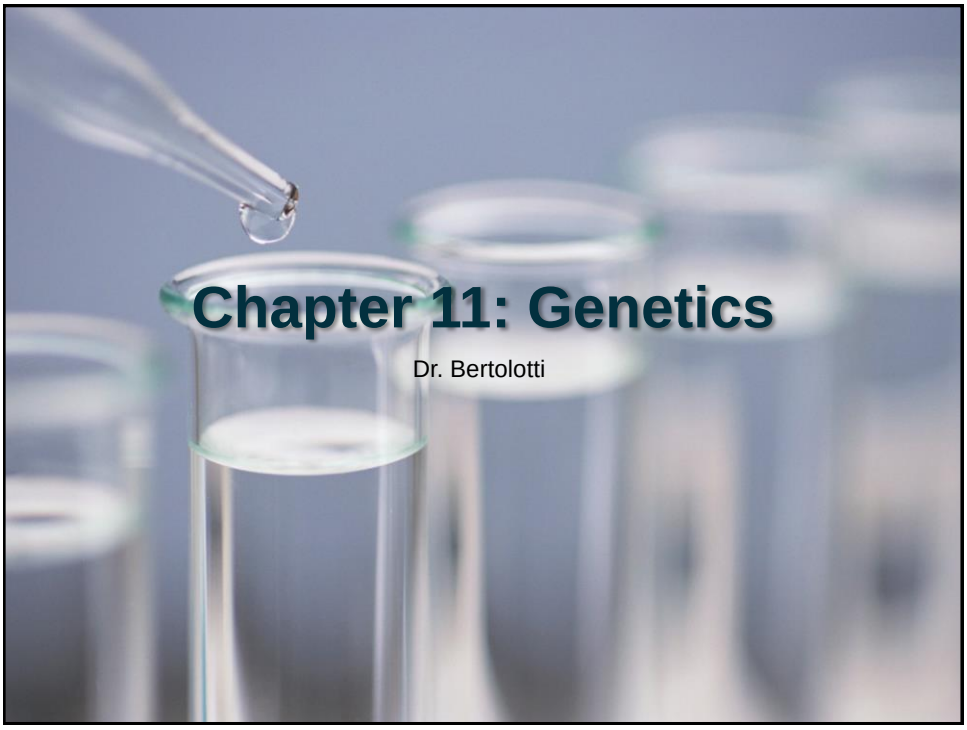


Chapter 11: Genetics

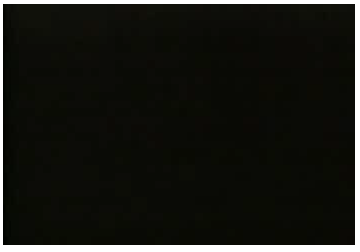
Dr. Bertolotti

Essential Question

- **How does biological information pass from one generation to another?**

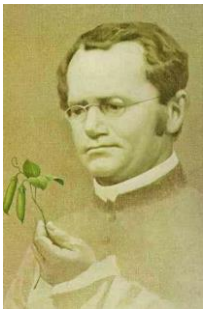
How are different forms of a gene distributed to offspring?

- ***Genetics*** is the scientific study of hereditary

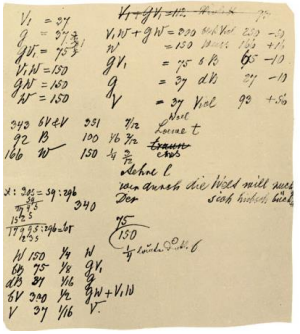


Genetics & The Work of Mendel

Gregor Mendel



- Modern genetics began in the mid-1800s in an abbey garden, where a monk named Gregor Mendel documented inheritance in peas
  - used good experimental design
  - used mathematical analysis
    - collected data & counted them
  - excellent example of scientific method

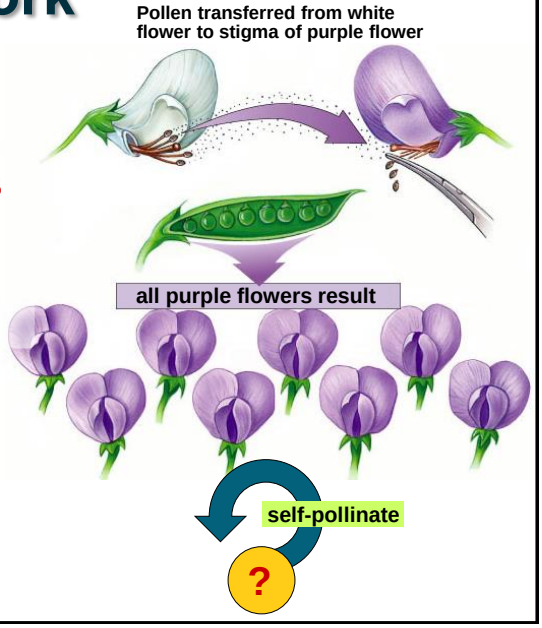


Vocabulary

- **Fertilization** is the process in sexual reproduction in which male and female reproductive cells join to form a new cell
- **True breeding** is a term used to describe organisms that produce offspring identical to themselves if allowed to self-pollinate
- **Self pollinating** means that sperm cells in pollen fertilize the egg cells in the same flower

Mendel's work

- Bred pea plants
  - cross-pollinate true breeding parents
  - raised seed & then observed traits
  - allowed offspring to self-pollinate & observed next generation

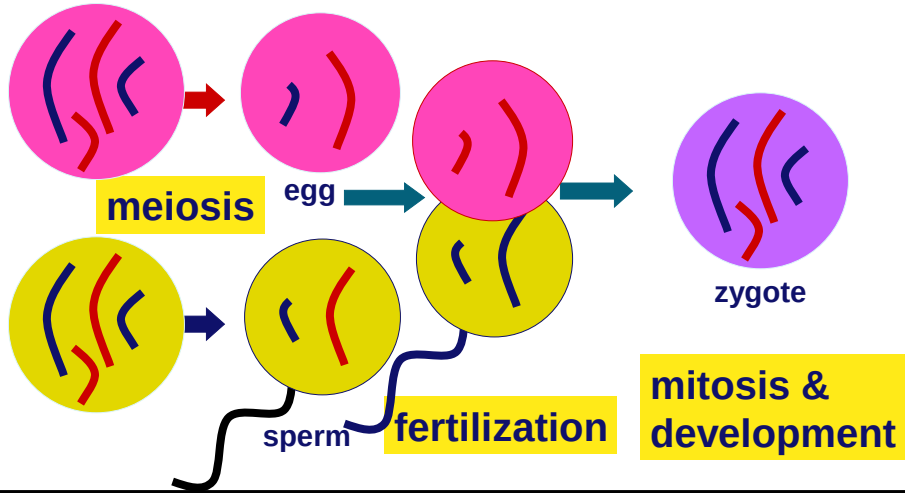


Genes and Dominance

- A **trait** is a very specific characteristic that varies from one individual to another
  - ex. Height, hair color, etc.
  - A trait is coded for by proteins
- The offspring of crosses between parents with different traits are called **hybrids**
  - ex. The child of one parent with curly hair and another with straight hair is a hybrid
- The chemical factors that determine traits are called **genes**
  - Different forms of genes are called **alleles**

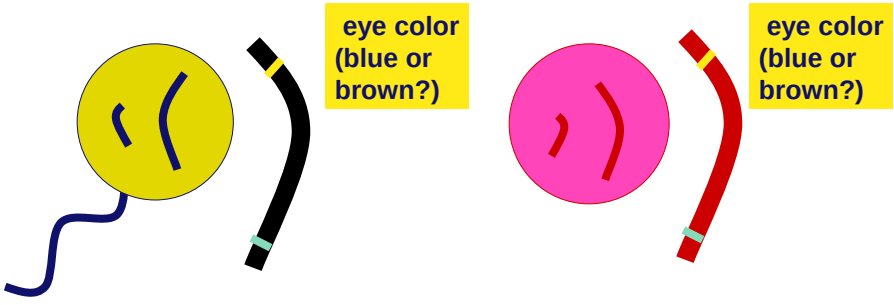
Inheritance of chromosomes

- Egg + sperm → zygote



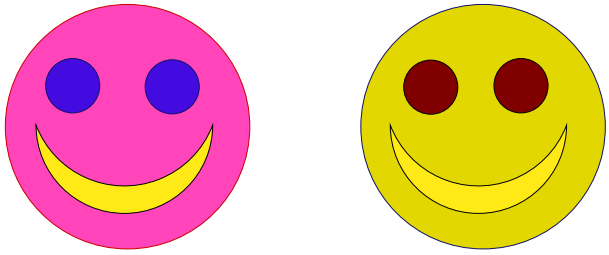
Inheritance of genes

- On the chromosomes passed from Mom & Dad to offspring are genes
  - may be same information
  - may be different information

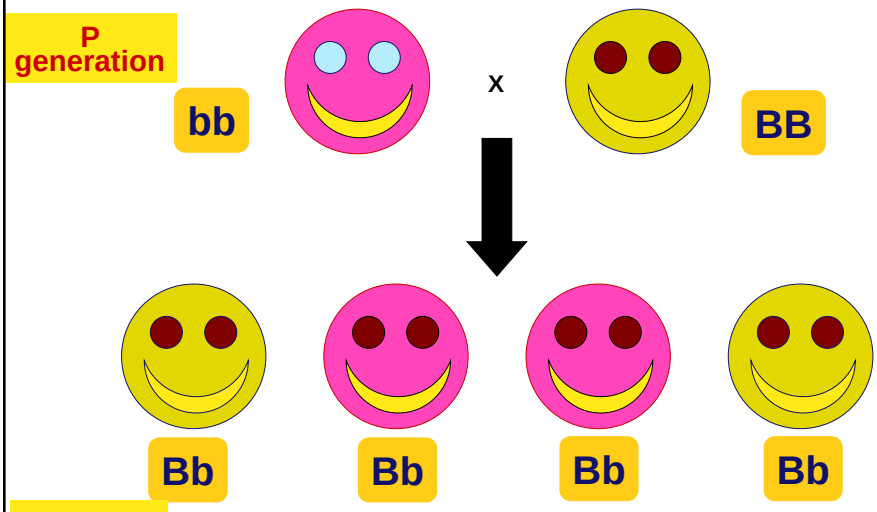


Effect of genes

- Genes come in different versions
  - brown vs. blue eyes
  - brown vs. blonde hair
  - **alleles**

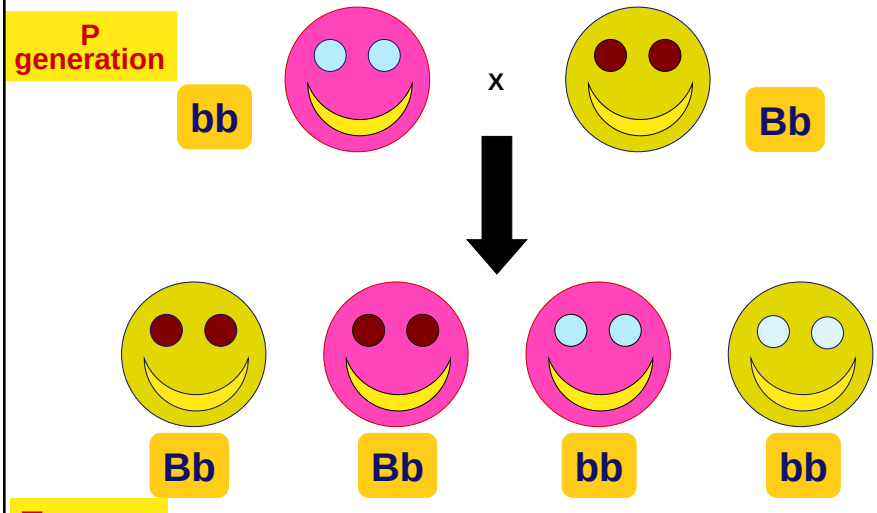


Genes affect what you look like



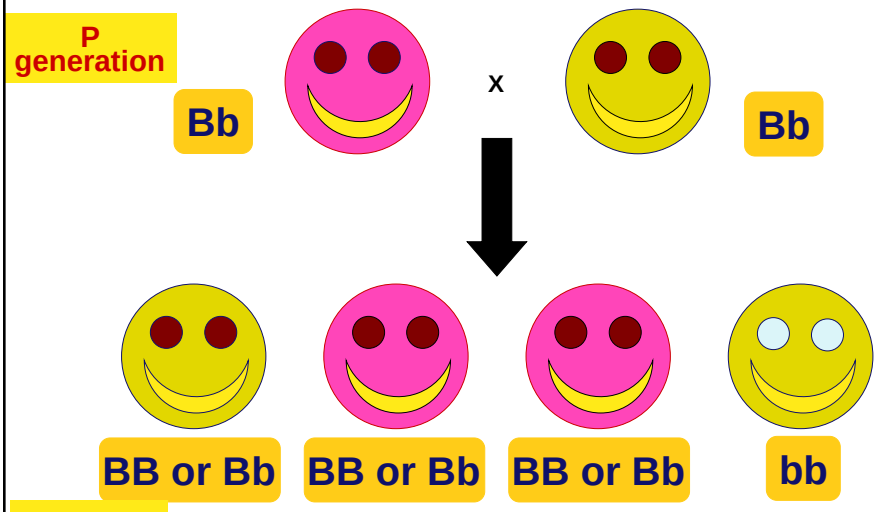
Where did the blue eyes go??

Genes affect what you look like…



Why did the blue eyes stay??

Genes affect what you look like…



Where did the blue eyes come from??

What did we show here?

- Genes come in "versions"
  - brown vs. blue eye color
  - alleles
- Alleles are inherited separately from each parent
  - brown & blue eye colors are separate & do not blend
    - either have brown or blue eyes, not a blend
- Some alleles mask others
  - brown eye color masked blue

Mendel collected data for 7 pea traits

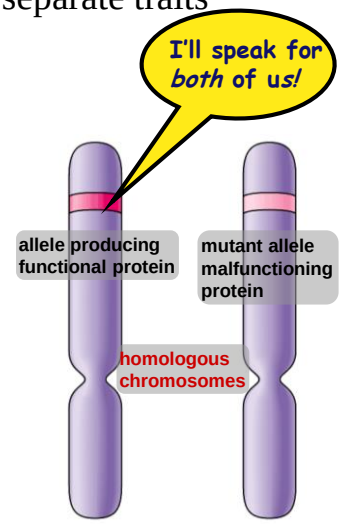
Table 13.1 Seven Characters Mendel Studied and His Experimental Results

Character				F_2 Generation	
	Dominant Form	×	Recessive Form	Dominant:Recessive	Ratio
	Purple flowers	×	White flowers	705:224	3.15:1
	Yellow seeds	×	Green seeds	6022:2001	3.01:1
	Round seeds	×	Wrinkled seeds	5474:1850	2.96:1
	Green pods	×	Yellow pods	428:152	2.82:1
	Inflated pods	×	Constricted pods	882:299	2.95:1
	Axial flowers	×	Terminal flowers	651:207	3.14:1
	Tall plants	×	Dwarf plants	787:277	2.84:1

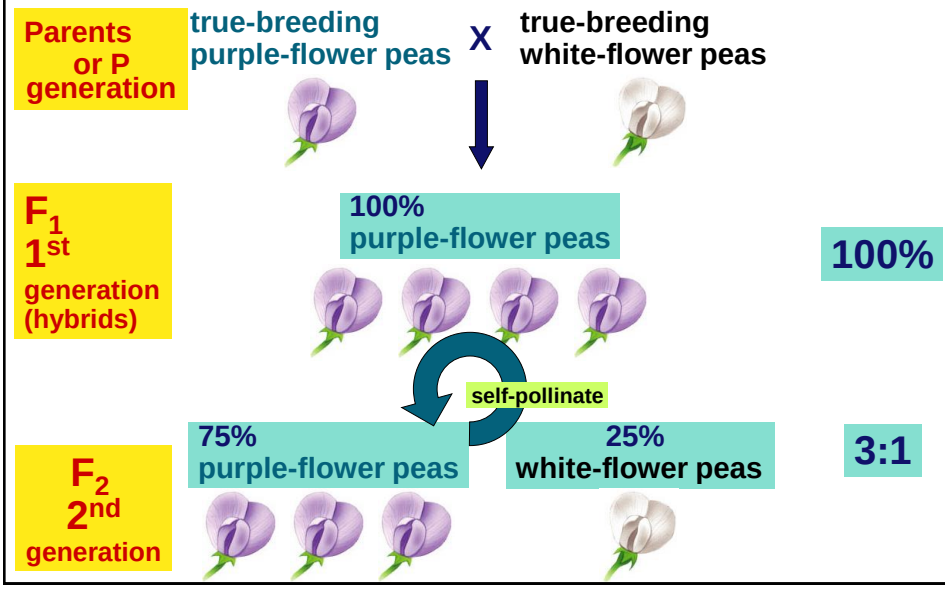
- Each original pair of organisms is called the parental generation or P
  - The offspring of parents are called the first filial or F_1 generation
- ***The principle of dominance states that some alleles are dominant and others are recessive***
  - An organism with a dominant allele for a particular form of a trait will **ALWAYS** exhibit that form of the trait
  - An organism with a recessive allele for a particular form of a trait will exhibit that form only when the dominant allele for the trait is **absent**
  - **AA (DOMINANT) Aa aa (recessive)**

What did Mendel's findings mean?

- Some traits mask others
  - purple & white flower colors are separate traits that do not blend
    - purple x white ≠ light purple
    - purple masked white
  - dominant allele
    - functional protein
      - affects characteristic
    - masks other alleles
  - recessive allele
    - no noticeable effect
    - allele makes a non-functioning protein

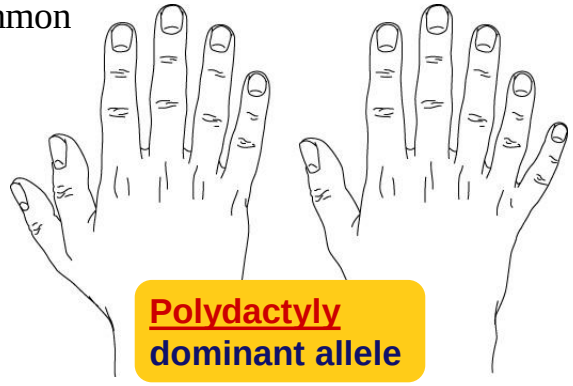


Looking closer at Mendel's work



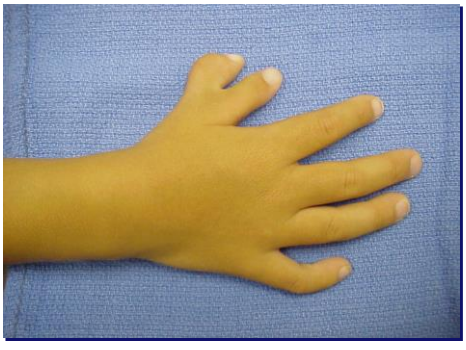
Dominant ≠ most common allele

- Because an allele is dominant does not mean…
  - it is better, or
  - it is more common



Polydactyly
dominant allele

Polydactyly



individuals are born with extra fingers or toes

the allele for >5 fingers/toes is DOMINANT & the allele for 5 digits is recessive

recessive allele far more common than dominant

→ **only 1 individual out of 500 has more than 5 fingers/toes**

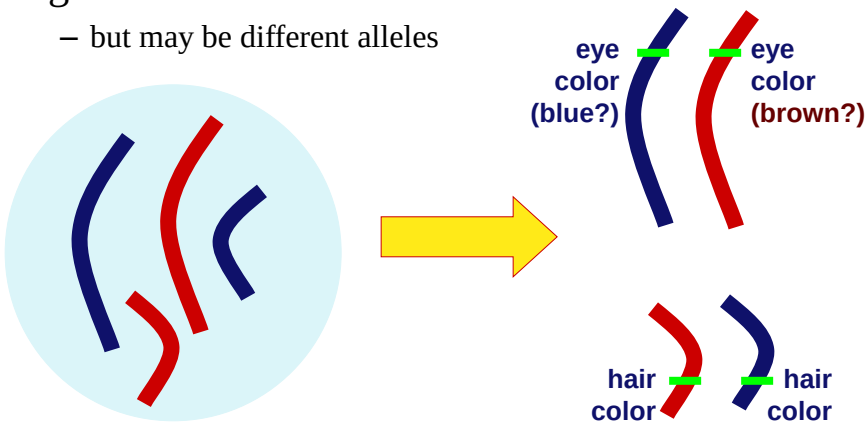
→ **so 499 out of 500 people are homozygous recessive (aa)**

Segregation

- **Gametes** are sex cells
- **Segregation** is the separation of alleles during gamete formation
- Segregation explains why when the offspring of an F_1 generation is crossed with another offspring of the F_1 generation (thus producing F_2 generation), 1 out of 4 of the offspring in the F_2 generation had the recessive allele

How does this work?

- Paired chromosomes have same kind of genes
  - but may be different alleles



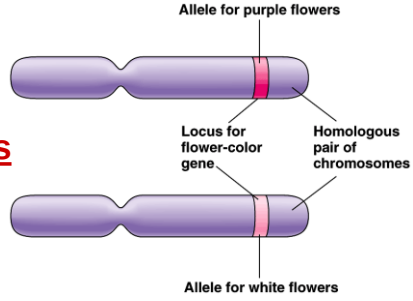
Traits are inherited as separate units

- For each trait, an organism inherits 2 copies of a gene, 1 from each parent
  - a diploid organism inherits 1 set of chromosomes from each parent
    - diploid = 2 sets of chromosomes

1 from Mom

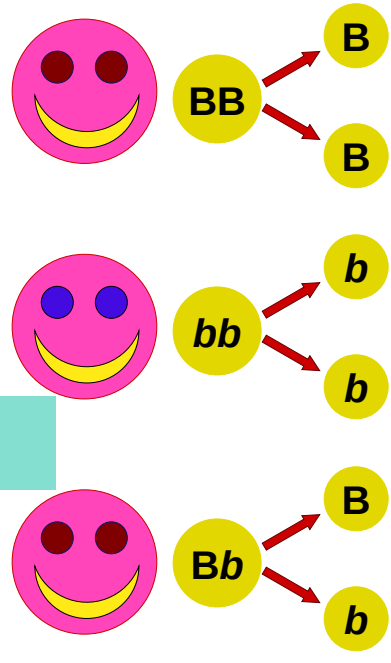
homologous chromosomes

1 from Dad



Making gametes

BB = brown eyes
***bb* = blues eyes**
B*b* = brown eyes



→ brown is dominant over blue
→ blue is recessive to brown

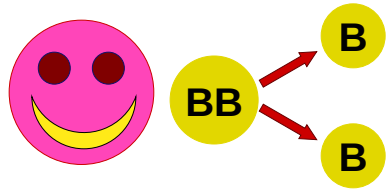
Remember meiosis!

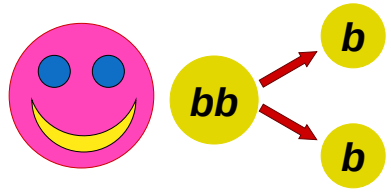
How do we say it?

2 of the same
Homozygous

BB = brown eyes
bb = blues eyes

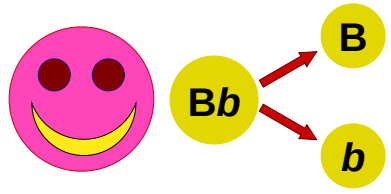
homozygous dominant
homozygous recessive





2 different
Heterozygous

B*b* = brown eyes



QUESTION AND ANSWER

How are different forms of a gene distributed to offspring?

How can we use probability to predict traits?

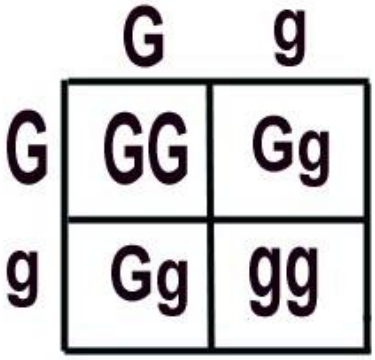
- Organisms that have 2 identical alleles for a particular trait are said to be **homozygous**, ex. *TT* or *tt*
  - Homozygous organisms are true breeding for a particular trait
- Organisms that have 2 different alleles for the same trait are **heterozygous,** ex *Tt*
  - Heterozygous organisms are hybrid for a particular trait
- **Phenotype** is physical characteristics or the "outward, physical manifestation" of the organism ex. Tall plants
- **Genotype** is genetic makeup or the "internally coded, inheritable information" carried by all living organisms, ex. *TT*

Probability and Punnett Squares

- **Probability** refers to the likelihood that a particular event will occur
- Past outcomes do not affect the future ones
- ***The principles of probability can be used to predict the outcomes of genetic crosses***
- E.g., What is the probability that a single coin flip will come up heads?
  - a. 100 percent b. 75 percent
  - c. 50 percent d. 25 percent

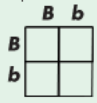
Punnett Squares

- A **Punnett Square** is a diagram showing the gene combinations that might result from a genetic cross

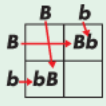


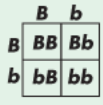
How to make a Punnett Square

Draw a table with enough squares for each pair of gametes from each parent. In this case, each parent can make two different types of gametes, B and b. Enter the genotypes of the gametes produced by both parents on the top and left sides of the table.



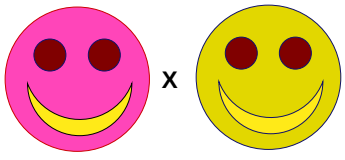
Fill in the table by combining the gametes' genotypes.





Punnett squares

Bb x Bb



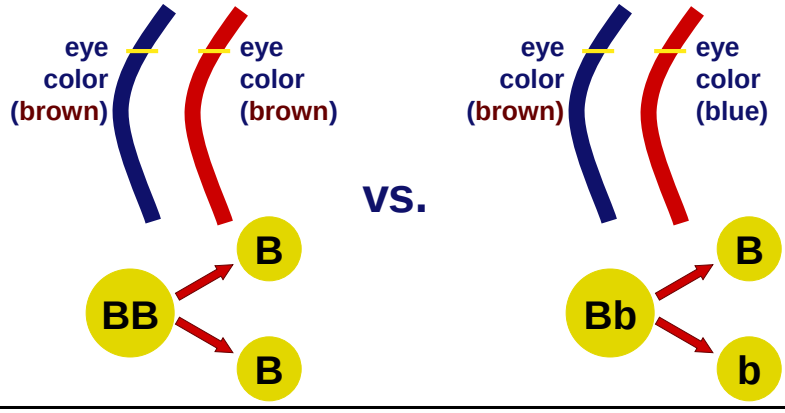
x

male / sperm

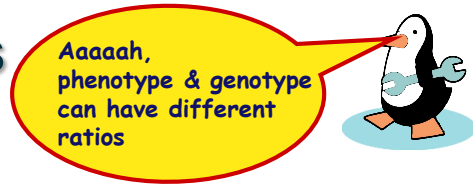
female / eggs	B	b
B	BB	B*b*
b	B*b*	*bb*

Genetics vs. appearance

How were these brown eyes made?



Punnett squares



1st generation (hybrids)

Pp x Pp

male / sperm

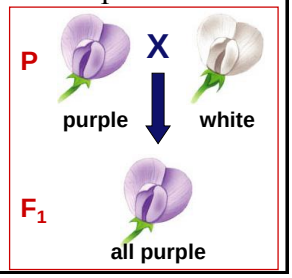
female / eggs	P	p
P	PP	Pp
p	Pp	pp

	% genotype	% phenotype
PP	25%	75%
Pp	50%	
Pp		
pp	25%	25%
	1:2:1	3:1

Genotype vs. phenotype

- Difference between how an organism "looks" & its genetics
  - phenotype
    - description of an organism's trait
  - genotype
    - description of an organism's genetic makeup

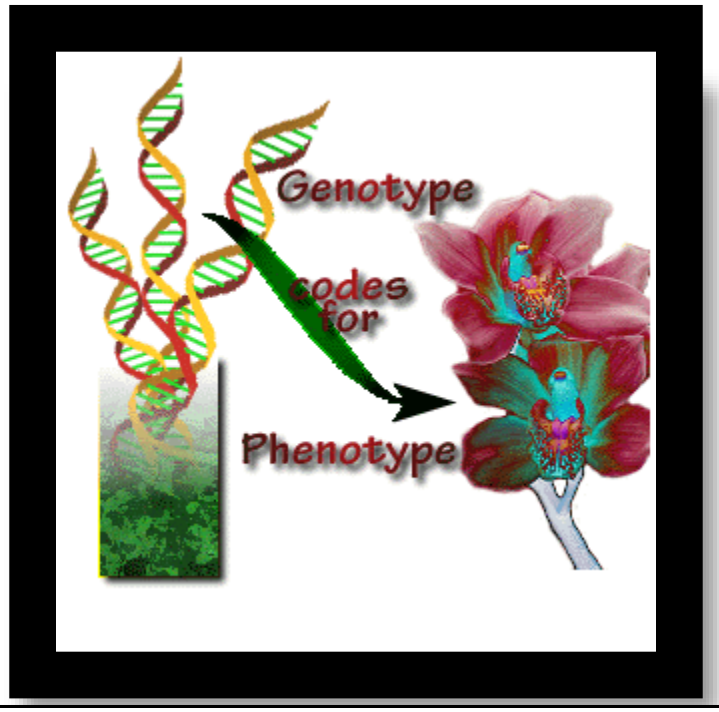
Explain Mendel's results using
…dominant & recessive
…phenotype & genotype



Genetics vs. appearance

- There can be a difference between how an organism looks & its genetics
  - appearance or trait = phenotype
    - brown eyes vs. blue eyes
  - genetic makeup = genotype
    - BB, B*b*, *bb*

2 people can have the same appearance but have different genetics: BB vs B*b*



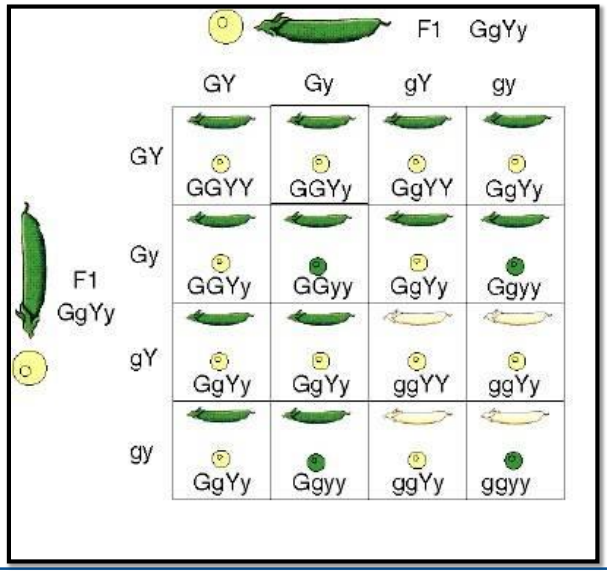
Independent Assortment

- How do alleles segregate when more than one gene is involved?
  - **The principle of independent assortment** states that genes for different traits can segregate independently during the formation of gametes.
  - Because it involves two different genes, Mendel's experiment is known as a two-factor, or dihybrid, cross. Single-gene crosses are monohybrid *crosses.*
  - Mendel's *Law of Segregation* states that allele pairs separate during gamete formation, and randomly unite at fertilization.

Independent Assortment

- A two factor cross is used to determine the outcome of 2 different genes that are passed from 1 generation to the next
  - True breeding organisms in the P generation when mated with another true breeding organism of a different trait, produce hybrid F_1 offspring. When these F_1 hybrid offsprings are mated, a variety of traits are expressed in the F_2 generation.
  - This exhibits the principle of **independent assortment** or that genes sort independently
- The law of **independent assortment** states that allele pairs separate independently during the formation of gametes. Therefore, traits are transmitted to offspring independently of one another giving different traits an equal opportunity of occurring together.

Dihybrid Cross

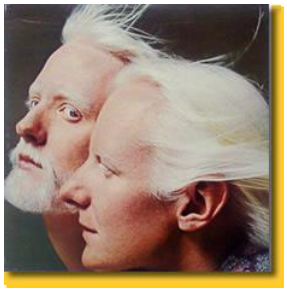


- Genes that segregate independently- such as the genes for height and hair color- do **NOT** influence each other's inheritance
- **The principle of independent assortment states that genes for different traits can segregate independently during the formation of gametes**
  - Independent assortment accounts for the genetic variations in organisms

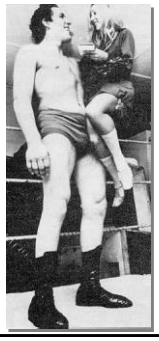
QUESTION AND ANSWER

How can we use probability to predict traits?

What are some exceptions to Mendel's principles?

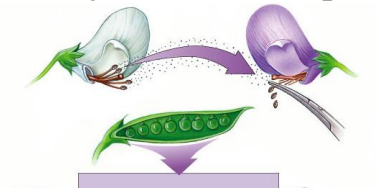


Beyond Mendel's Laws of Inheritance



Extending Mendelian genetics

- Mendel worked with a simple system
  - peas are genetically simple
  - most traits are controlled by single gene
  - each gene has only 2 version
    - 1 completely dominant (*A*)
    - 1 recessive (*a*)
- But its usually not that simple!



Gregor Mendel

Exceptions to Mendel's principles

- Not all genes show simple patterns of dominant and recessive alleles as the majority have more than 2 alleles and many important traits are controlled by multiple alleles

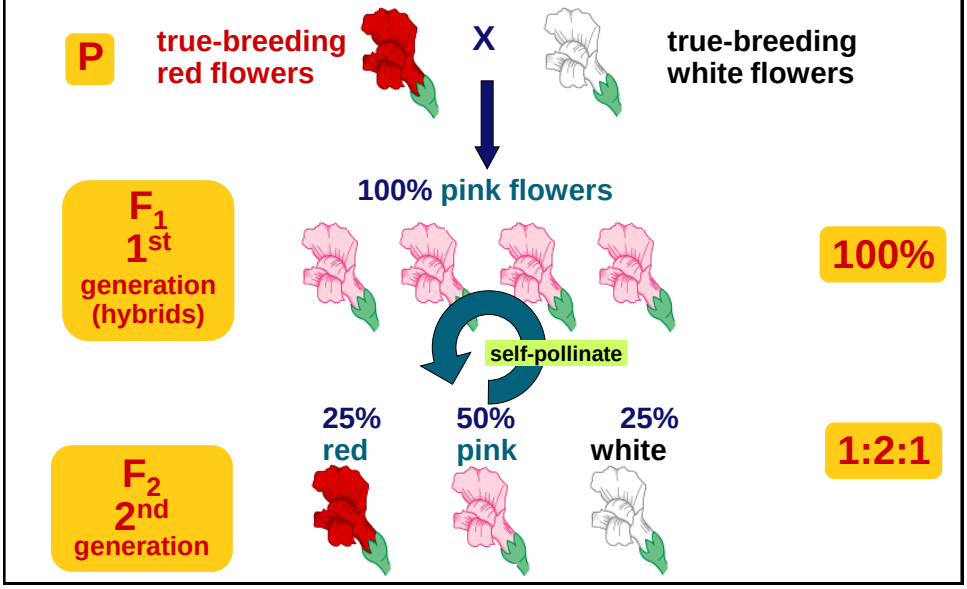
Incomplete dominance

- **Incomplete dominance** occurs when one allele is not completely dominant over another
- The heterozygous phenotype is "in between" or "blended" version the two homozygous phenotypes of the parents
  - E. g. Crossing a red and a white flower produces pink flowers

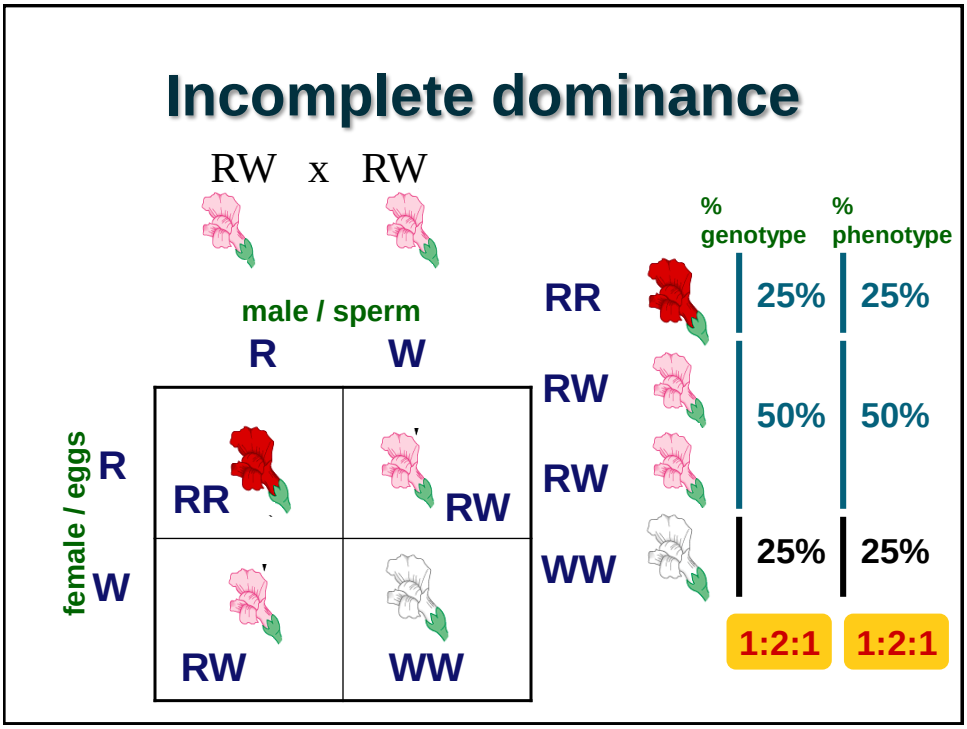
Incomplete dominance

- Hybrids have "in-between" appearance
  - RR = red flowers →**RR**
  - rr = white flowers →**WW**
  - Rr = pink flowers →**RW**
    - make 50% less color

Incomplete dominance

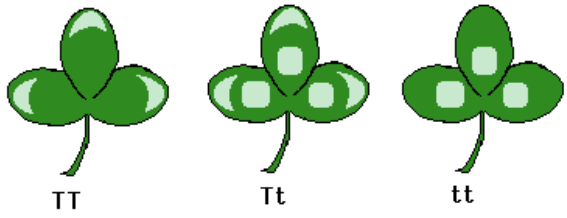


Incomplete dominance



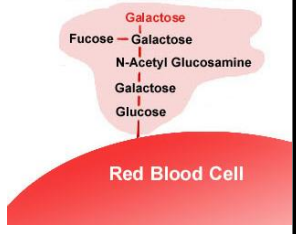
Codominance

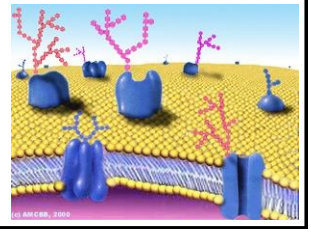
- Occurs when both alleles contribute to the phenotype
- E.g. The offspring of a black feathered chicken and a white feathered chicken would be both black and white feathered (or speckled)

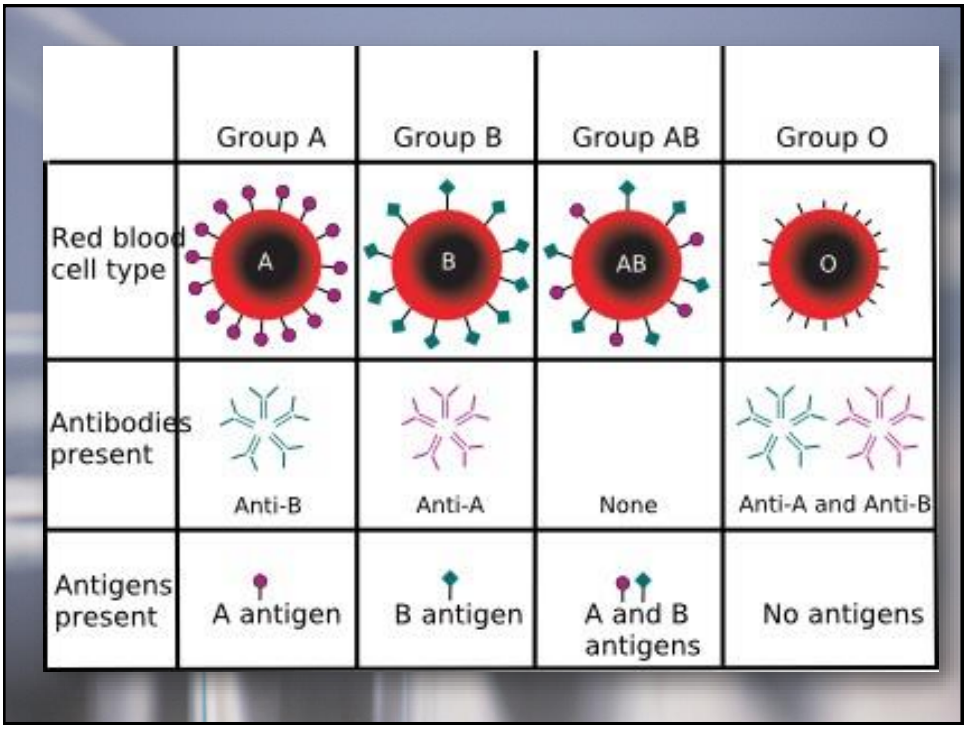


Codominance

- Equal dominance
  - human ABO blood groups
  - 3 version
    - A, B, i
    - A & B alleles are codominant
    - both A & B alleles are dominant over *i* allele
  - the genes code for different sugars on the surface of red blood cells
    - "name tag" of red blood cell







- There are 3 alleles for the ABO blood group- I^A, I^B, and I
  - I^A and I^B are codominant and produce both A and B antigens on the surface of red blood cells
  - Blood type O (*ii*) produce no antigens on the surface
  - I^AI^B (AB) is the universal acceptor of blood and *ii* or O is the universal donor of blood

- **Genotypes of the blood groups are:**
  - Group A: I^AI^A, I^Ai
  - Group B: I^BI^B, I^Bi
  - Group AB: I^AI^B
  - Group O: *ii*

Blood donation

(a) Phenotype (blood group)	(b) Genotypes	(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
A	$I^A I^A$ *or* $I^A i$	Anti-B		clotting	clotting	
B	$I^B I^B$ *or* $I^B i$	Anti-A	clotting		clotting	
AB	$I^A I^B$	—				
O	*ii*	Anti-A Anti-B	clotting	clotting	clotting	

Multiple alleles

- 3 or more alleles of the same gene
- E.g. Coat color in rabbits- 4 possible coat colors exist

Polygenic traits

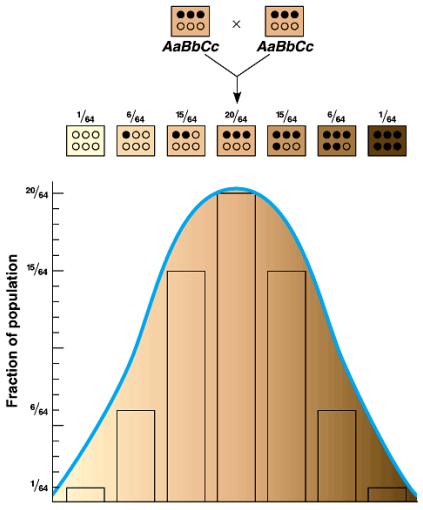
- Polygenic traits are traits that are controlled by two or more genes
- Often show a wide range of phenotypes
  - E.g. Skin color in humans is determined by 4 or more different genes

Many genes: one trait

- **Polygenic inheritance**
  - additive effects of many genes
  - humans
    - skin color
    - height
    - weight
    - eye color
    - intelligence
    - behaviors

Human skin color

- AaBbCc x AaBbCc
  - can produce a wide range of shades
  - most children = intermediate skin color
  - some can be very light & very dark



Environment effect on genes

- Phenotype is controlled by both the environment & genes

Human skin color is influenced by both genetics & environmental conditions

Color of Hydrangea flowers is influenced by soil pH

Coat color in arctic fox influenced by heat sensitive alleles

(a)

(b)