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A wild carrot has a small, tough, pale, bitter white root; while modern domestic carrots usually have a swollen, sweet, orange root. Carrots originated in present day Afghanistan about 5000 years ago, probably originally as a purple or yellow root. Purple, white and yellow carrots were imported to southern Europe in the 14th century and were widely grown in Europe into the 17th Century.



The Dutch growers developed them to be sweeter, more practical and more orange. Finally we have the French to thank for popular modern varieties, with credit to the 19th century horticulturist Louis de Vilmorin, who laid the foundations for modern plant breeding.

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**Selective Breeding** is the process by which humans choose the traits they find most desirable and allow only those individuals to reproduce.



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## What is Heredity?

Heredity is the passing of traits from parents to offspring

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## How did you get your traits?

Half of your genes (on chromosomes) came from your mom, and half from your dad.

This gives each person a unique genetic code.

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## What is Genetics?

Genetics is the Study of Heredity

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## ***Why Is Your Combination of Genes/Traits Unique?***

When the egg and sperm were formed during meiosis, **crossing over** and **independent assortment** mixed up your genes, giving you a one-of-a-kind genotype.

In addition **genetic recombination** as a result of sexual reproduction (egg + sperm = zygote), is all based on **random chance**.

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## **Every day we observe variations among individuals in a population.**

(human eyes vary from brown, green, blue, to gray)

We already know how these traits are transmitted from parents to offspring **but how does your body decide what color eyes to have?**

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## **The History of Genetics**

(or all the section of the notes where we talk about all the silly things people used to think were true)

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### *The “Blending” Hypothesis.*

This hypothesis proposed that the **genetic material** contributed by each parent **mixes** in a manner analogous to the way **blue** and **yellow** paint blend to make **green**.

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Under the **blending system** we would expect to see large populations of **uniform individuals**.



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For example **blue** and **brown** eyed parents should have children with eyes that are whatever color is blue and brown mixed together

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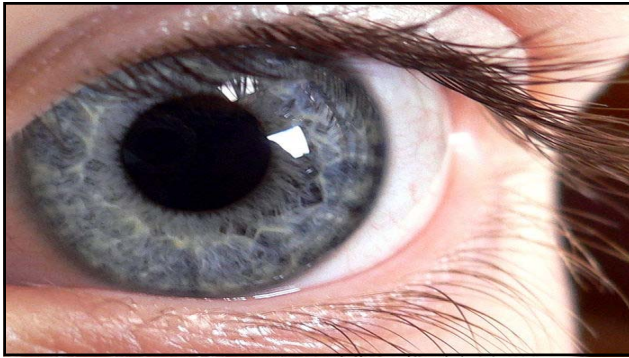
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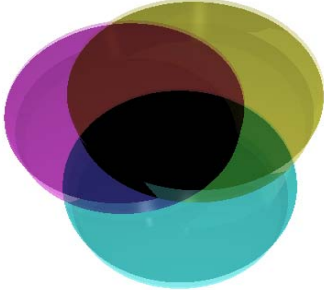
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The problem with blending is that eventually everyone would have the same color eyes

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The “blending” hypothesis is shown to be incorrect by everyday observations

Parents with **brown** and **blue** eyes usually have **brown eyed offspring**

The results of breeding experiments also contradict blending predictions.

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An alternative to the blending model is called “particulate” inheritance.

Particulate Inheritance proposes that parents pass on discrete heritable units - called genes - that retain their separate identities in offspring.

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**In “particulate” inheritance genes can be sorted and passed on, generation after generation, in an unmixed form.**

Modern genetics began in an abbey garden, where a monk names **Gregor Mendel** documented the particulate mechanism of inheritance.

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- Mendel grew up on a small farm in what is today the Czech Republic.
- In 1843, Mendel entered an Augustinian monastery.
- He studied at the University of Vienna from 1851 to 1853 where he was influenced by a physicist who encouraged experimentation and the application of mathematics to science and by a botanist who aroused Mendel’s interest in the causes of variation in plants.
- These influences came together in Mendel’s experiments.

**Mendel**  
**Father of Genetics**



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- After the university, Mendel taught at the Brunn Modern School and lived in the local monastery.
- The monks at this monastery had a long tradition of interest in the breeding of plants, including peas.
- **Around 1857, Mendel began breeding garden peas to study inheritance.**
- Pea plants have several advantages for genetics.
- Pea plants are available in many varieties with distinct heritable features (**characters**) with different variants (**traits**).

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In 1865 Mendel announced his findings about the laws of heredity, resulting from eight years of study on pea plants and their traits.

**But nobody noticed**

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Unfortunately Mendel's work was not appreciated by scientists until 1900, sixteen years after his death.

Today Mendel's *Laws of Heredity* are the basis of modern genetics.

**They are the Laws of Dominance,  
Segregation,  
Independent Assortment.**

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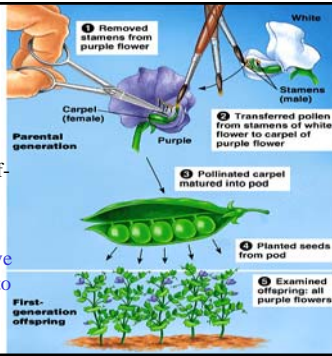
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Another advantage of peas is that Mendel had strict control over which plants mated with which.

Each pea plant has male and female sexual organs.

In nature, pea plants typically self-fertilize, fertilizing ova with their own sperm.

However, Mendel could also move pollen from one plant to another to cross-pollinate plants.



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In a typical breeding experiment, Mendel would cross-pollinate (**hybridize**) two contrasting, **true-breeding** pea varieties.

- Parents are called the **P generation**
- Their offspring are called the **F<sub>1</sub> generation**.

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Mendel would then allow the F<sub>1</sub> hybrids to self-pollinate to produce an F<sub>2</sub> generation.

It was mainly Mendel's quantitative analysis of F<sub>2</sub> plants that revealed the two fundamental principles of heredity:

- the law of segregation
- and
- the law of independent assortment.

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**By the law of segregation, the two alleles for a character are packaged into separate gametes**

- If the blending model were correct, the F<sub>1</sub> hybrids from a cross between purple-flowered and white-flowered pea plants would have **pale purple flowers**.
- Instead, the F<sub>1</sub> hybrids **all have purple flowers, just as purple as the purple-flowered parents**.



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- When Mendel allowed the F<sub>1</sub> plants to self-fertilize, the F<sub>2</sub> generation included both purple-flowered and white-flowered plants.
- **The white trait, absent in the F<sub>1</sub>, reappeared in the F<sub>2</sub>.**



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- This cross produced a **three purple** to one white ratio of traits in the F<sub>2</sub> offspring.
- Mendel reasoned that the heritable factor for white flowers was present in the F<sub>1</sub> plants, but it did not affect flower color.
- **Purple flower color is a dominant trait and white flower color is a recessive trait.**

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The reappearance of white-flowered plants in the F<sub>2</sub> generation indicated that the heritable factor for the **white trait was not diluted** or “blended” by coexisting with the purple-flower factor in F<sub>1</sub> hybrids.

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Mendel found similar 3 to 1 ratios of two traits among F<sub>2</sub> offspring when he conducted crosses for six other characters, each represented by **two different varieties**.

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- For example, when Mendel crossed two true-breeding varieties, one of which produced **round seeds**, the other of which produced **wrinkled seeds**
- All the F<sub>1</sub> offspring had round seeds
- The F<sub>2</sub> plants, 75% of the seeds were round and 25% were wrinkled.

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Table 14.1 The Results of Mendel's F <sub>2</sub> Crosses for Seven Characters in Pea Plants					
Character	Dominant Trait	×	Recessive Trait	F <sub>2</sub> Generation Dominant:Recessive	Ratio
Flower color	 Purple	×	 White	705:224	3.15:1
Flower position	 Axial	×	 Terminal	651:207	3.14:1
Seed color	 Yellow	×	 Green	6022:2001	3.01:1
Seed shape	 Round	×	 Wrinkled	5474:1850	2.96:1
Pod shape	 Inflated	×	 Constricted	882:299	2.95:1
Pod color	 Green	×	 Yellow	428:152	2.82:1
Stem length	 Tall	×	 Dwarf	787:277	2.84:1

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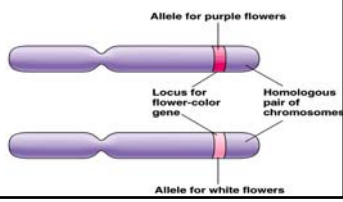
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- Mendel developed a hypothesis to explain these results that consisted of **four** related ideas.
  - Alternative versions of genes (different **alleles**) account for variations in inherited characters.
    - The purple-flower allele and white-flower allele are two DNA variations at the flower-color locus.




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- For each character, an organism inherits **two alleles**, one from each parent.
  - A diploid organism inherits one set of chromosomes from each parent.
  - Each diploid organism has **a pair** of homologous chromosomes and therefore **two copies** of each locus.

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- These homologous loci may be identical or the two alleles may differ.
  - In the flower-color example, the F<sub>1</sub> plants inherited a purple-flower allele from one parent and a white-flower allele from the other.

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3. If two alleles differ, then one, the **dominant allele**, is fully expressed in the organism's appearance.

- The other, the **recessive allele**, has no noticeable effect on the organism's appearance.
  - Mendel's F<sub>1</sub> plants had purple flowers because the purple-flower allele is dominant and the white-flower allele is recessive.

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4. The two alleles for each character segregate (separate) during gamete production.

- This segregation of alleles corresponds to the distribution of homologous chromosomes to different gametes in **meiosis**.
  - If an organism has identical alleles for a particular character, then that allele exists as a single copy in all gametes.
  - If different alleles are present, then 50% of the gametes will receive one allele and 50% will receive the other.

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The separation of alleles into separate gametes is summarized as Mendel's law of Segregation.

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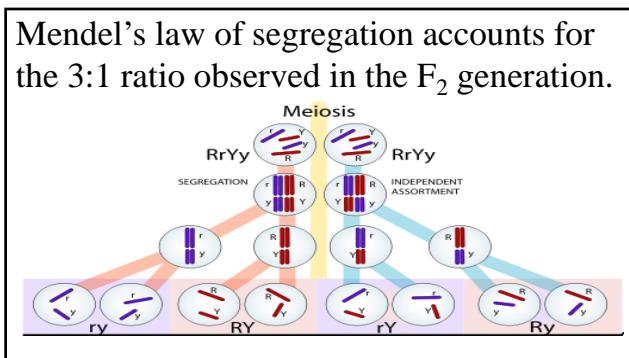
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Mendel's law of segregation accounts for the 3:1 ratio observed in the F<sub>2</sub> generation.




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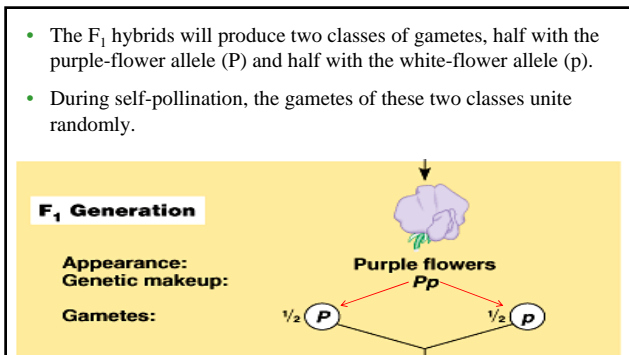
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- The F<sub>1</sub> hybrids will produce two classes of gametes, half with the purple-flower allele (P) and half with the white-flower allele (p).
- During self-pollination, the gametes of these two classes unite randomly.




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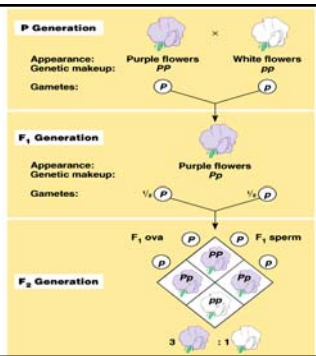
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- A **Punnett square** predicts the results of a genetic cross between individuals of known genotype.
- This can produce four equally likely combinations of sperm and ovum.
- (PP) (Pp) (Pp) (pp)




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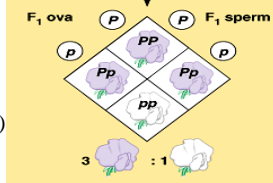
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- A Punnett square analysis of the flower-color example demonstrates Mendel's model.
- **One** in four F<sub>2</sub> offspring will inherit two white-flower alleles and produce white flowers. (pp)
- **Half** of the F<sub>2</sub> offspring will inherit one white-flower allele and one purple-flower allele and produce purple flowers. (Pp)
- **One** in four F<sub>2</sub> offspring will inherit two purple-flower alleles and produce purple flowers too. (PP)




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An organism with two identical alleles for a character is **homozygous** for that character.

**tt**  
homozygous recessive

**TT**  
homozygous dominant

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Organisms with two different alleles for a characteristic is **heterozygous** for that trait.

**Tt**  
heterozygous

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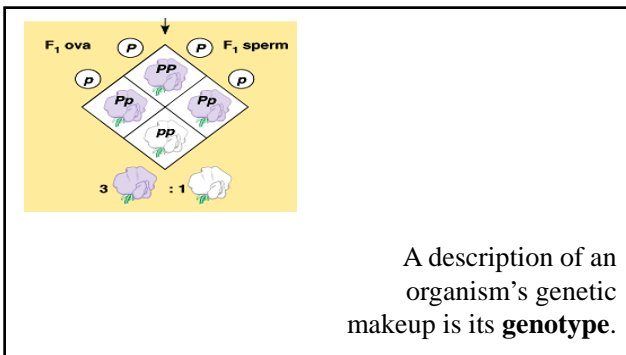
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## MENDEL AND THE GENE IDEA

- **Segregation**, the two alleles for a character are packaged into separate gametes
- **Independent assortment**, each pair of alleles segregates into gametes independently
- **Dominance**, some alleles are more “powerful” than other (recessive) alleles

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Mendelian inheritance reflects the rules of probability

Mendel discovered the particulate behavior of genes

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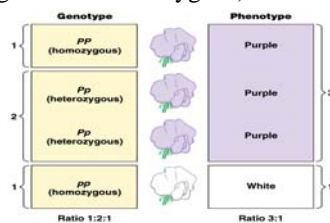
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- For flower color in peas, both  $PP$  and  $Pp$  plants have the same phenotype (purple) but different genotypes (homozygous and heterozygous).
- The only way to produce a white phenotype is to be homozygous recessive ( $pp$ ) for the flower-color gene.



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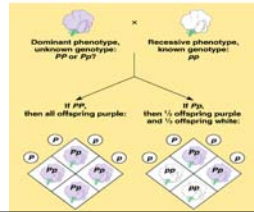
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- It is not possible to predict the genotype of an organism with a dominant phenotype.
  - The organism must have one dominant allele, but it could be homozygous dominant or heterozygous.
- A **testcross**, breeding a homozygous recessive with dominant phenotype, but unknown genotype, can determine the identity of the unknown allele.




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**Table 14.1 The Results of Mendel's F<sub>1</sub> Crosses for Seven Characters in Pea Plants**

Character	Dominant Trait	×	Recessive Trait
Flower color	Purple	×	White
Flower position	Axial	×	Terminal
Seed color	Yellow	×	Green

Cross a Heterozygous Axial Flower with a Terminal

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**Cross a Heterozygous Axial flower with a Terminal flower**

The first step is to define your variables:  
 A = Axial  
 a = Terminal

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Cross a Heterozygous Axial flower with a Terminal flower

A = Axial  
a = Terminal

The second step is to define the parent genotypes

Heterozygous Axial = Aa

Terminal = aa

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Cross a Heterozygous Axial flower with a Terminal flower

Heterozygous Axial = Aa

Terminal = aa

A = Axial  
a = Terminal

Third step is to load the gametes onto the outside of the Punnett square

	A	a
a		
a		

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Cross a Heterozygous Axial flower with a Terminal flower

Heterozygous Axial = Aa

Terminal = aa

A = Axial  
a = Terminal

Fourth step is to load the inner cells with the gametes to predict the possible zygotes

1<sup>st</sup> do one set of gametes

	A	a
a	A	a
a	A	a

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Cross a Heterozygous Axial flower with a Terminal flower

Heterozygous Axial =  $Aa$                        $A$  = Axial  
 Terminal =  $aa$                                        $a$  = Terminal

Fourth step is to load the inner cells with the gametes to predict the possible zygotes

Then do the other

	$A$	$a$
$a$	$Aa$	$aa$
$a$	$Aa$	$aa$

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Cross a Heterozygous Axial flower with a Terminal flower

Heterozygous Axial =  $Aa$                        $A$  = Axial  
 Terminal =  $aa$                                        $a$  = Terminal

The fifth step is to count the zygotes and make ratios

**Genotype**

50%  $Aa$

50%  $aa$

**Phenotype**

50% Axial

50% terminal

	$A$	$a$
$a$	$Aa$	$aa$
$a$	$Aa$	$aa$

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Table 14.1 The Results of Mendel's  $F_1$  Crosses for Seven Characters in Pea Plants

Character	Dominant Trait	×	Recessive Trait
Flower color	 Purple	×	 White
Flower position	 Axial	×	 Terminal
Seed color	 Yellow	×	 Green

Cross a Homozygous Yellow seed with a Heterozygous seed

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Cross a Homozygous Yellow seed with a Heterozygous seed

Homozygous = **YY**  
Heterozygous = **Yy**

Y = Yellow  
y = green

	<b>Y</b>	<b>Y</b>
<b>Y</b>	<b>YY</b>	<b>YY</b>
<b>y</b>	<b>Yy</b>	<b>Yy</b>

Genotype: 50% YY 50% Yy

Phenotype: 100% Yellow

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Table 14.1 The Results of Mendel's F<sub>1</sub> Crosses for Seven Characters in Pea Plants

Character	Dominant Trait	×	Recessive Trait
Flower color	 Purple	×	 White
Flower position	 Axial	×	 Terminal
Seed color	 Yellow	×	 Green

Cross Two Heterozygous Yellow Seeds

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Cross a Two Heterozygous Yellow seeds

Heterozygous = **Yy**  
Heterozygous = **Yy**

Y = Yellow  
y = green

	<b>Y</b>	<b>y</b>
<b>Y</b>	<b>YY</b>	<b>Yy</b>
<b>y</b>	<b>Yy</b>	<b>yy</b>

Genotype: 25% YY 50% Yy 25% yy

Phenotype: 75% Yellow 25% Green

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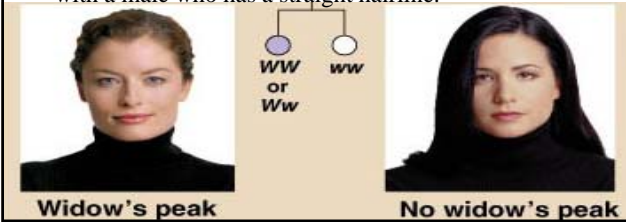
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If the occurrence of widows peak (W) is dominant to a straight hairline (w) then

- Cross a Female who is heterozygous for widows peak with a male who has a straight hairline.




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Cross a Female who is heterozygous for widows peak with a male who has a straight hairline.

Heterozygous =  $Ww$        $W$  = Widows Peak  
Homozygous Recessive =  $ww$        $w$  = straight hairline

	$W$	$w$
$w$	$Ww$	$ww$
$w$	$Ww$	$ww$

Genotype: 50%  $Ww$  | 50%  $ww$   
Phenotype: 50% Peak | 50% Straight

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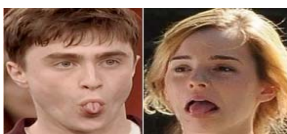
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Tongue rolling is dominant  
To no rolling... or is it?

Most people, when first asked, either can easily roll their tongue (here called "R"), or cannot roll it at all ("NR"). The proportion of people who can roll their tongue ranges from 65 to 81 percent, with a slightly higher proportion of tongue-rollers in females than in males.

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Komai (1951)

Parents	R offspring	NR offspring	Percent R
R x R	928	104	90%
R x NR	468	217	68%
NR x NR	48	92	34%

Family studies clearly demonstrate that tongue rolling is not a simple genetic character, and twin studies demonstrate that it is influenced by both genetics and the environment.

Despite this, tongue rolling is probably the most commonly used classroom example of a simple genetic trait in humans.

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If tongue rolling was a simple dominant and recessive trait what would happen if a female who is homozygous dominant crosses with a male who is homozygous recessive?

Homozygous Dominant = **TT**      T = can roll

Homozygous Recessive = **tt**      t = can't roll

	<b>T</b>	<b>T</b>
<b>t</b>	<b>Tt</b>	<b>Tt</b>
<b>t</b>	<b>Tt</b>	<b>Tt</b>

Genotype: 100% Tt

Phenotype: 100% Rolls

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Some people have earwax that is wet, sticky and yellow or brown; other people's earwax is dry, crumbly and grayish. Variation at a single gene determines which kind of earwax you have; the allele for wet earwax is dominant over the allele for dry earwax.

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The allele for dry earwax appears to have originated by mutation in northeastern Asia about 2,000 generations ago, then spread outwards because it was favored by natural selection. It is very common in eastern Asia, becomes much less common towards Europe, and is very rare in Africa.

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**Molecular genetics**

Tomita et al. (2002) used eight Japanese families to determine that the gene for wet/dry earwax is on chromosome 16, near the centromere.

Yoshiura et al. (2006) then found the gene responsible: ABCC11 (ATP-binding cassette, subfamily C, member 11).

The allele for wet earwax has a G at site 538 of the coding region, which causes a glycine at position 180 in the amino acid sequence; most dry alleles have an A at site 538, coding for arginine.

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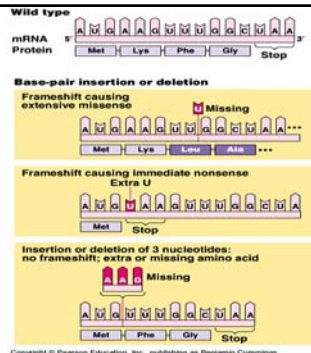
Point mutations and base-pair insertion or deletion

Swapping a G for an A is an example of a Point Mutation. A point mutation will only change one codon.

A Base Insertion is when one is added

A Base Deletion is when one base is removed

Insertions and Deletions are worse for protein synthesis as they will change all of the codons from the point of the mutation forward.




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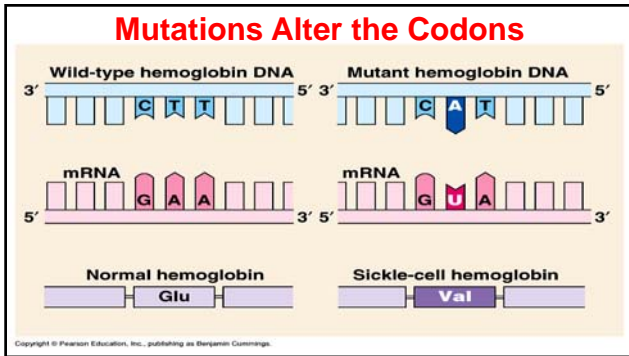
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**By the law of independent assortment, each pair of alleles segregates into gametes independently**

- Mendel's experiments that followed the inheritance of flower color or other characters focused on only a single character via **monohybrid** crosses.
- He conducted other experiments in which he followed the inheritance of two different characters, a **dihybrid** cross.

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- In one dihybrid cross experiment, Mendel studied the inheritance of **seed color and seed shape**.
  - The allele for yellow seeds (*Y*) is dominant to the allele for green seeds (*y*).
  - The allele for round seeds (*R*) is dominant to the allele for wrinkled seeds (*r*).
- Mendel crossed true-breeding plants that had yellow, round seeds (*YYRR*) with true-breeding plants that has green, wrinkled seeds (*yyrr*).

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Cross a **YYRR** x **yyrr**

**Y = Yellow peas**    **R = Round**  
**y = Green peas**    **r = Wrinkled**

Show your work and ratios

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When you cross **YYRR** x **yyrr** and get an F<sub>1</sub> phenotype of 100% Yellow and Round peas there are two possibilities:

- One possibility is that the two characters are transmitted from parents to offspring **as a package**.
- The **Y** and **R** alleles and **y** and **r** alleles stay together.
- If this were the case, the F<sub>1</sub> offspring would produce yellow, round seeds in the F<sub>2</sub> generation.

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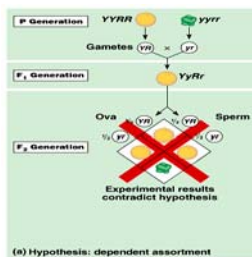
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If the **Y** and **R** alleles and **y** and **r** alleles stay together.

- The F<sub>2</sub> offspring would produce two phenotypes in a 3:1 ratio, just like a monohybrid cross.
- This was not consistent with Mendel's results.



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- An alternative hypothesis is that the two pairs of alleles **segregate independently** of each other.
  - The presence of one specific allele for one trait has no impact on the presence of a specific allele for the second trait.
- In our example, the F<sub>1</sub> offspring would still produce yellow, round seeds.
- However, when the F<sub>1</sub>'s produced gametes, genes would be packaged into gametes with all possible allelic combinations.
  - Four combinations of gametes (*YR*, *Yr*, *yR*, and *yr*) would be produced in equal amounts.

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**Cross a YYRR x yyrr**

**Y = Yellow peas    R = Round**  
**y = Green peas    r = Wrinkled**

**Show your work and ratios**

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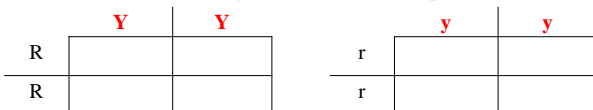
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**Cross a YYRR x yyrr**

**Y = Yellow peas    R = Round**  
**y = Green peas    r = Wrinkled**

1<sup>st</sup> find the gametes for each parent




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**Cross a YYRR x yyrr**

**Y = Yellow peas    R = Round**  
**y = Green peas    r = Wrinkled**

	<b>Y</b>	<b>Y</b>		<b>y</b>	<b>y</b>
R	YR	YR	r		
R	YR	YR	r		

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**Cross a YYRR x yyrr**

**Y = Yellow peas    R = Round**  
**y = Green peas    r = Wrinkled**

	<b>Y</b>	<b>Y</b>		<b>y</b>	<b>y</b>
R	YR	YR	r	y $r$	y $r$
R	YR	YR	r	y $r$	y $r$

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**Cross a YYRR x yyrr**

**Y = Yellow peas    R = Round**  
**y = Green peas    r = Wrinkled**

	<b>YR</b>	<b>YR</b>	<b>YR</b>	<b>YR</b>
<b>y<math>r</math></b>				
<b>y<math>r</math></b>				
<b>y<math>r</math></b>				
<b>y<math>r</math></b>				

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**Cross a YYRR x yyrr**

Y = Yellow peas      R = Round  
y = Green peas      r = Wrinkled

	YR	YR	YR	YR
yr	YyRr	YyRr	YyRr	YyRr
yr	YyRr	YyRr	YyRr	YyRr
yr	YyRr	YyRr	YyRr	YyRr
yr	YyRr	YyRr	YyRr	YyRr

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**Cross a YYRR x yyrr**

Y = Yellow peas      R = Round  
y = Green peas      r = Wrinkled

Genotype = 100% YyRr  
Phenotype = 100% Yellow Round Peas

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**Cross a YyRr x YyRr**

**Y = Yellow peas      R = Round**  
**y = Green peas      r = Wrinkled**

**Show your work and ratios**

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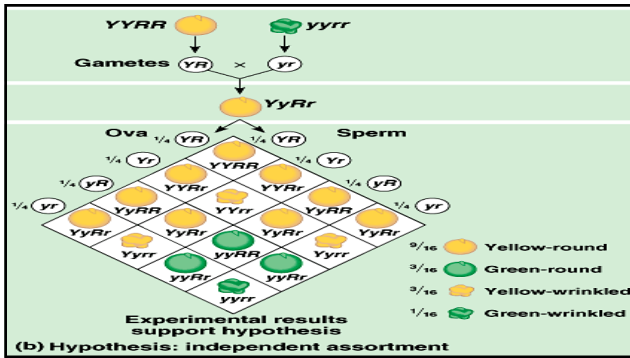
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- When sperm with four classes of alleles and ova with four classes of alleles combined, there would be 16 equally probable ways in which the alleles can combine in the  $F_2$  generation.
- These combinations produce four distinct phenotypes in a 9:3:3:1 ratio.

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- Each character is inherited independently.
- The independent assortment of each pair of alleles during gamete formation is now called Mendel's **law of independent assortment**.
- One other aspect that you can notice in the dihybrid cross experiment is that if you follow just one character, you will observe a 3:1  $F_2$  ratio for each, just as if this were a monohybrid cross.

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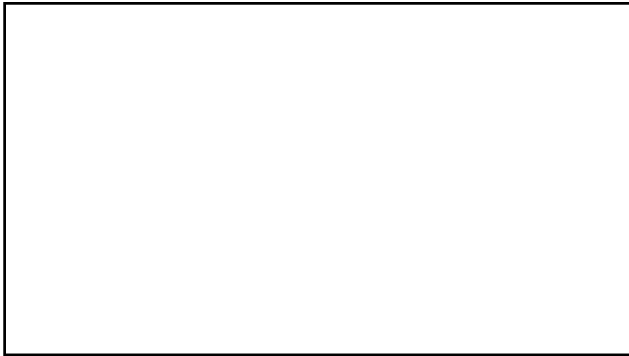
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## Coin Toss Lab

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### Mendelian inheritance rules of probability

- Mendel's laws of segregation and independent assortment reflect the same laws of probability that apply to tossing coins or rolling dice.
  - The probability of tossing heads with a normal coin is  $1/2$ .
  - The probability of rolling a 3 with a six-sided die is  $1/6$ , and the probability of rolling any other number is  $1 - 1/6 = 5/6$ .

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- When tossing a coin, the outcome of one toss has no impact on the outcome of the next toss.
- Each toss is an independent event, just like the distribution of alleles into gametes.

Each time you toss a coin the odds of heads vs tails is the same 50/50

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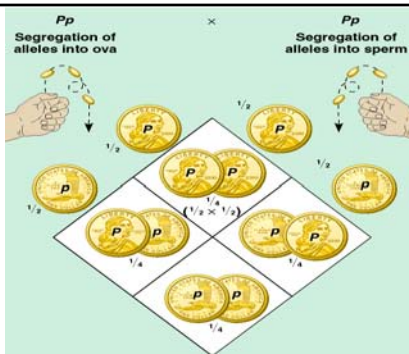
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Like a coin toss, each ovum from a heterozygous parent has a 1/2 chance of carrying the dominant allele and a 1/2 chance of carrying the recessive allele.

The same odds apply to the sperm.




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- We can use the rule of multiplication to determine the chance that two or more independent events will occur together in some specific combination.
  - Compute the probability of each independent event.
  - Then, multiply the individual probabilities to obtain the overall probability of these events occurring together.
  - The probability that two coins tossed at the same time will land heads up is  $1/2 \times 1/2 = 1/4$ .
  - Similarly, the probability that a heterozygous pea plant ( $Pp$ ) will produce a white-flowered offspring ( $pp$ ) depends on an ovum with a white allele mating with a

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- The rule of multiplication also applies to dihybrid crosses.
  - For a heterozygous parent ( $YyRr$ ) the probability of producing a  $YR$  gamete is  $1/2 \times 1/2 = 1/4$ .
  - We can use this to predict the probability of a particular  $F_2$  genotype without constructing a 16-part Punnett square.
  - The probability that an  $F_2$  plant will have a  $YYRR$  genotype from a heterozygous parent is  $1/16$  ( $1/4$  chance for a  $YR$  ovum and  $1/4$  chance for a  $YR$  sperm).

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- The rule of addition also applies to genetic problems.
  - Under the rule of addition, the probability of an event that can occur two or more different ways is the sum of the separate probabilities of those ways.
    - For example, there are two ways that  $F_1$  gametes can combine to form a heterozygote.
      - The dominant allele could come from the sperm and the recessive from the ovum (probability =  $1/4$ ).
      - Or, the dominant allele could come from the ovum and the recessive from the sperm (probability =  $1/4$ ).

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- We can combine the rules of multiplication and addition to solve complex problems in Mendelian genetics.
  - Let's determine the probability of finding two recessive phenotypes for at least two of three traits resulting from a trihybrid cross between pea plants that are  $PpYyRr$  and  $Ppyyrr$ .
    - There are five possible genotypes that fulfill this condition:  $ppyyRr$ ,  $ppYyrr$ ,  $Ppyyrr$ ,  $PPyyrr$ , and  $ppyyrr$ .
    - We would use the rule of multiplication to calculate the probability for each of these genotypes and then use the

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- The probability of producing a *ppyyRr* offspring:
  - The probability of producing *pp* =  $1/2 \times 1/2 = 1/4$ .
  - The probability of producing *yy* =  $1/2 \times 1 = 1/2$ .
  - The probability of producing *Rr* =  $1/2 \times 1 = 1/2$ .
  - Therefore, the probability of all three being present (*ppyyRr*) in one offspring is  $1/4 \times 1/2 \times 1/2 = 1/16$ .
- For *ppYyrr*:  $1/4 \times 1/2 \times 1/2 = 1/16$ .
- For *Ppyyrr*:  $1/2 \times 1/2 \times 1/2 = 2/16$
- For *PPyyrr*:  $1/4 \times 1/2 \times 1/2 = 1/16$

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### 5. Mendel discovered the particulate behavior of genes: a review

- While we cannot predict with certainty the genotype or phenotype of any particular seed from the F2 generation of a dihybrid cross, we can predict the probabilities that it will fit a specific genotype of phenotype.
- Mendel's experiments succeeded because he counted so many offspring and was able to discern this statistical feature of inheritance and had a keen sense

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- Mendel's laws of independent assortment and segregation explain heritable variation in terms of alternative forms of genes that are passed along according to simple rule of probability.
- These laws apply not just to garden peas, but to all other diploid organisms that reproduce by sexual reproduction.
- Mendel's studies of pea inheritance endure not only in genetics, but as a case study of the power of scientific reasoning using the hypothetico-

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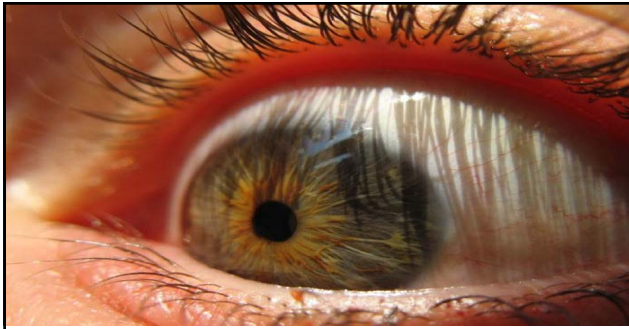
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The relationship between genotype and phenotype is rarely simple

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### Sex Linkage

- The discovery of the different sizes and shapes of the X and Y chromosomes led to the hypothesis that more genes could be carried by the X chromosome than by the smaller Y chromosome.

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### Sex Linkage

- Genes found on the **X chromosome** are said to be X-linked genes.
- Genes found on the Y chromosome are Y linked genes.
- The presence of a gene on a sex chromosome is called sex linkage.

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### Sex Linked Notation

$X^R X^R$  = a Homozygous dominant female

Sex linked traits use a superscript to show that the allele is **on the X chromosome**.

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### Sex Linked Notation

$X^R X^r$  = a Heterozygous female

Sex linked traits use a superscript to show that the allele is **on the X chromosome**.

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### Sex Linked Notation

$X^R Y$  = a dominant male

Sex linked traits use a superscript to show that the allele is **on the X chromosome**.

**There are no alleles on the Y**

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## Sex Linked Notation

**$X^r Y$**  = a recessive male

The allele is **on the X chromosome**.

**There are no alleles on the Y.**

**Males cannot be heterozygous.**

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**Males cannot be heterozygous  
for sex linked traits**

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Although most fruit flies have red eyes, a few males have white eyes. When a white-eyed male with a red-eyed female, the results of the cross followed Mendel's predictions: the F1 generation all had red eyes.

When you cross members of the F1 generation the F2 exhibited the expected ratio of three red-eyed flies to one white-eyed fly. However, all of the white-eyed flies were male.

**Why were there no white-eyed females?**

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The gene for eye color is carried on the X chromosome. the X chromosome carries the gene for eye color, either  $X^R$  (red eyes) or  $X^r$  (white eyes).

Cross a  $X^R X^R$  female (red-eyed) with a  $X^r Y$  male (white-eyed)

	$X^R$	$X^R$	Genotype: 50% $X^R X^r$ 50% $X^R Y$
$X^r$	$X^R X^r$	$X^R X^r$	
$Y$	$X^R Y$	$X^R Y$	

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	$X^R$	$X^R$	Genotype: 50% $X^R X^r$ 50% $X^R Y$
$X^r$	$X^R X^r$	$X^R X^r$	
$Y$	$X^R Y$	$X^R Y$	

**Phenotype**  
Males: 100% Red Eyed  
Females: 100% Red Eyed

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Cross a  $X^R X^r$  female (red-eyed) with a  $X^r Y$  male (white-eyed)

	$X^R$	$X^r$	Genotype: 25% $X^R X^r$ 25% $X^r X^r$ 25% $X^R Y$ 25% $X^r Y$
$X^r$	$X^R X^r$	$X^r X^r$	
$Y$	$X^R Y$	$X^r Y$	

**Phenotype**  
Males: 50% Red Eyed 50% White Eyed  
Females: 50% Red Eyed 50% White Eyed

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Cross a  $X^R X^r$  female (red-eyed) with a  $X^R Y$  male (Red-eyed)

	$X^R$	$X^r$	Genotype: 25% $X^R X^R$ 25% $X^R X^r$ 25% $X^R Y$ 25% $X^r Y$
$X^R$	$X^R X^R$	$X^r X^R$	
$Y$	$X^R Y$	$X^r Y$	

**Phenotype**

Males: 50% Red Eyed 50% White Eyed

Females: 100% Red Eyed

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**Incomplete Dominance**

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- The heterozygous  $F_1$  offspring of Mendel's crosses always looked like one of the parents because one allele was dominant to the other.
  
- However, some alleles show **incomplete dominance** where heterozygotes show a distinct **intermediate phenotype**, not seen in homozygotes.

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- **Incomplete dominance is not blended** inheritance because the traits are separable (particulate) as seen in further crosses.
- Offspring of a cross between heterozygotes will show three phenotypes: both parents and the heterozygote.
- The phenotypic and genotypic ratios are identical, 1:2:1.

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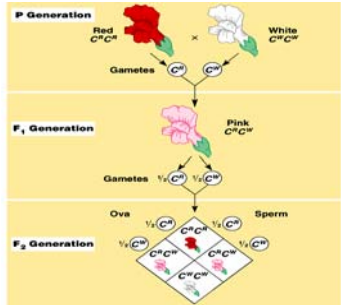
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**Incomplete dominance as seen in snapdragon flower color**

- A cross between a white-flowered plant and a red-flowered plant will produce 100% pink F<sub>1</sub> offspring.
- Self-pollination of the F<sub>1</sub> offspring produces 25% white, 25% red, and 50% pink offspring.




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**Cross a Pink flower with a White Flower**

$C^W$  = White     $C^R$  = Red

Pink Parent:  $C^W C^R$     White Parent:  $C^W C^W$

	$C^W$	$C^R$
$C^W$	$C^W C^W$	$C^R C^W$
$C^W$	$C^W C^W$	$C^R C^W$

Genotype: 50%  $C^W C^W$   
 50%  $C^R C^W$   
 Phenotype: 50% White  
 50% Pink

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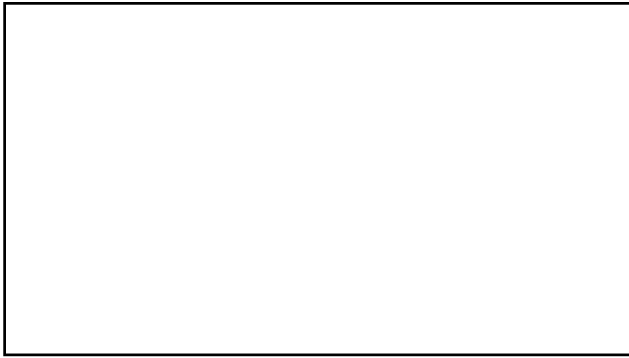
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