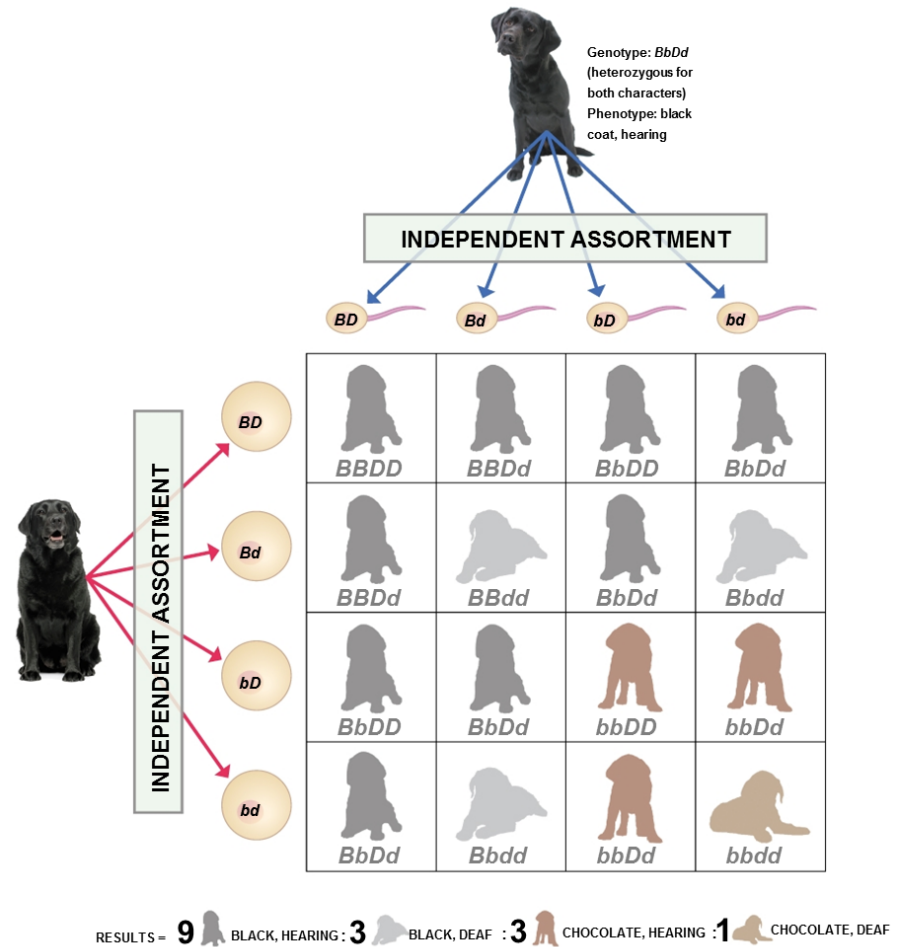


2 genes: coat color and deafness
Alleles for each gene are inherited independently

- When examining 2 genes, the inheritance of one allele has no effect on the inheritance of another
- Each gamete has an equal chance of containing any combination of alleles

Dihybrid Cross

- **Monohybrid cross** – each individual is heterozygous for one gene
- **Dihybrid cross** – each individual is heterozygous for two genes
 - Phenotypic ratio is 9:3:3:1



Human Genetics

- **Wild-type trait** = most common, one most often seen in nature
- **Mutant trait** = less common

DOMINANT	RECESSIVE
Freckles	No freckles
Normal pigmentation	Albinism
Healthy	Cystic fibrosis
Achondroplasia (dwarfism)	Normal height

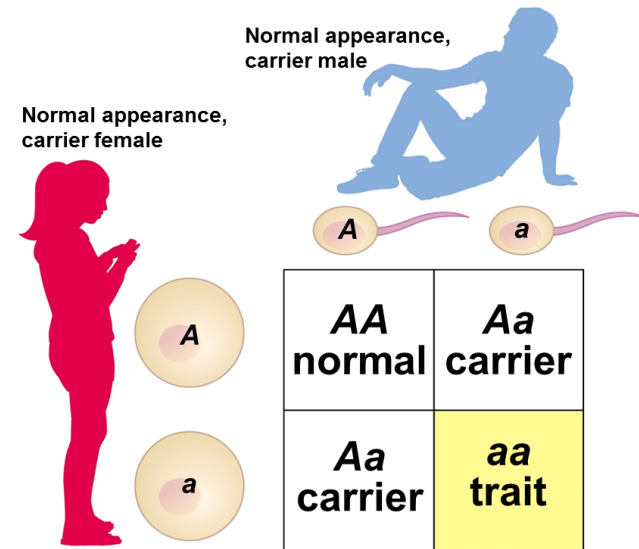
☒ Mutant trait (less common)

☐ Wild-type trait (more common)

Wild-type traits may be dominant or recessive!
Mutant traits are not always recessive!

Human Genetics: Carriers

- Humans are diploid (have two alleles for each gene)
- For **recessive diseases**:
 - Individual must have two copies of the recessive allele to express the trait (aa)
 - Normal individuals without the disease may be either homozygous dominant (AA) or heterozygous (Aa)
 - A normal individual who is heterozygous (Aa) can pass along the disease allele (a) so is considered a **carrier** for the trait

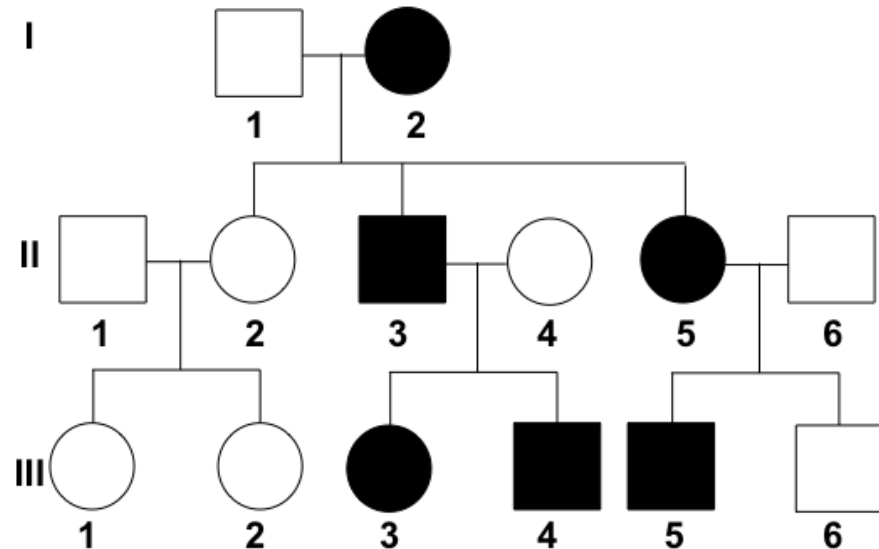
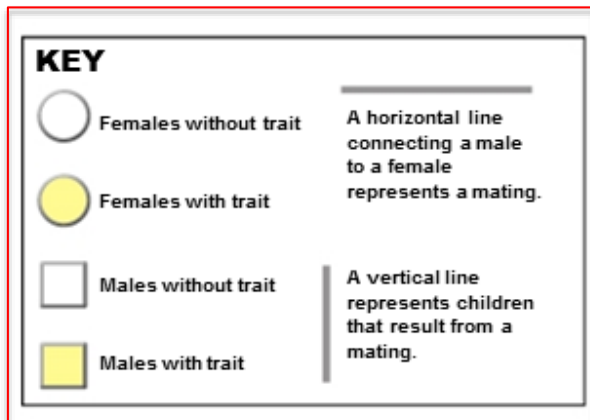


RESULTS = 1 AA (NORMAL) : 2 Aa (CARRIER) : 1 aa (TRAIT)
Ratio of phenotypes = 3 normal : 1 trait

Two normal individuals can produce a child with the recessive disease if they are both carriers

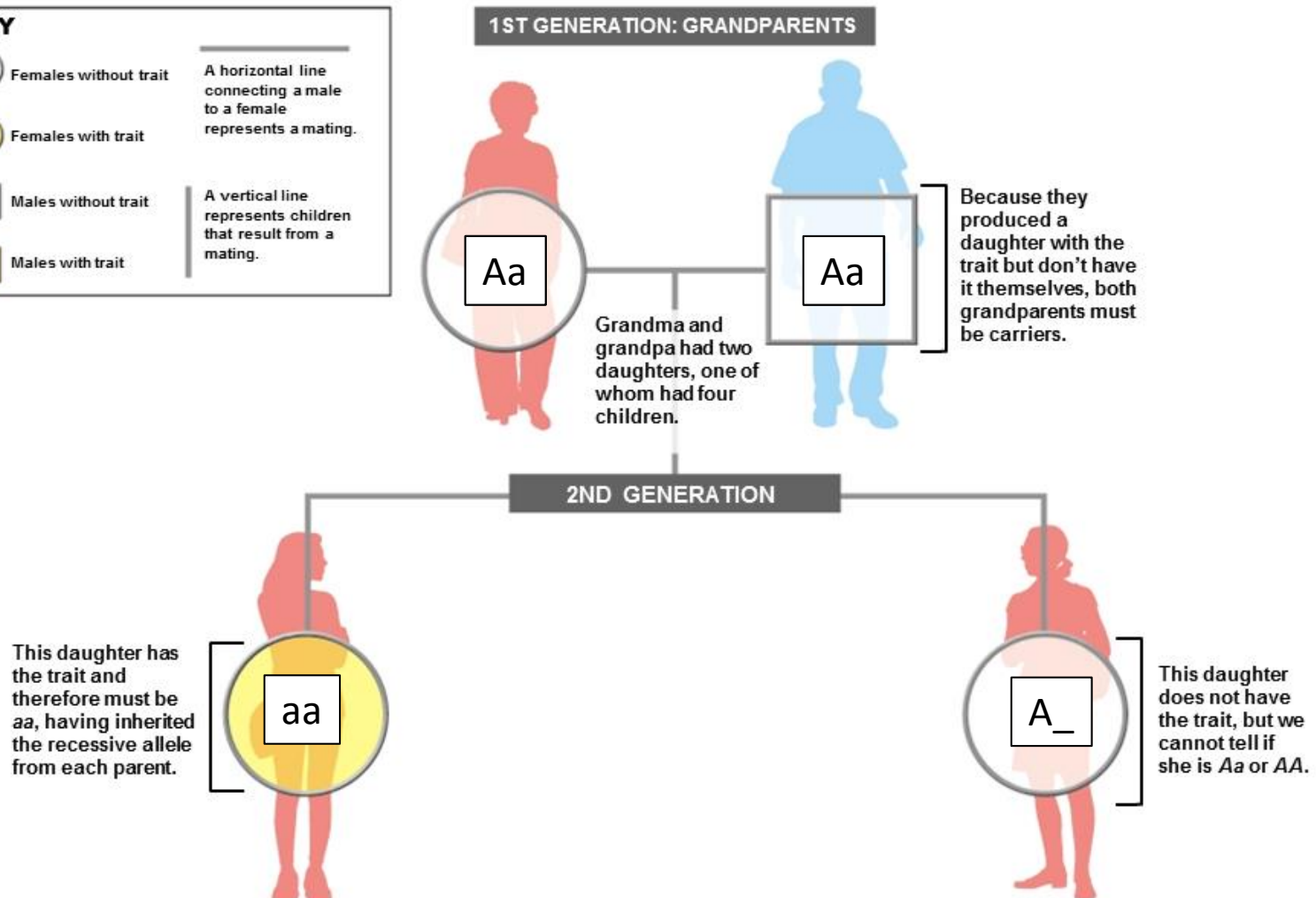
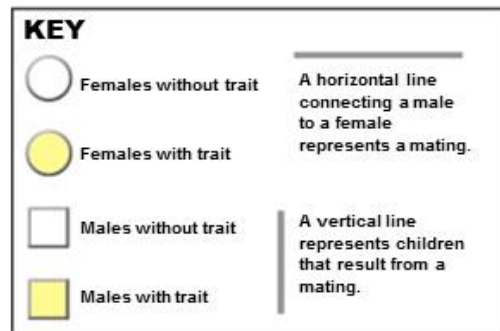
Human Genetics: Pedigrees

- **Pedigree** – a genetic family history



1. Describe individual I-2:
2. Describe individual III-6:
3. What is the relationship between individuals I-2 and II-3?
4. What is the relationship between individuals II-3 and II-4?

Human Genetics: Pedigree Problem Solving



Other Inheritance Patterns: Multiple Alleles

- **Multiple alleles:** more than two alleles possible
 - Each individual is still diploid (has 2 alleles)

Human blood type alleles:

I^A = A allele

I^B = B allele

i = O allele

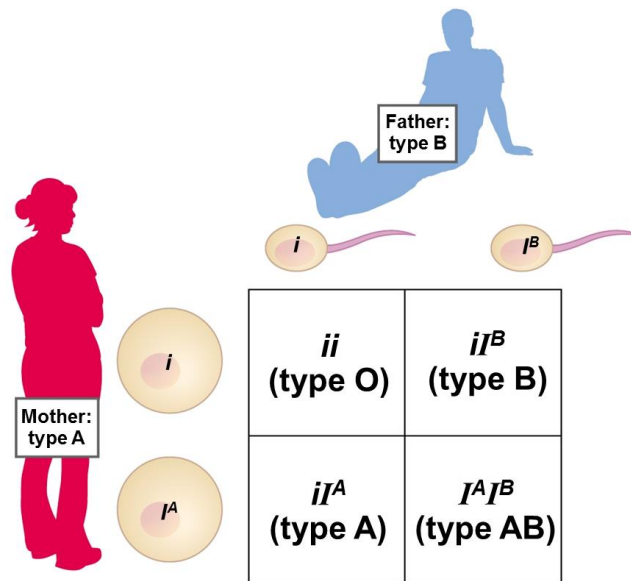
GENOTYPE	PHENOTYPE (blood type)	Blood Group (Phenotype)	Genotypes	Carbohydrates Present on Red Blood Cells
ii	O	A	$I^A I^A$ or $I^A i$	 Carbohydrate A
$i I^A$	A	B	$I^B I^B$ or $I^B i$	 Carbohydrate B
$I^A I^A$	A	AB	$I^A I^B$	 Carbohydrate A and Carbohydrate B
$i I^B$	B	O	ii	 Neither
$I^B I^B$	B			
$I^A I^B$	AB			

A and B alleles are dominant to O

A and B alleles are codominant to each other

Other Inheritance Patterns: Multiple Alleles

- If a man heterozygous for Type B blood and a woman heterozygous for Type A blood have a child, what is the probability their child will have Type O blood?

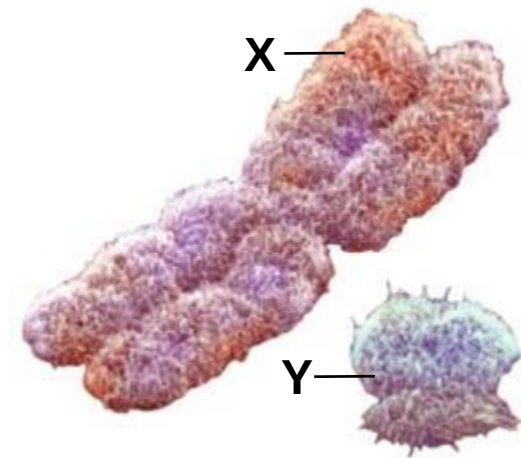


$\frac{1}{4}$ or 25% probability of having a child with Type O blood

RESULTS = 1 ii (TYPE O) : 1 iI^B (TYPE B) : 1 iI^A (TYPE A) : 1 $I^A I^B$ (TYPE AB)

Sex Chromosomes

- Most genes are on homologous chromosomes (autosomes)
 - Humans have 22 pairs
- Last set of homologous chromosomes isn't really homologous (sex chromosomes)
 - Human 23rd pair
 - Y chromosome is smaller than X

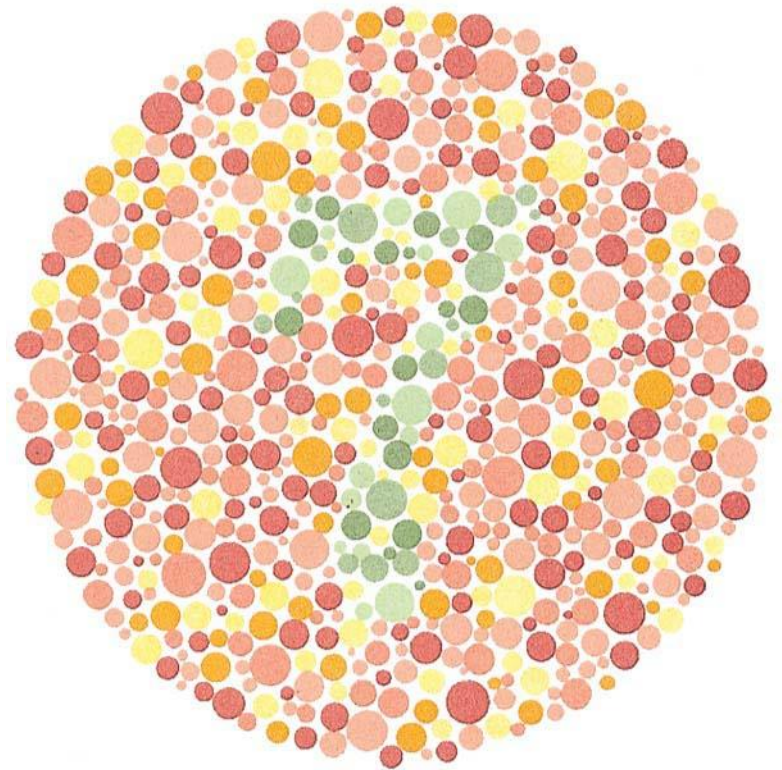


Sex-Linked Genes

- **Sex-linked genes** – genes located on the X sex chromosome
- Inherited differently by males and females
- Females
 - Inherit two X chromosomes (XX)
 - Have two alleles for each gene on the X chromosome
- Males
 - Inherit one X and one Y chromosome (XY)
 - Have only one allele for each gene on the X chromosome

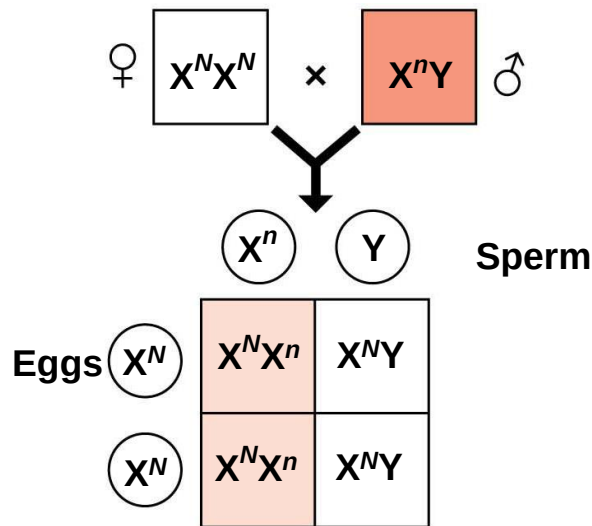
Sex-Linked Gene: Colorblindness

- Sex-linked gene alleles are written as superscripts
 - X^N = normal vision
 - X^n = colorblind
 - Y (no allele)
- Possible genotypes
 - $X^N X^N$ = female, normal
 - $X^N X^n$ = female, normal
 - $X^n X^n$ = female, colorblind
 - $X^N Y$ = male, normal
 - $X^n Y$ = male, colorblind

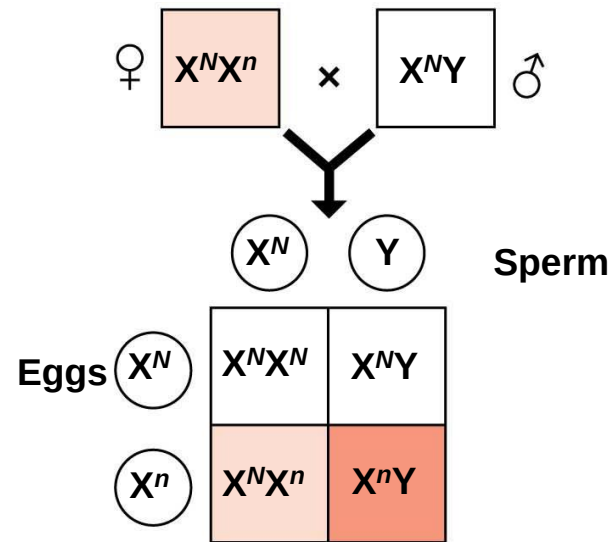


Sex-Linked Gene: Colorblindness

- Examine the Punnett Squares...
 - How many offspring are colorblind?
 - How many female offspring are colorblind?
 - How many male offspring are colorblind?



(a) Normal female $\times$ colorblind male



(b) Carrier female $\times$ normal male

You should now be able to:

- Define the terms

Gene	Allele	Genotype	Phenotype
Homozygous	Heterozygous	Dominant	Recessive
Wild-type	Mutant	Carrier	Sex-linked gene

- Understand the relationship between genotype and phenotype
- Describe how genes and alleles relate to homologous chromosomes
- Explain the Law of Segregation and Law of Independent Assortment

You should now be able to:

- Identify and solve problems of typical genetic crosses using Punnett squares
 - Monohybrid cross
 - Testcross
 - Dihybrid cross
 - Human genetic traits (carriers)
 - Human blood types
 - Sex-linked traits
- Solve problems involving human pedigrees