LECTURE 1 : GENETICS

- Introduction to Genetics and heredity
- Gregor Mendel
- Genetic terminology (glossary)
- Monohybrid crosses
- Patterns of inheritance
- Dihybrid crosses
- Test cross

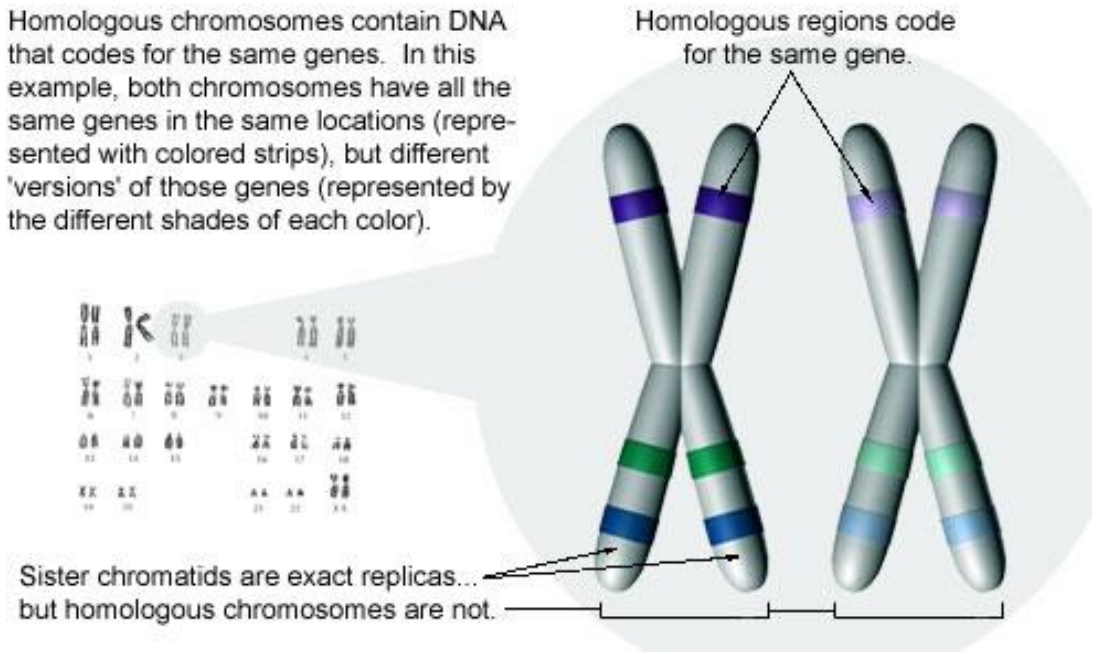
Introduction to Genetics

- **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

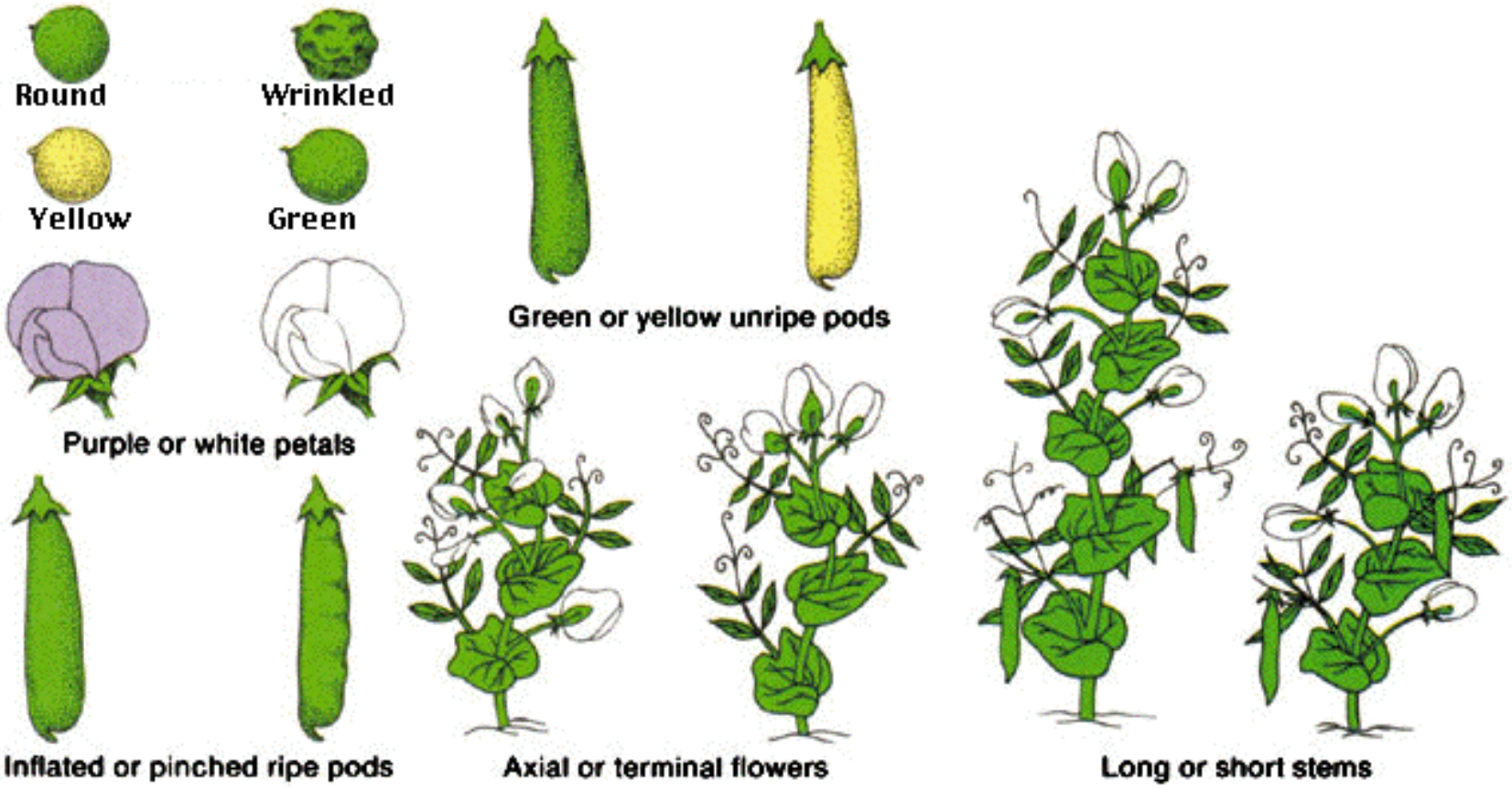
- Austrian Monk, born in what is now Czech Republic in 1822
- **Called the "Father of Genetics"**


Gregor Mendel

- 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:



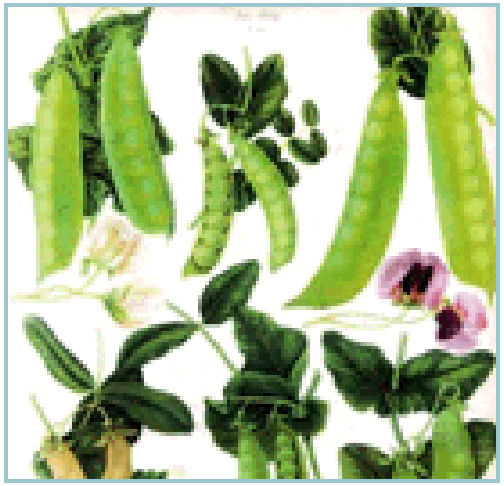
- In 1866 he published *Experiments in Plant Hybridization*, in which he established his three Principles of Inheritance
- He tried to repeat his work in another plant, but didn't work because the plant reproduced asexually! If…
- Work was largely ignored for 34 years, until 1900, when 3 independent botanists rediscovered Mendel's work.

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

  The concept of genes

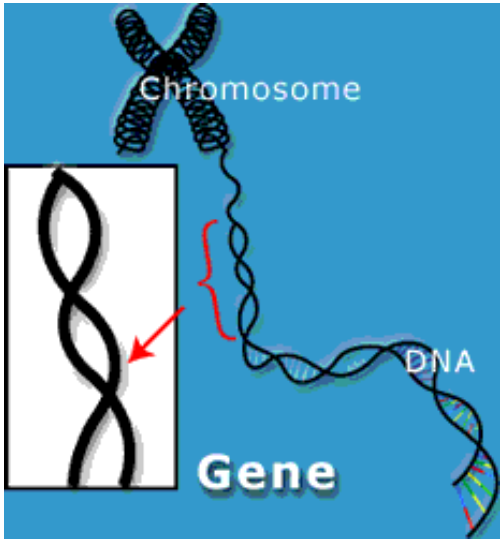
  That genes occur in pairs

  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** – A gene which can occupy the same locus as another gene in a pair of homologous chromosomes
- **Locus** – a fixed location on a strand of DNA where a gene or one of its alleles is located.

- **Homozygous** – Identical alleles present on the homologous chromosomes
- **Heterozygous** – Non identical alleles present on the homologous chromosomes

- **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 organisms
- **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.
- $\mathbf{F_2}$ = Second filial generation of 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

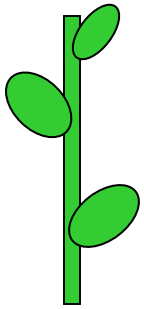
  t t = homozygous dwarf plant

Long or short stems

T T × t t

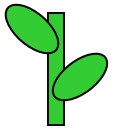
Monohybrid cross for stem length:

P = parentals
true breeding,
homozygous plants:



T T × *t t*

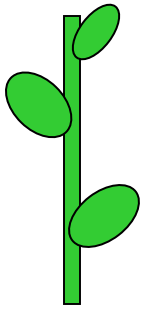
(tall) (dwarf)



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…

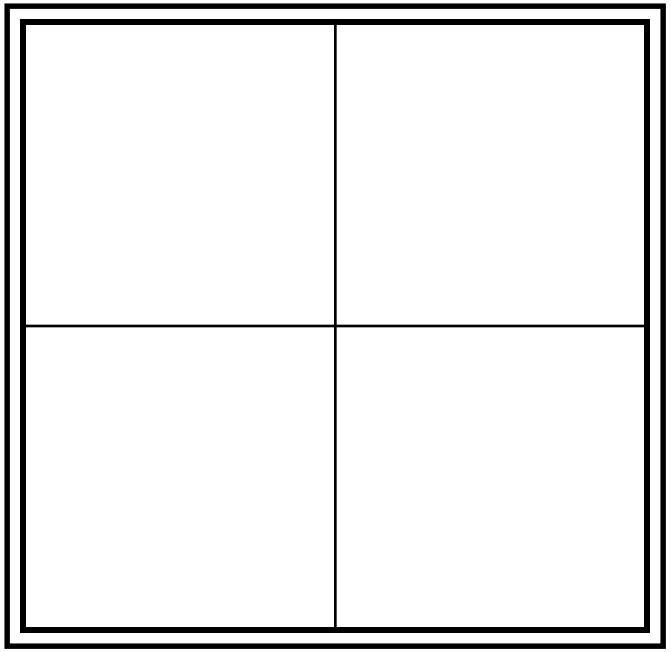
  We use the

  Punnett square

  to predict the

  genotypes and phenotypes of

  the offspring.



Using a Punnett Square

STEPS:

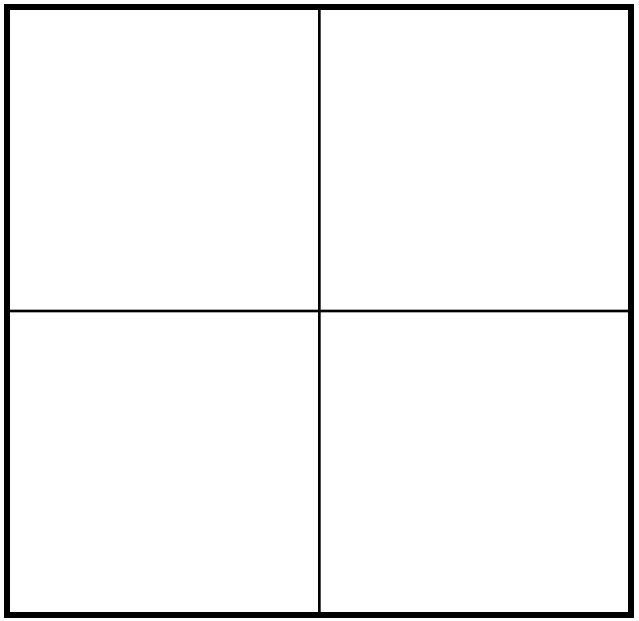
1. determine the genotypes of the parent organisms
2. write down your "cross" (mating)
3. draw a p-square

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

Monohybrid cross: F_2 generation

- If you let the F1 generation self-fertilize, the next monohybrid cross would be:

T *t* × **T *t***

(tall) (tall)

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

Genotypes:

1 TT= Tall

2 T*t* = Tall

1 *tt* = dwarf

Genotypic ratio= 1:2:1

Phenotype:

3 Tall

1 dwarf

Phenotypic ratio= 3:1

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 **T T** × ***t t***

The gametes are:

Once you have the gametes…

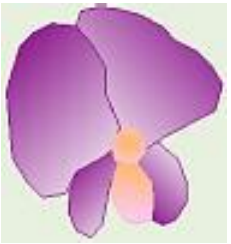
T **T** × ***t*** ***t***

	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*

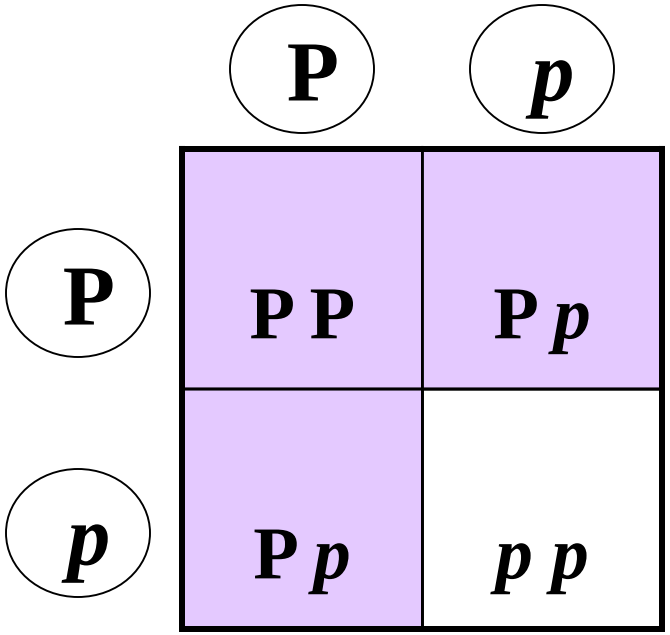
↓

P *p*

ALL PURPLE (P*p*)

Cross the F1 generation:

P *p* × P *p*

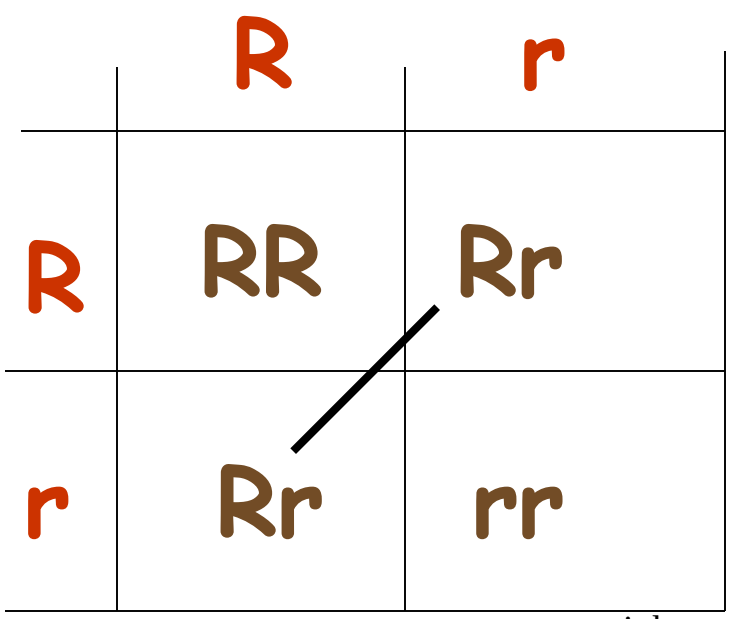


Genotypes:
1 PP
2 Pp
1 pp

Phenotypes:
3 Purple
1 White

F_1 Monohybrid Cross

- **Trait: Seed Shape**
- **Alleles: R – Round r – Wrinkled**
- **Cross: Round seeds x Round seeds**
- **Rr x Rr**



Genotype: RR, Rr, rr

Phenotype: Round & wrinkled

G.Ratio: 1:2:1

P.Ratio: 3:1

What Do the Peas Look Like?

Dominance and Recessiveness

- In all the experiments mendel observed that, one of the each pair of contrasting characters appeared in F1 hybrids
- He called the one which appeared the dominant trait, and the one which did not as the recessive trait.
- A dominant character is denoted by a capital letter and its recessive is represented by the simple letter

Mendel's Principles

- **<u>1. Principle of Dominance</u>**:

  One allele masked another, one allele was dominant over the other in the F_1 generation.

- **<u>2. Principle of Segregation</u>**:

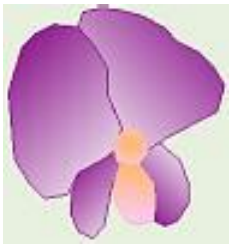
  When gametes are formed, the pairs of hereditary factors (genes) become separated, so that each sex cell (egg/sperm) receives only one kind of gene.

Dihybrid crosses

- Matings that involve parents that differ in **<u>two</u>** genes (two independent traits)

For example, flower color:

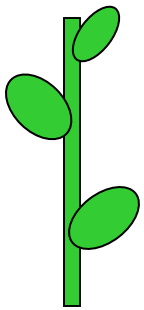
P = purple (dominant)



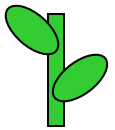
p = white (recessive)

and stem length:

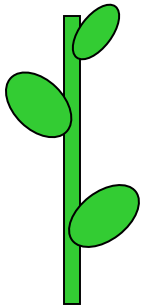
T = tall



t = short

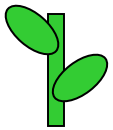


Dihybrid cross: flower color and stem length




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:

T*t* P*p* × T*t* P*p*

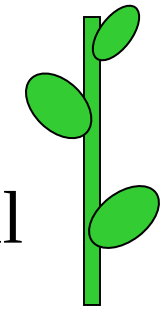
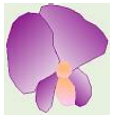
(tall, purple) (tall, purple)

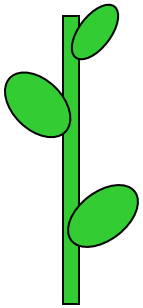
Possible gametes:
TP T*p* *t*P *tp*

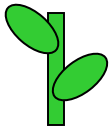
	TP	T*p*	*t*P	*tp*
TP	TTPP	TTP*p*	T*t*PP	T*t*P*p*
T*p*	TTP*p*	TT*pp*	T*t*P*p*	T*tpp*
*t*P	T*t*PP	T*t*P*p*	*tt*PP	*tt*P*p*
tp	T*t*P*p*	T*tpp*	*tt*P*p*	*ttpp*

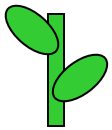
Four phenotypes observed
Tall, purple (9); Tall, white (3); Short, purple (3); Short white (1)

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*

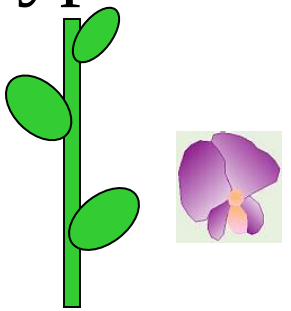
Phenotype Ratio = 9:3:3:1

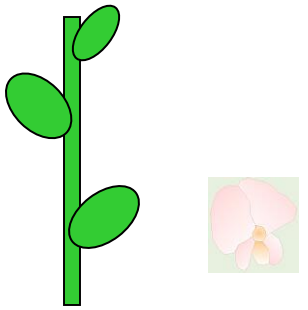
Dihybrid cross: 9 genotypes

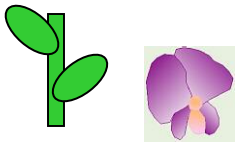
Genotype ratios (9):

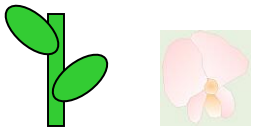
Four Phenotypes:

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)









Principle of Independent Assortment

- Based on these results, Mendel postulated the **3. Principle of Independent Assortment**:

  "Members of one gene pair segregate independently from other gene pairs during gamete formation"

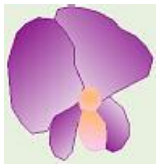
Genes get shuffled – these many combinations are one of the advantages of sexual reproduction

Test cross

When you have an individual with an unknown genotype, you do a **test cross.**

Test cross: Cross with a homozygous recessive individual.

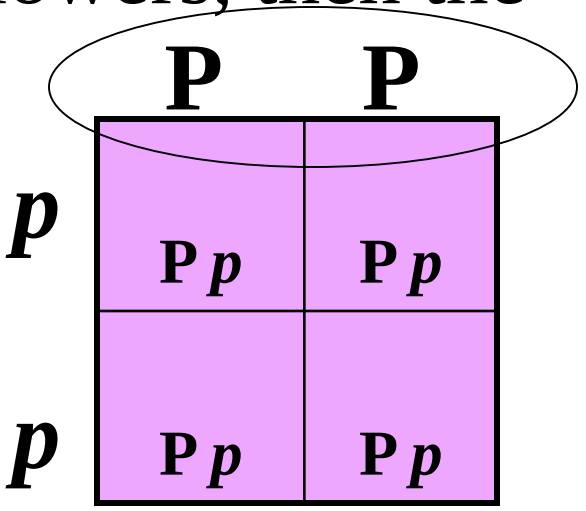
For example, a plant with **purple** flowers can either be **PP** or **P*p***… therefore, you cross the plant with a *pp* (white flowers, homozygous recessive)



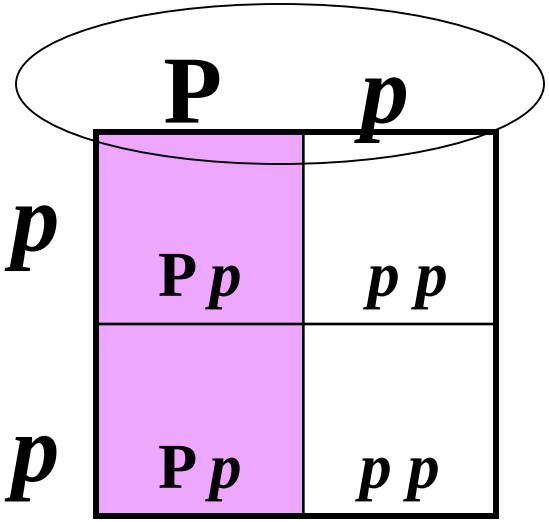
P ? × *pp*

Test cross

- If you get all 100% purple flowers, then the unknown parent was PP…



- If you get 50% white, 50% purple flowers, then the unknown parent was P*p*…

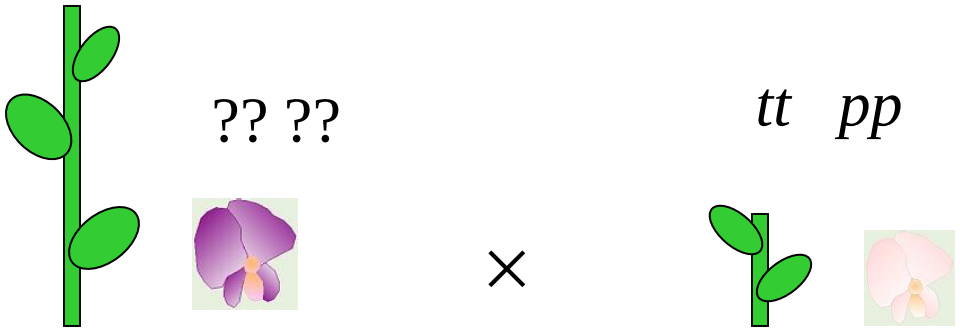


Back cross vs. test cross

- The mating between the F2 progeny back to one of its parents is termed backcross
- Ex: Tt x TT
- Tt x tt

Dihybrid test cross??

If you had a tall, purple plant, how would you know what genotype it is?



1. TTPP
2. TTP*p*
3. T*t*PP
4. T*t*P*p*

Summary of Genetics

- Chromosomes carry hereditary info (genes)
- Chromosomes (and genes) occur in pairs
- New combinations of genes occur in sexual reproduction
- Monohybrid vs. Dihybrid crosses
- Mendel's Principles:
  - Dominance: one allele masks another
  - Segregation: genes become separated in gamete formation
  - Independent Assortment: Members of one gene pair segregate independently from other gene pairs during gamete formation