LECTURE 7 : GENETICS

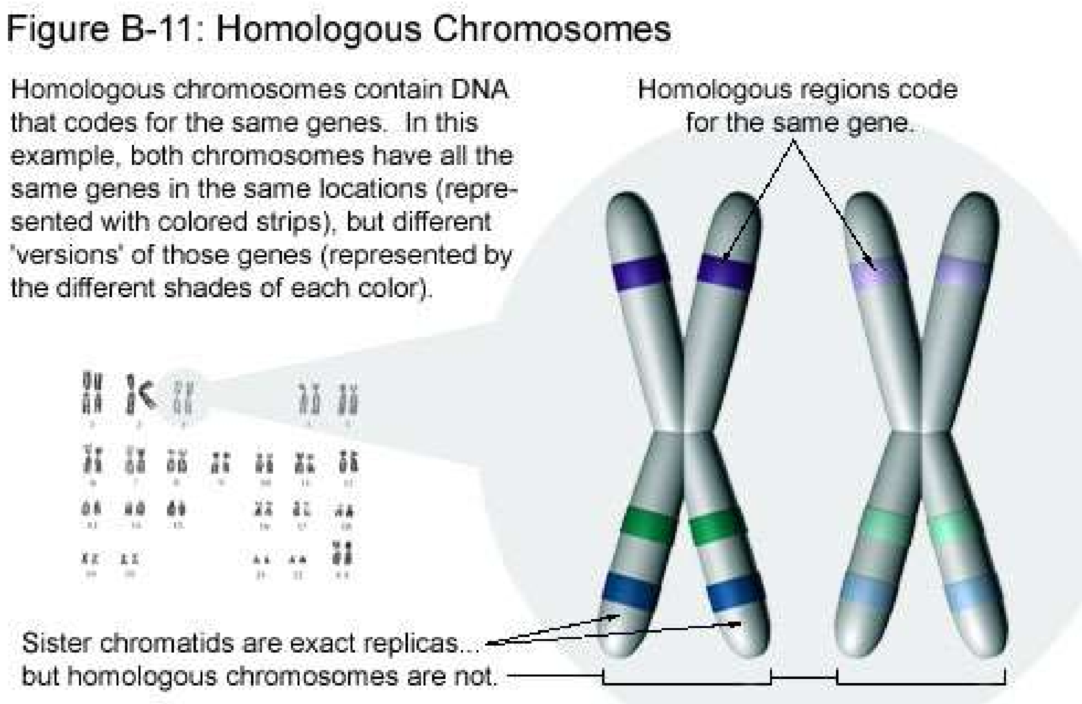
- Introduction to Genetics and heredity
- Gregor Mendel – a brief bio
- Genetic terminology (glossary)
- Monohybrid crosses
- Patterns of inheritance
- Dihybrid crosses
- Test cross
- Beyond Mendelian Genetics – incomplete dominance

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



Warm-Up

- You need something to write with for today’s class

Gregor Mendel

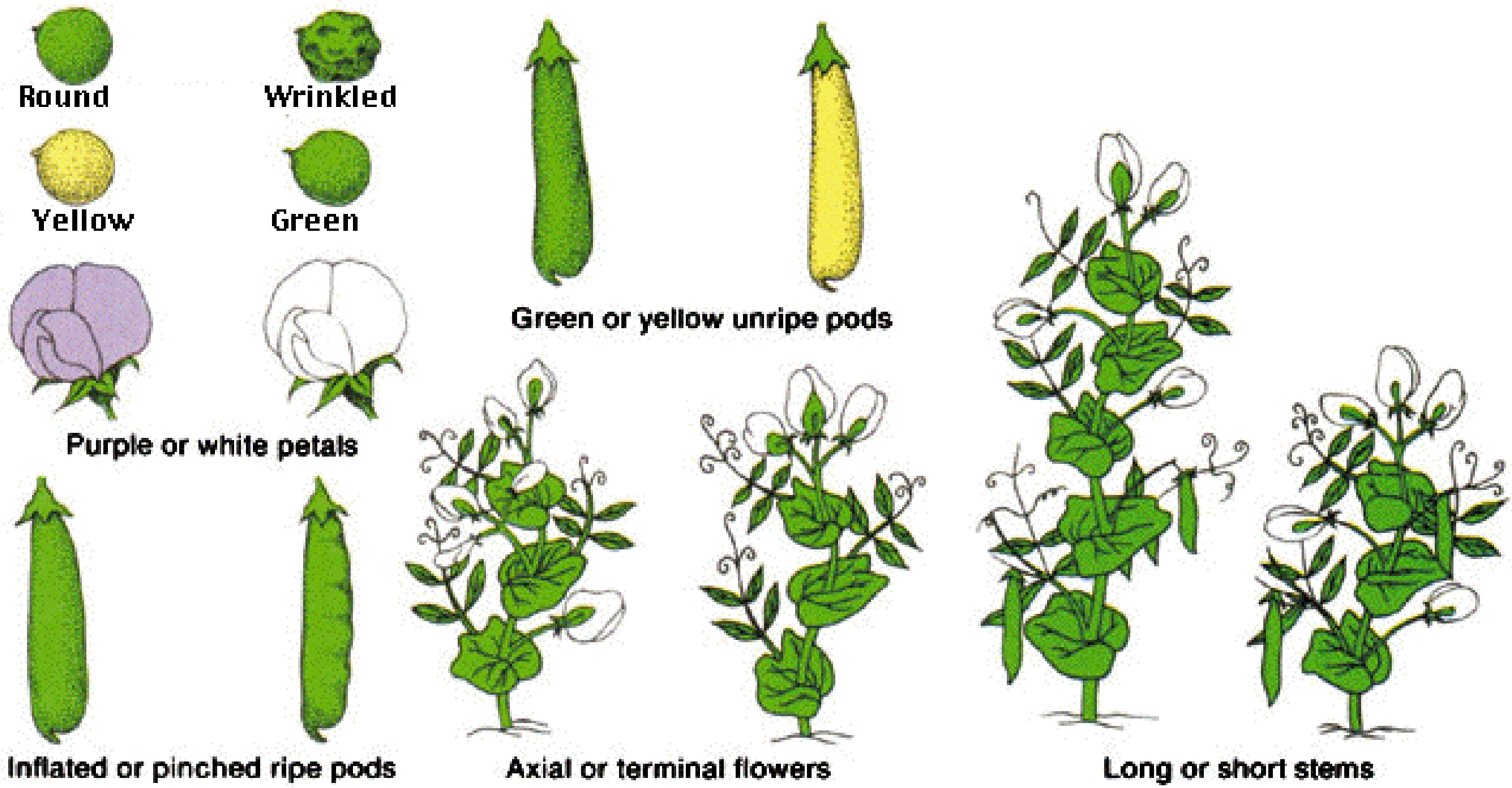
- Austrian Monk, born in what is now Czech Republic in 1822
- Son of peasant farmer, studied Theology and was ordained priest Order St. Augustine.

Gregor Mendel

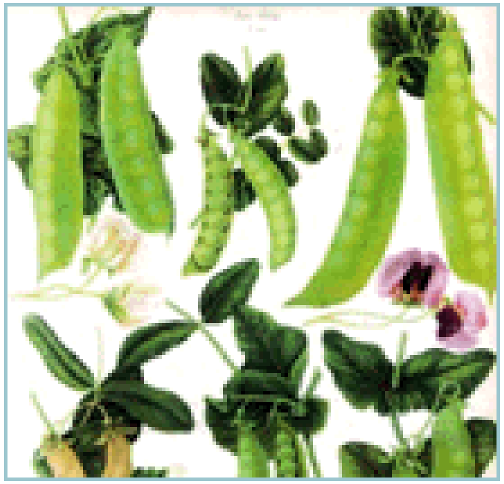
- Went to the university of Vienna, where he studied botany and learned the Scientific Method
- Worked with pure lines of peas for eight years
- Prior to Mendel, heredity was regarded as a "blending" process and the offspring were essentially a "dilution"of the different parental characteristics.

Mendel’s peas

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



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



The concept of genes

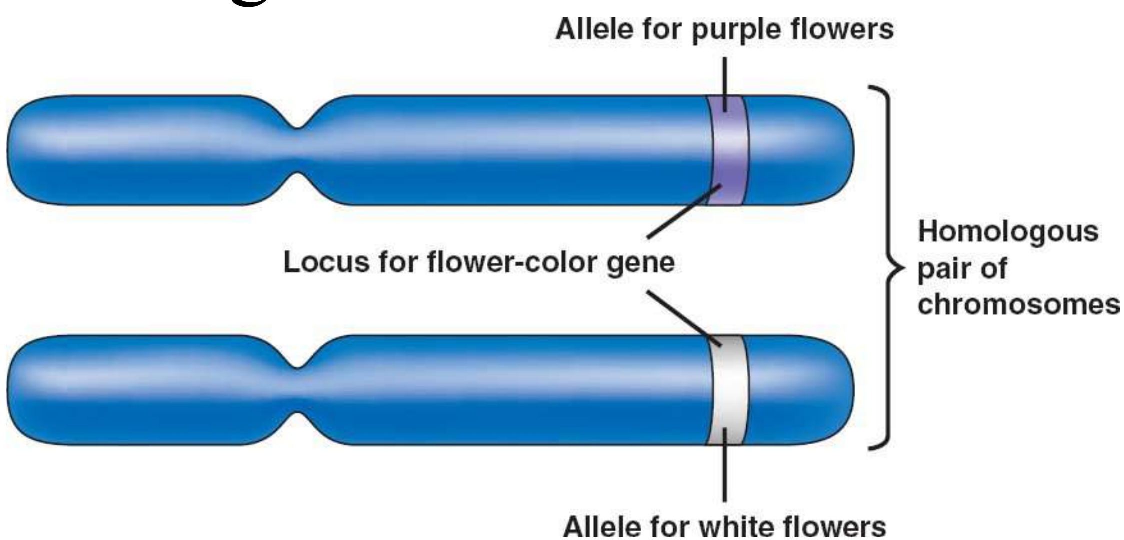
That genes occur in pairs

That one gene of each pair is present in the gametes

Considered the Father of Genetics

Genetics terms you need to know:

- **Heredity** – passing of traits from
parent to offspring
- **Alleles** – the different forms of a trait that make up a gene pair
- **Genetics-**the study of how traits are inherited through interactions of alleles



- **Homozygous** – having same genes (one from each parent) for a particular characteristic.

Ex: AA, aa, BB, bb

- **Heterozygous** – having two different genes for a particular characteristic.

Ex: Aa, Bb, Cc

- **Genotype** – the genetic makeup of an organism

(GENES)

- **Phenotype**- the physical appearance of an organism (what the organism looks like)

- **Dominant** – the allele of a gene that dominates or suppresses the expression of an allele
- **Recessive** – an allele that is covered up by a dominant allele;
- **Punnett Square-** a handy tool used to predict the phenotype and genotype of offspring
  - Upper case letter = dominant (A)
  - Lower case letter= recessive (a)

  - **Hybrid-** the result of mixing, through sexual reproduction, two animals or plants of different breeds, varieties or species. **AKA half breed**

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 (Dominant)

t = allele for dwarf (recessive)

TT = homozygous tall plant

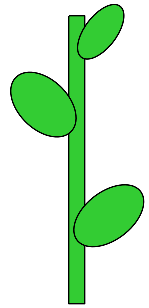
t t = homozygous dwarf plant

Long or short stems

T T × t t

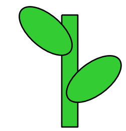
Monohybrid cross for stem length:

P = parentals
true breeding,
homozygous plants:



T T × *t t*

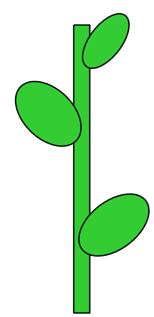
(tall) (dwarf)



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…

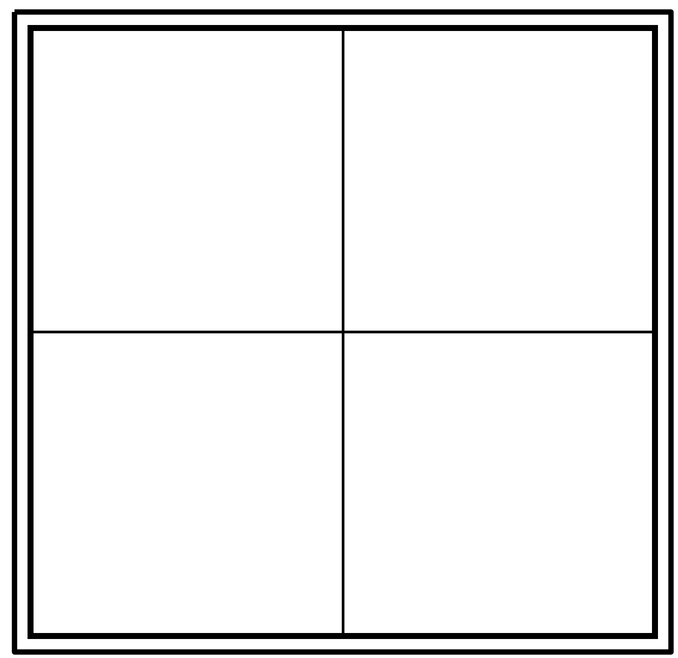
We use the

Punnett square

to predict the

genotypes and phenotypes of

the offspring.



Using a Punnett Square

STEPS:

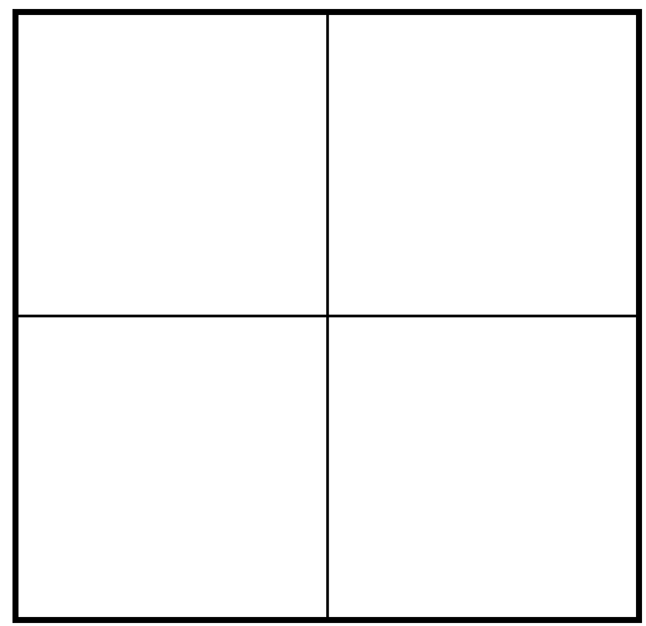
1. determine the genotypes of the parent organisms
2. write down your "cross" (mating)
3. draw a p-square

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:

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

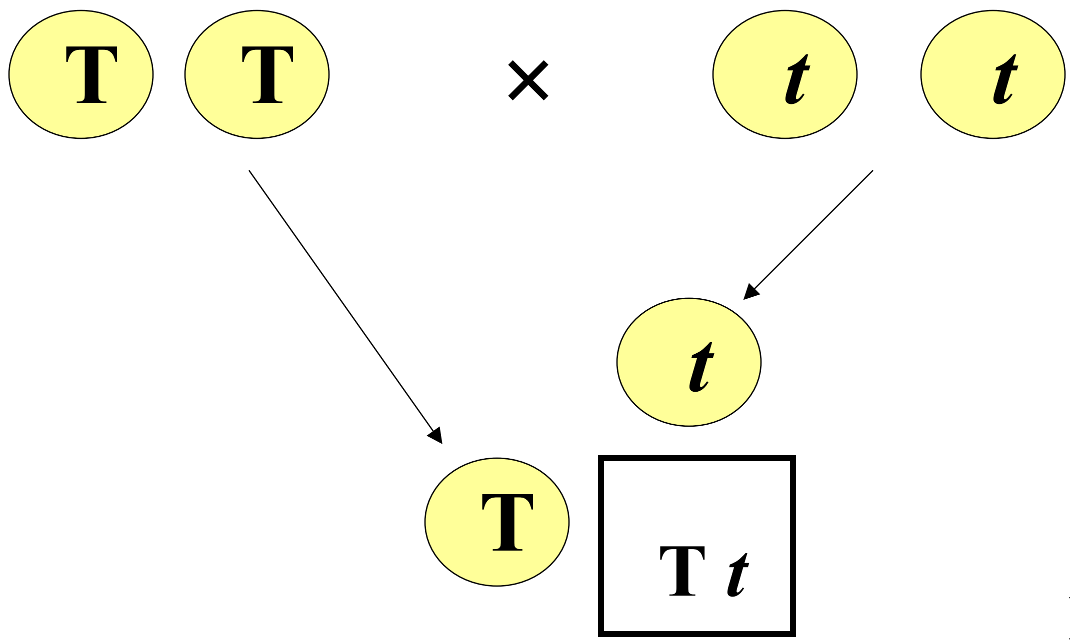
Once you have the gametes…

T **T** × ***t*** ***t***

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

Shortcut for Punnett Square…

If either parent is HOMOZYGOUS



Genotypes:
100% T t

Phenotypes:
100% Tall plants

- **You only need <u>one</u> box!**

Understanding the shortcut…

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

=

	t
T	**T *t***

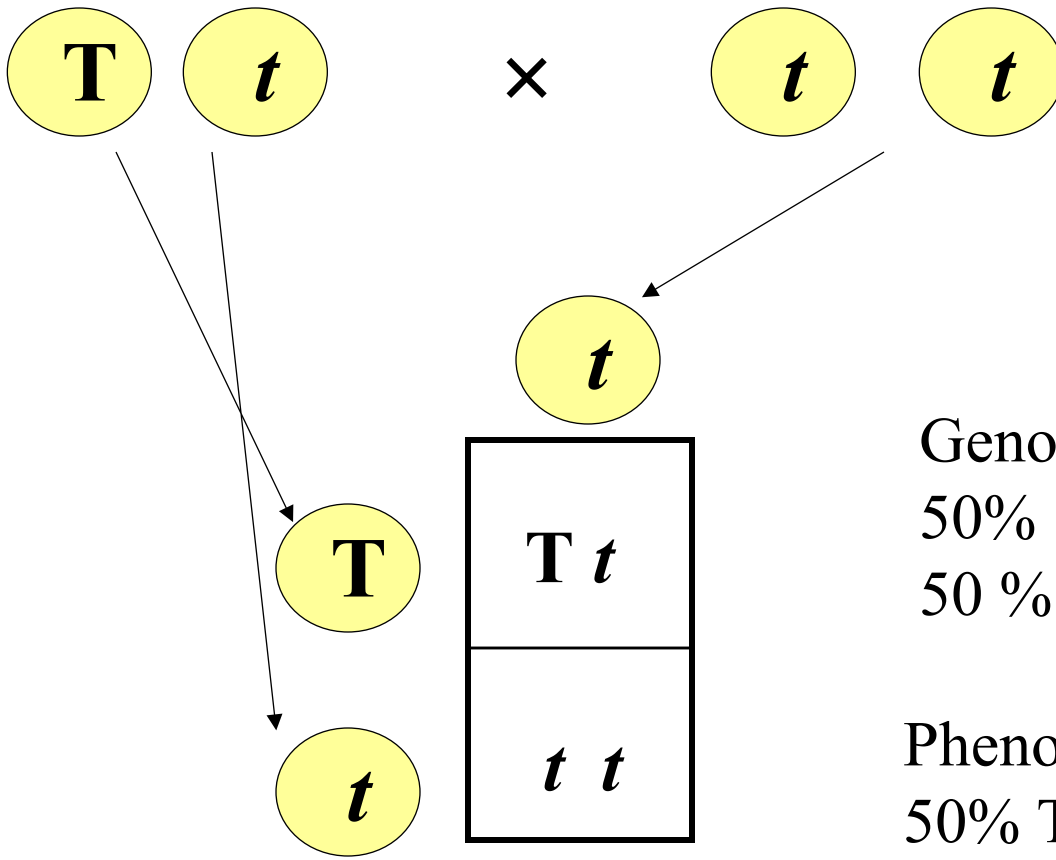
Genotypes:
100% T t

Phenotypes:
100% Tall plants

If you have another cross…

- A heterozygous with a homozygous

You can still use the shortcut!



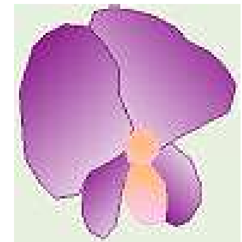
Genotypes:
50% T t
50 % *t t*

Phenotypes:
50% Tall plants
50% Dwarf plants

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

- **2. Principle of Segregation**:

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.

Human case: CF

- Mendel's Principles of Heredity apply universally to all organisms.
- Cystic Fibrosis: a lethal genetic disease affecting 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 = normal
C *c* = carrier, no symptoms
c *c* = has cystic fibrosis

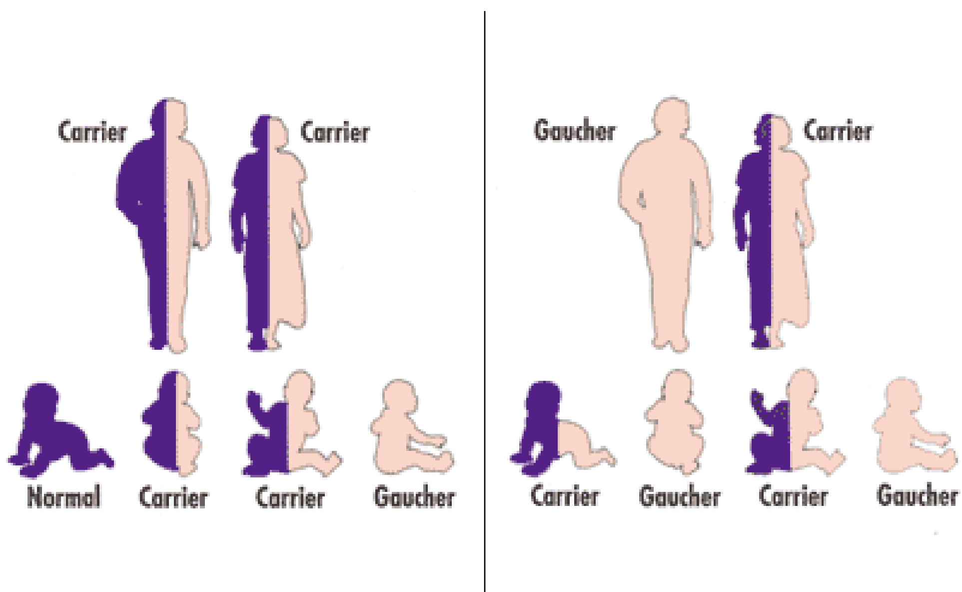
	C	***c***
C	**C C**	**C *c***
c	**C *c***	***c c***

Probabilities…

- Of course, the 1 in 4 probability of getting the disease is just an **expectation**, and in reality, any two carriers may have normal children.
- However, the greatest probability is for 1 in 4 children to be affected.
- Important factor when prospective parents are concerned about their chances of having affected children.
- Now, 1 in 29 Americans is a symptom-less carrier (Cf *cf*) of the gene.

Gaucher Disease

- **Gaucher Disease** is a rare, genetic disease. It causes lipid-storage disorder (lipids accumulate in spleen, liver, bone marrow)
- It is the most common genetic **disease** affecting Jewish people of Eastern European ancestry (1 in 500 incidence; rest of pop. 1 in 100,000)

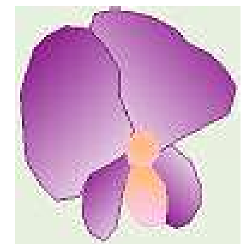


Dihybrid crosses

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

For example, flower color:

P = purple (dominant)

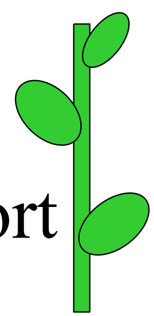


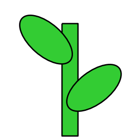
p = white (recessive)

and stem length:

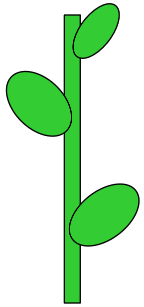
T = tall

t = short



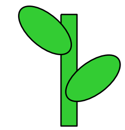


Dihybrid cross: flower color and stem length

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: flower color and stem length (shortcut)

TT PP × *tt pp*

(tall, purple) (short, white)

Possible Gametes for parents

T P

t p

	T P
t 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:

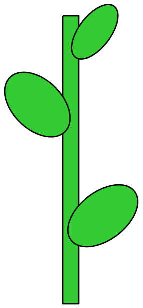
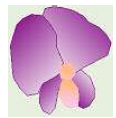
TP T*p* *t*P *tp*

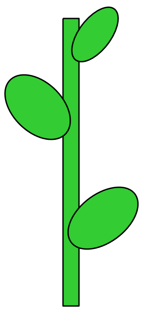
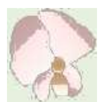
	TP	T*p*	*t*P	*tp*
TP	TTPP	TTP*p*	T*t*PP	T*t*P*p*
T*p*	TTP*p*	TT*pp*	T*t*P*p*	T*tpp*
*t*P	T*t*PP	T*t*P*p*	*tt*PP	*tt*P*p*
tp	T*t*P*p*	T*tpp*	*tt*P*p*	*ttpp*

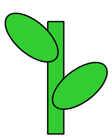
Four phenotypes observed

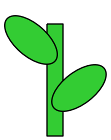
Tall, purple (9); Tall, white (3); Short, purple (3); Short white (1)

Dihybrid cross

9 Tall  purple 

3 Tall  white 

3 Short  purple 

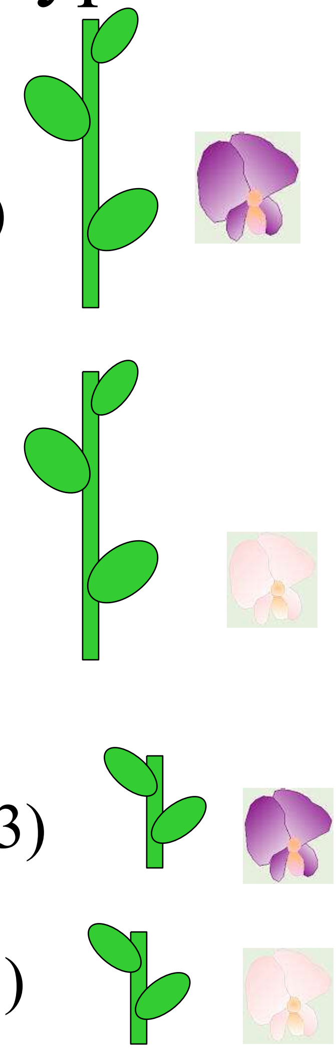
1 Short  white 

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



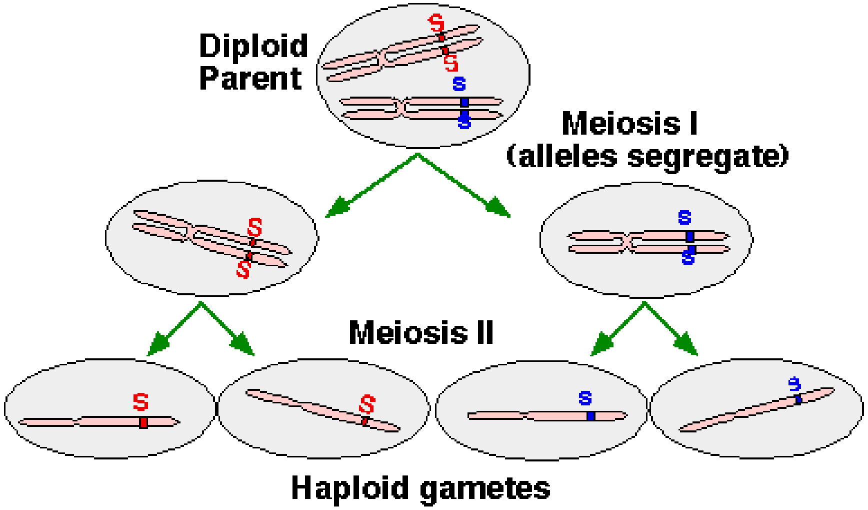
Principle of Independent Assortment

- 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

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

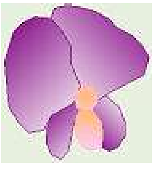


Test cross

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…

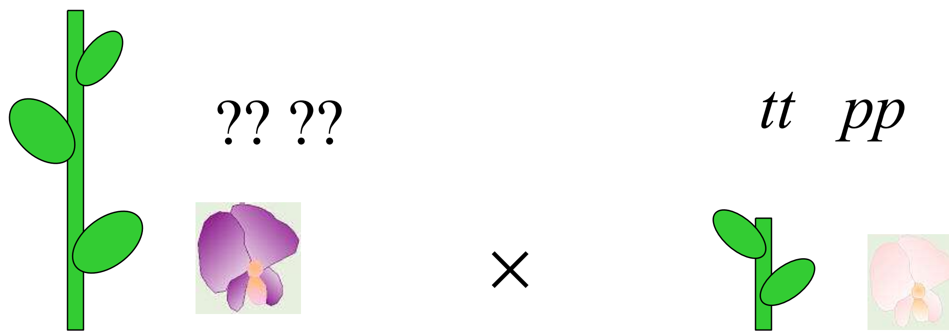
	P	P
p	P*p*	P*p*
p	P*p*	P*p*

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

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

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*

Beyond Mendelian Genetics: Incomplete Dominance

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.

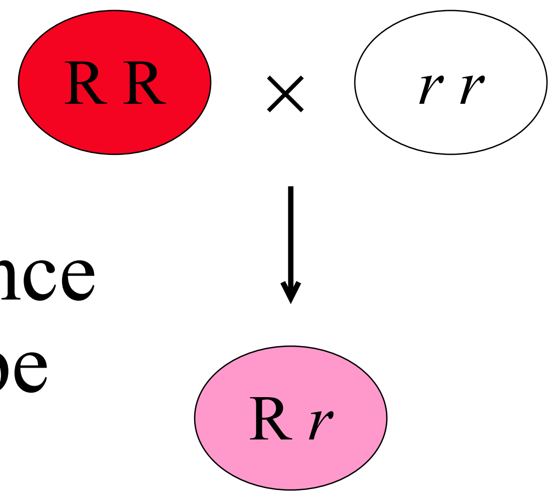
But sometimes phenotypes are not very obvious…

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)!



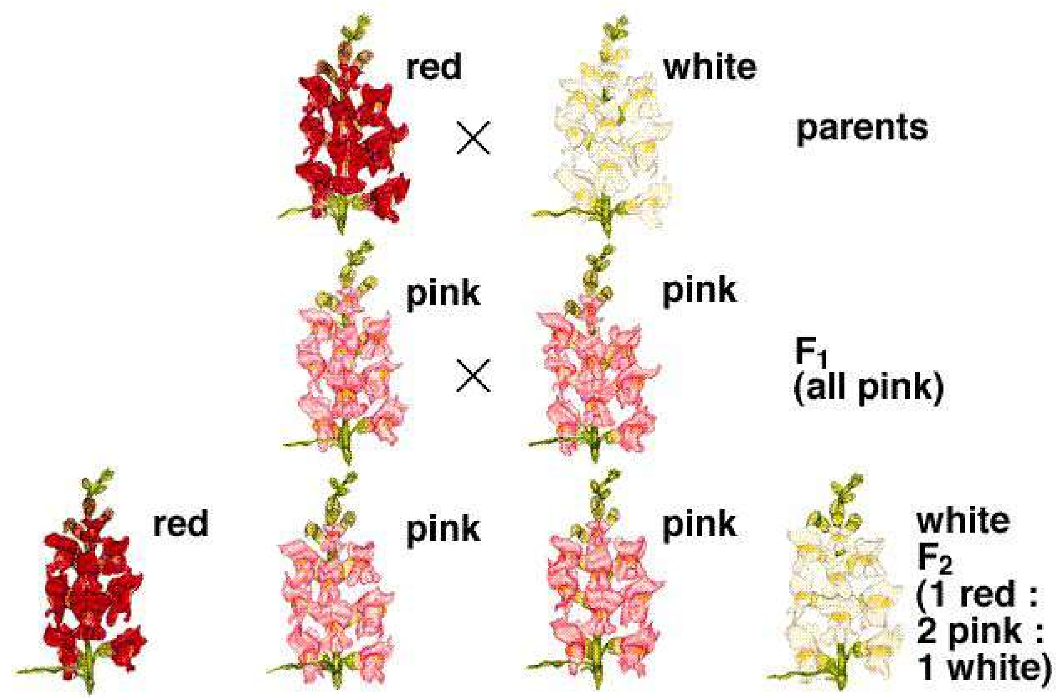
Genes show incomplete dominance when the heterozygous phenotype is intermediate.

Incomplete dominance

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

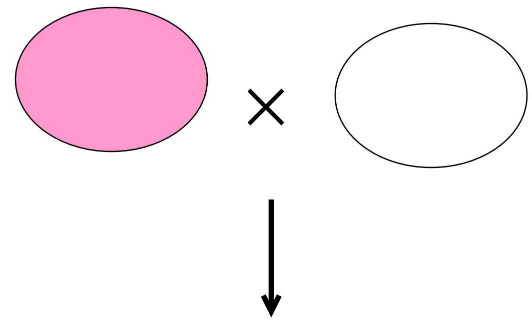
	R	***r***
R	**R R**	**R *r***
r	**R *r***	***r r***

Incomplete Dominance



Incomplete dominance

What happens if you cross a pink with a white?



A pink with a red?