GENETICS

- Introduction to Genetics and heredity
- Gregor Mendel – a brief bio
- Genetic terminology (glossary)
- Monohybrid crosses
- Patterns of inheritance
- Dihybrid crosses
- Test cross
- Beyond Mendelian Genetics – incomplete dominance

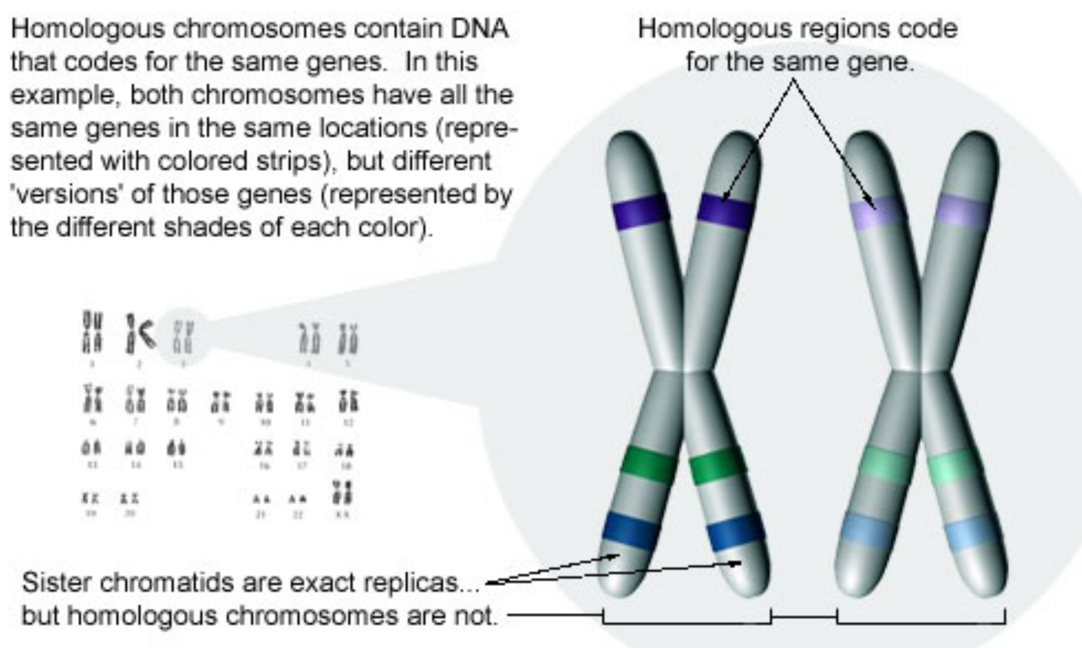
Introduction to Genetics

- **GENETICS** – branch of biology that deals with heredity and variation of organisms.

- **Chromosomes** carry the hereditary information (genes)
  - Arrangement of nucleotides in DNA
  - DNA → RNA → Proteins

- Chromosomes (and genes) occur in pairs **Homologous Chromosomes**
- New combinations of genes occur in sexual reproduction
  - Fertilization from two parents

Figure B-11: Homologous Chromosomes



Gregor Johann Mendel

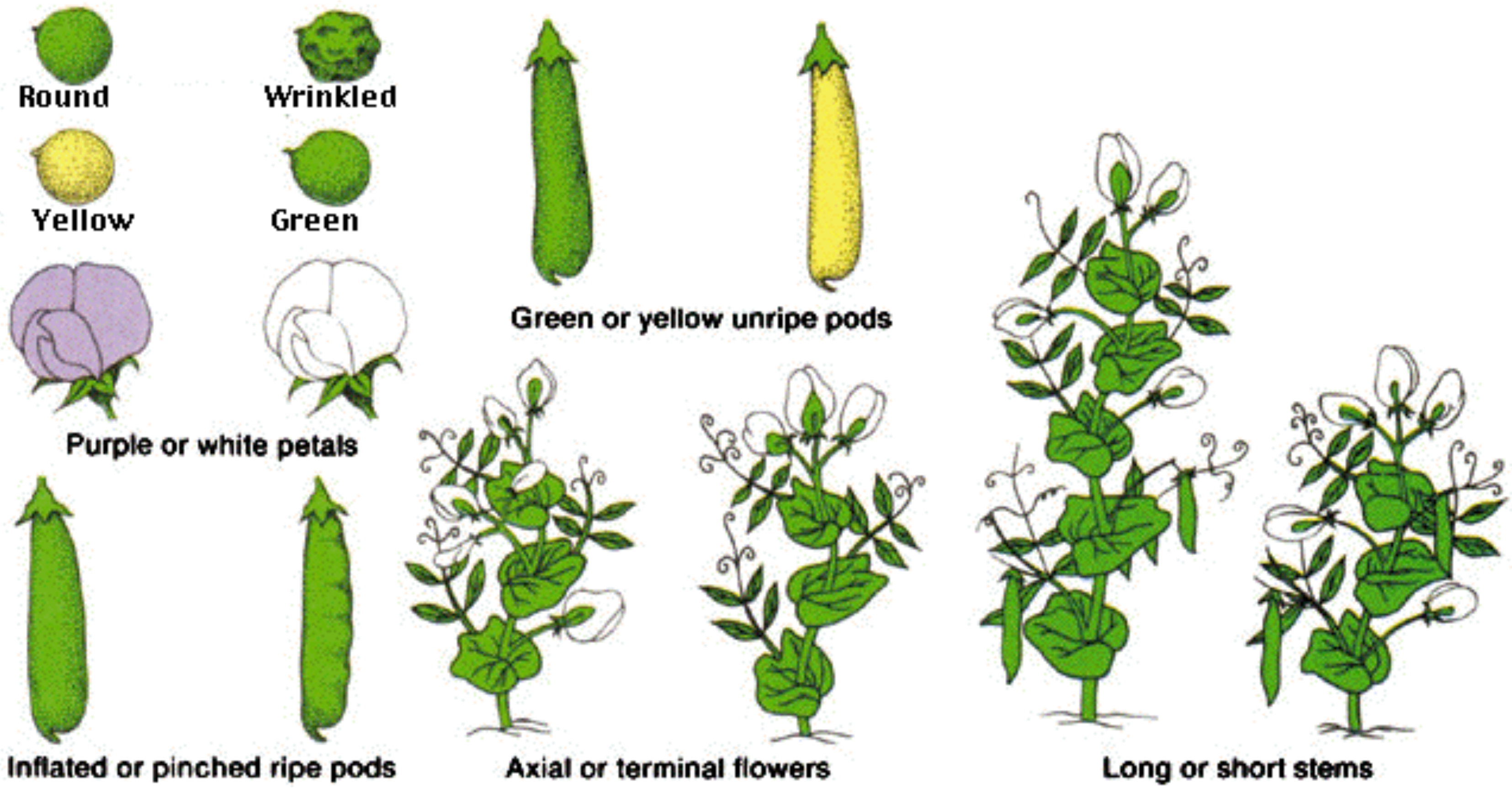
- Austrian Monk, born in what is now Czech Republic in 1822

Gregor Mendel

- Son of peasant farmer, studied Theology and was ordained priest Order St. Augustine.
- Went to the university of Vienna, where he studied botany and learned the Scientific Method
- Worked with pure lines of peas for eight years
- Prior to Mendel, heredity was regarded as a "blending" process and the offspring were essentially a "dilution"of the different parental characteristics.

Mendel's peas

- Mendel looked at seven traits or characteristics of pea plants:



- In 1866 he published *Experiments in Plant Hybridization*, (*Versuche über Pflanzen-Hybriden*) in which he established his three Principles of Inheritance
- He tried to repeat his work in another plant, but didnt work because the plant reproduced asexually!
- Work was largely ignored for 34 years, until 1900, when 3 independent botanists rediscovered Mendel's work.

- Mendel was the first biologist to use Mathematics – to explain his results quantitatively.
- Mendel predicted

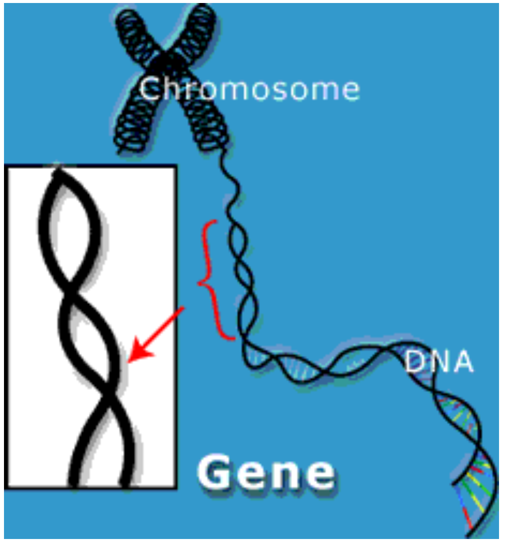
  The concept of genes

  That genes occur in pairs

  That one gene of each pair is present in the gametes

Genetics terms you need to know:

- **Gene** – a unit of heredity; a section of DNA sequence encoding a single protein
- **Genome** – the entire set of genes in an organism



- **Alleles** – two genes that occupy the same position on homologous chromosomes and that cover the same trait (like 'flavors' of a trait).
- **Locus** – a fixed location on a strand of DNA where a gene or one of its alleles is located.

- **Homozygous** – having identical alleles (one from each parent) for a particular characteristic.
- **Heterozygous** – having two different alleles for a particular characteristic.

- **Dominant** – the allele of a gene that masks or suppresses the expression of an alternate allele; the trait appears in the heterozygous condition.
- **Recessive** – an allele that is masked by a dominant allele; does not appear in the heterozygous condition, only in homozygous.

- **Genotype** – the genetic makeup of an organism
- **Phenotype** – the physical appearance of an organism (Genotype + environment)

- **Monohybrid cross**: a genetic cross involving a single pair of genes (one trait); parents differ by a single trait.
- **P** = Parental generation
- $\mathbf{F_1}$ = First filial generation; offspring from a genetic cross.

7 Characteristics in Peas

Trait	Stem length	Pod shape	Seed shape	Seed color	Flower position	Flower color	Pod color
Characteristics	 Tall	 Inflated	 Smooth	 Yellow	 Lateral	 Purple	 Green
	 Dwarf	 Constricted	 Wrinkled	 Green	 Terminal	 White	 Yellow

Monohybrid cross

- Parents differ by a single trait.
- Crossing two pea plants that differ in stem size, one tall one short

T = allele for Tall

t = allele for dwarf

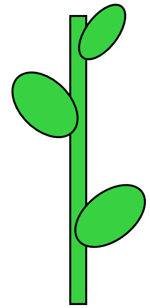
TT = homozygous tall plant

t t = homozygous dwarf plant

T T × t t

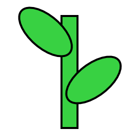
Monohybrid cross for stem length:

P = parentals
true breeding,
homozygous plants:



T T × *t t*

(tall) (dwarf)



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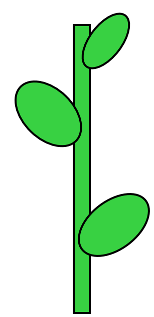
F_1 generation
is heterozygous:

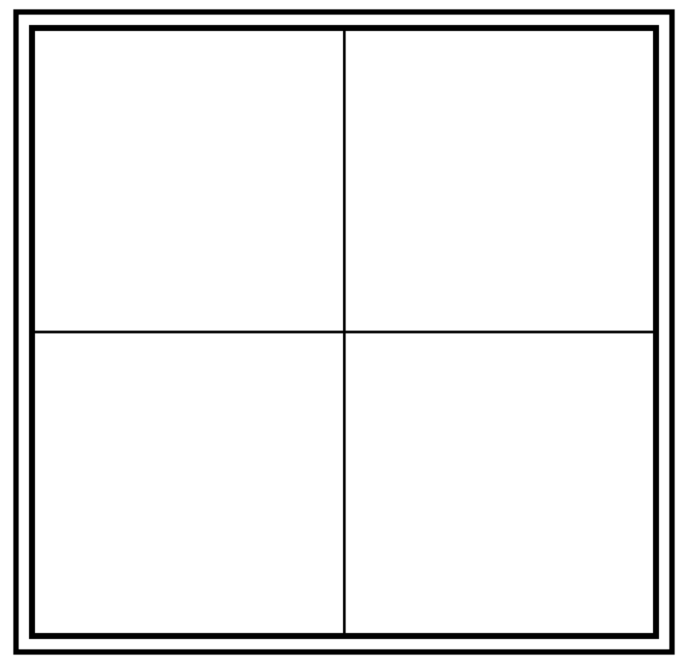
T *t*

(all tall plants)



Punnett square

- A useful tool to do genetic crosses
- For a monohybrid cross, you need a square divided by four….
- Looks like
  a window
  pane…
  We use the
  Punnett square
  to predict the
  genotypes and phenotypes of
  the offspring.



Using a Punnett Square

<u>STEPS</u>:

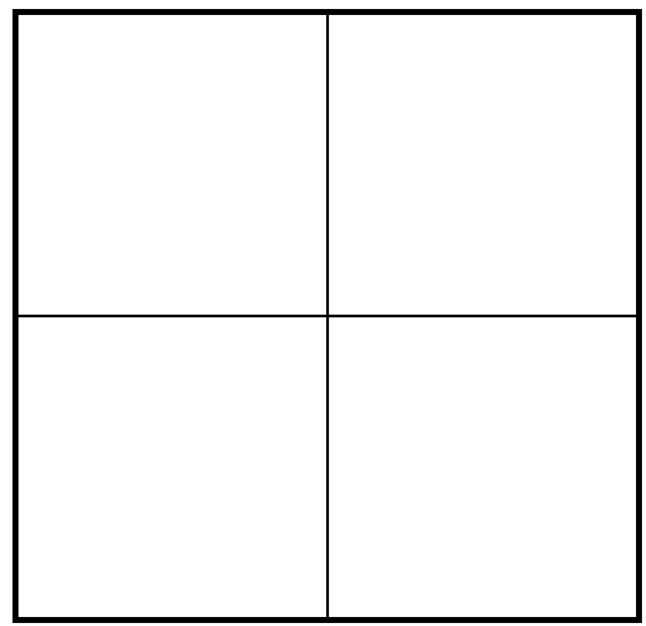
1. determine the genotypes of the parent organisms
2. write down your "cross" (mating)
3. draw a p-square

Parent genotypes:

TT and *t t*

Cross

T T × *t t*



Punnett square

4. **"split" the letters of the genotype for each parent & put them "outside" the p-square**
5. determine the possible genotypes of the offspring by filling in the p-square
6. summarize results (genotypes & phenotypes of offspring)

T T × *t t*

	T	**T**
t	**T *t***	**T *t***
t	**T *t***	**T *t***

Genotypes:
100% T t

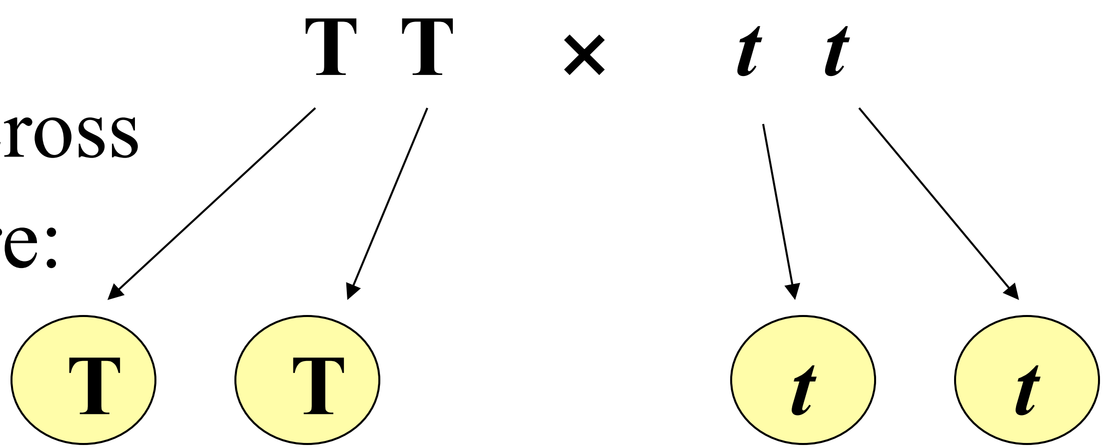
Phenotypes:
100% Tall plants

Secret of the Punnett Square

- Key to the Punnett Square:
- Determine the gametes of each parent…
- How? By "splitting" the genotypes of each parent:

If this is your cross

The gametes are:



Once you have the gametes…

T **T** × ***t*** ***t***

	t	***t***
T	**T *t***	**T *t***
T	**T *t***	**T *t***

Another example: Flower color

For example, flower color:

P = purple (dominant)

p = white (recessive)

If you cross a homozygous Purple (PP) with a homozygous white (*pp*):

P P × *p p*

↓

P *p*

ALL PURPLE (P*p*)

Cross the F1 generation:

P *p* × P *p*

	P	***p***
P	**P P**	**P *p***
p	**P *p***	***p p***

Genotypes:
1 PP
2 Pp
1 pp

Phenotypes:
3 Purple
1 White

Mendel’s Principles

- **1. Principle of Dominance**:

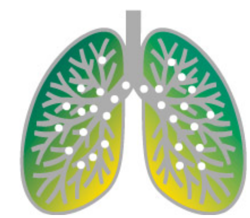
  One allele masks another

- **2. Principle of Segregation**:

  When gametes are formed, the pairs of hereditary factors (alleles) become separated, so that each sex cell (egg/sperm) receives only one kind of gene.

Human case: CF

- Mendel's Principles of Heredity apply universally to all organisms.
- Cystic Fibrosis: a lethal genetic disease affecting primarily Caucasians.
- Caused by mutant recessive gene carried by 1 in 20 people of European descent (12M)
- One in 400 Caucasian couples will be both carriers of CF – 1 in 4 children will have it.



- CF disease affects transport in tissues – mucus is accumulated in lungs, causing infections.

Inheritance pattern of CF

IF two parents carry the recessive gene of Cystic Fibrosis (*c*), that is, they are heterozygous (C *c*), one in four of their children is expected to be homozygous for *cf* and have the disease:

	C	***c***
C	**C C**	**C *c***
c	**C *c***	***c c***

C C = normal
C *c* = carrier, no symptoms
c c = has cystic fibrosis

Probabilities…

- Of course, the 1 in 4 is a ***probability*** of getting the disease, not a guarantee that 1 of 4 will have
- Important factor when prospective parents are concerned about their chances of having affected children. → Genetic Test for gene
- 1 in 29 Americans is a symptom-less carrier (Cf *cf*) of the gene.

Dihybrid crosses

- Matings that involve parents that differ in **two** genes (two independent traits)

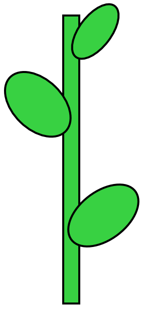
For example, flower color:

P = purple (dominant)

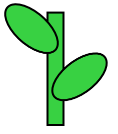
p = white (recessive)

and stem length:

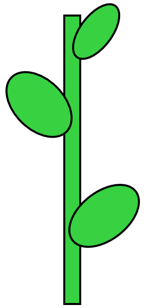
T = tall



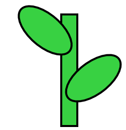
t = short



Dihybrid cross: flower color and stem length




TT PP × *tt pp*
(tall, purple) (short, white)



Possible Gametes for parents

T P and *t p*

	tp	*tp*	*tp*	*tp*
TP	T*t*P*p*	T*t*P*p*	T*t*P*p*	T*t*P*p*
TP	T*t*P*p*	T*t*P*p*	T*t*P*p*	T*t*P*p*
TP	T*t*P*p*	T*t*P*p*	T*t*P*p*	T*t*P*p*
TP	T*t*P*p*	T*t*P*p*	T*t*P*p*	T*t*P*p*

F1 Generation: All tall, purple flowers (T*t* P*p*)

Dihybrid cross F_2

If F_1 generation is allowed to self pollinate, Mendel observed 4 phenotypes:

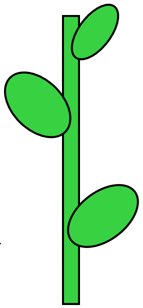
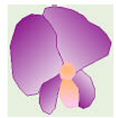
Tt Pp × Tt Pp
(tall, purple) (tall, purple)

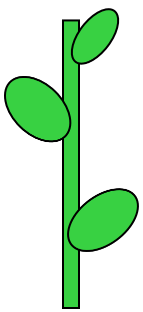
Possible gametes:
TP T*p* *t*P *tp*

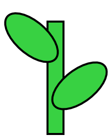
	TP	T*p*	*t*P	*tp*
TP	TTPP	TTP*p*	T*t*PP	T*t*P*p*
T*p*	TTP*p*	TT*pp*	T*t*P*p*	T*tpp*
*t*P	T*t*PP	T*t*P*p*	*tt*PP	*tt*P*p*
tp	T*t*P*p*	T*tpp*	*tt*P*p*	*ttpp*

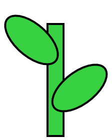
Four phenotypes observed
Tall, purple (9); Tall, white (3); Short, purple (3); Short white (1)

Dihybrid cross

9 Tall  purple 

3 Tall  white 

3 Short  purple 

1 Short  white 

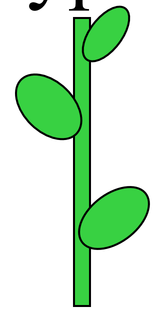
	TP	T*p*	*t*P	*tp*
TP	TTPP	TTP*p*	T*t*PP	T*t*P*p*
T*p*	TTP*p*	TT*pp*	T*t*P*p*	T*tpp*
*t*P	T*t*PP	T*t*P*p*	*tt*PP	*tt*P*p*
tp	T*t*P*p*	T*tpp*	*tt*P*p*	*ttpp*

Phenotype Ratio = 9:3:3:1

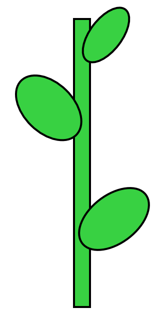
Dihybrid cross: 9 genotypes

Genotype ratios (9):		Four Phenotypes:
1	TTPP	Tall, purple (9)
2	TTP*p*	
2	T*t*PP	
4	T*t*P*p*	
1	TT*pp*	Tall, white (3)
2	T*tpp*	
1	*tt*PP	Short, purple (3)
2	*tt*P*p*	
1	*ttpp*	Short, white (1)

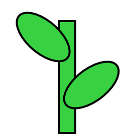
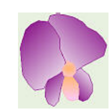
Tall, purple (9)



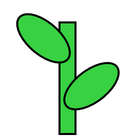

Tall, white (3)



Short, purple (3)

Short, white (1)



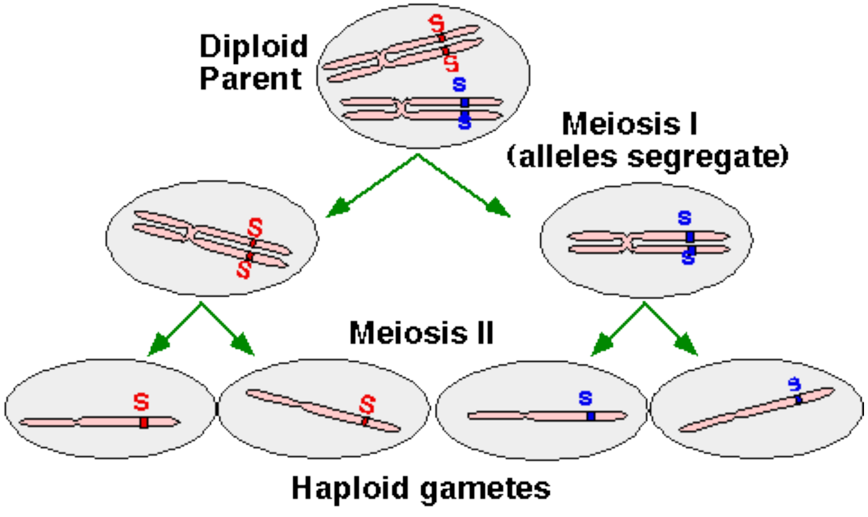
Principle of Independent Assortment

- Based on these results, Mendel postulated the **<u>3. Principle of Independent Assortment</u>**:
  "Members of one gene pair segregate independently from other gene pairs during gamete formation"

Genes get shuffled – these many combinations are one of the advantages of sexual reproduction

Relation of gene segregation to meiosis…

- There' s a correlation between the movement of chromosomes in meiosis and the segregation of alleles that occurs in meiosis



Beyond Mendelian Genetics:

Mendel was lucky!
Traits he chose in the pea plant showed up very clearly…

One allele was dominant over another, so phenotypes were easy to recognize.

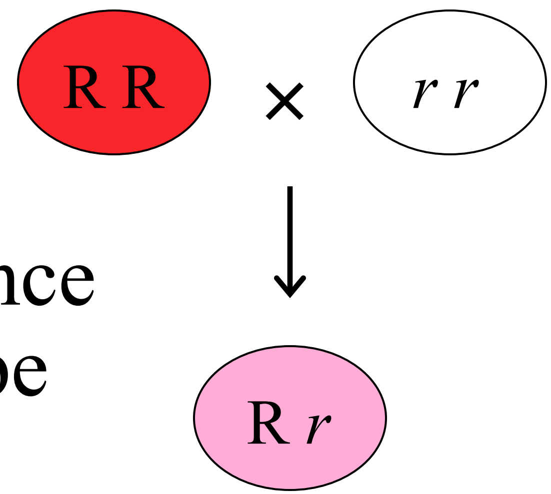
But sometimes phenotypes are not very obvious…

Incomplete Dominance

Snapdragon flowers come in many colors.

If you cross a red snapdragon (RR) with a white snapdragon (rr)

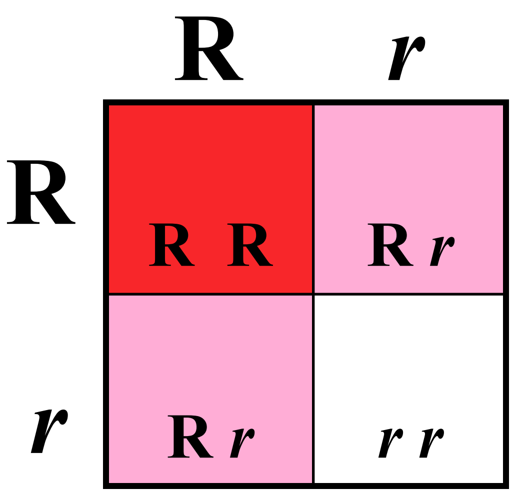
You get PINK flowers (Rr)!



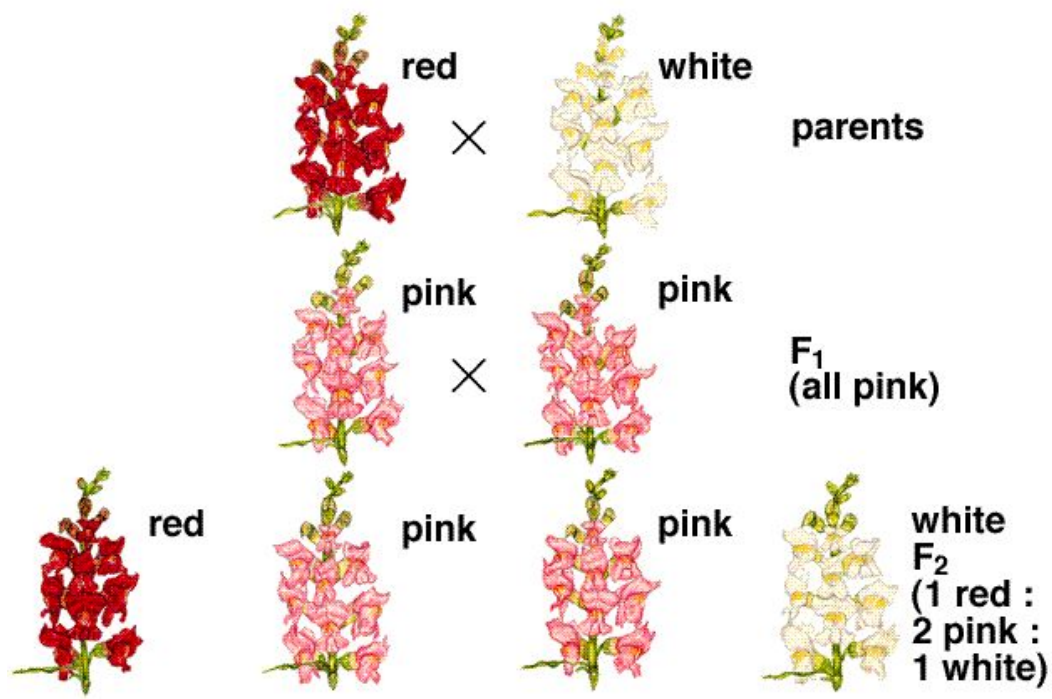
Genes show incomplete dominance when the heterozygous phenotype is intermediate.

Incomplete dominance

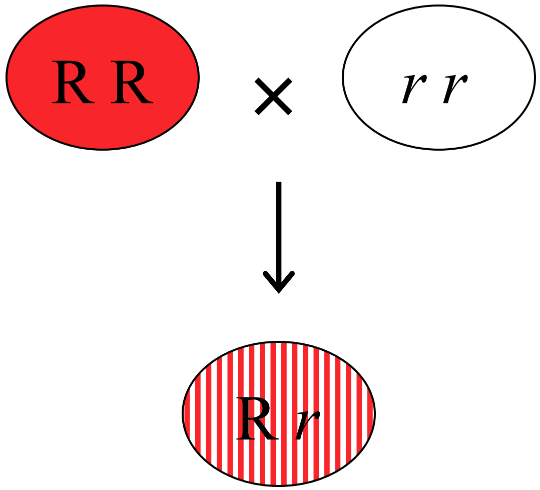
When F1 generation (all pink flowers) is self pollinated, the F2 generation is 1:2:1 red, pink, white



Incomplete Dominance



Co-dominance



Genes show Co- dominance when the heterozygous phenotype shows characteristics of both alleles.