Introduction to Genetics

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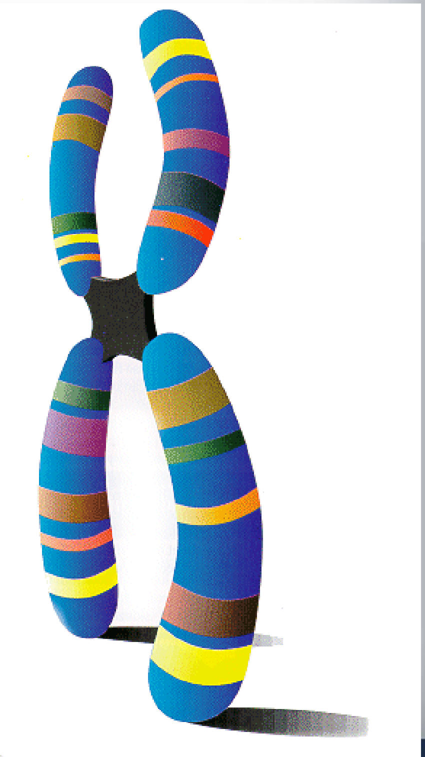
GENETICS

- 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

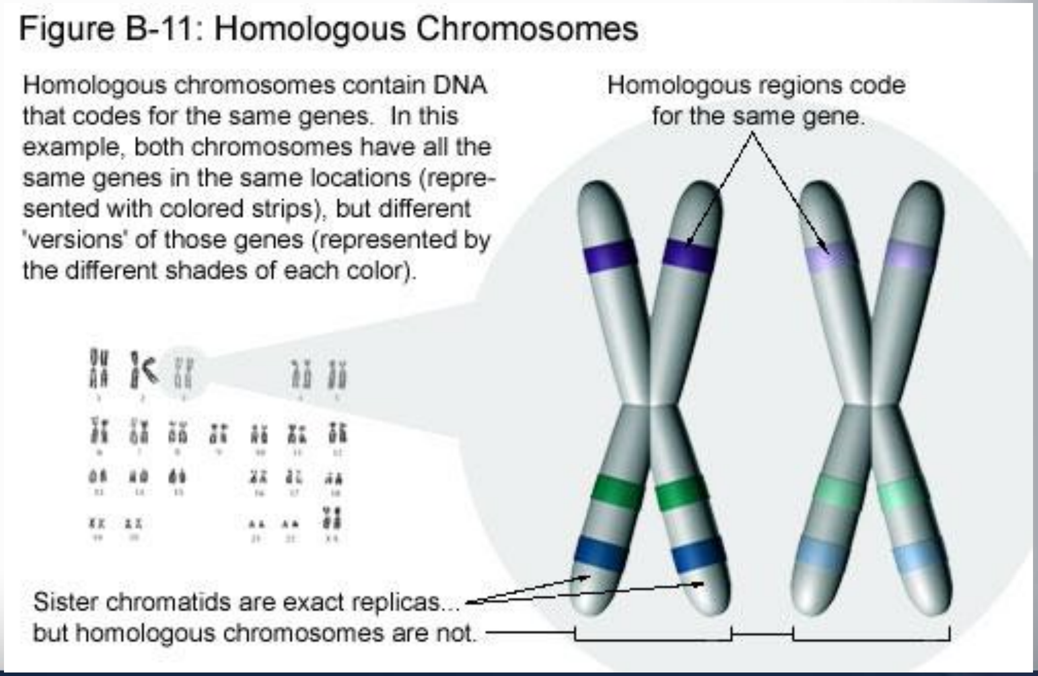
Introduction to Genetics

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

- **Chromosomes** carry the hereditary inf (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



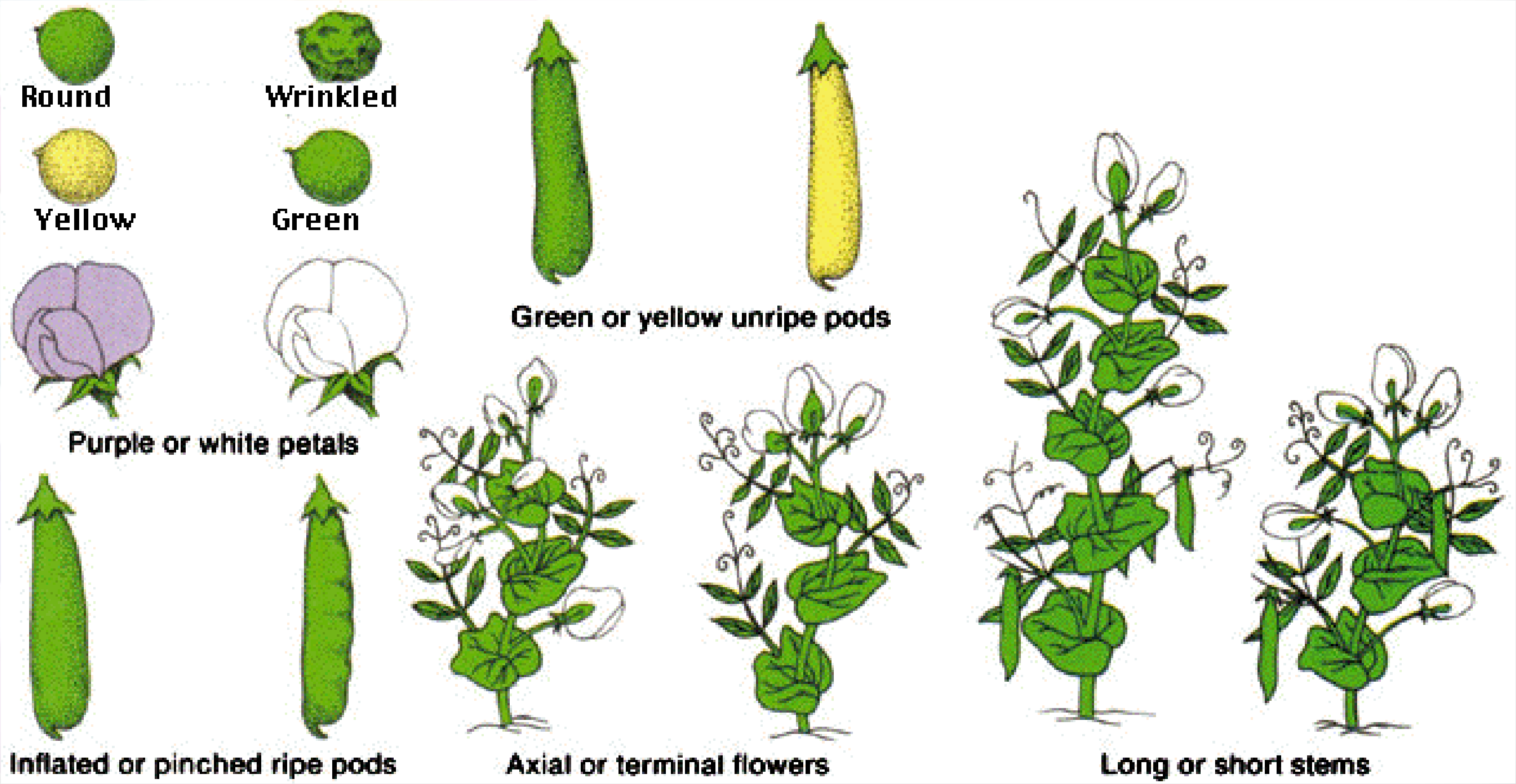
Gregor Johann Mendel

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.
- 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 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.

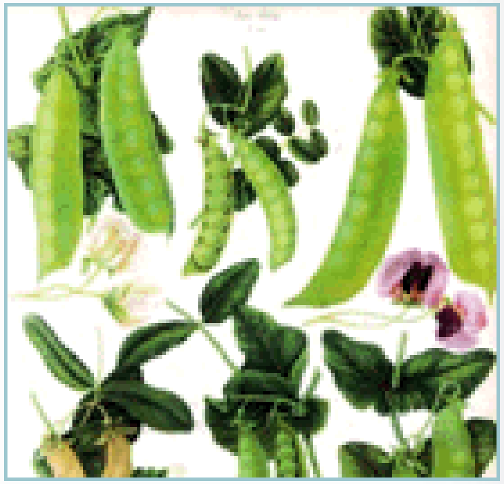
Mendel's Works

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

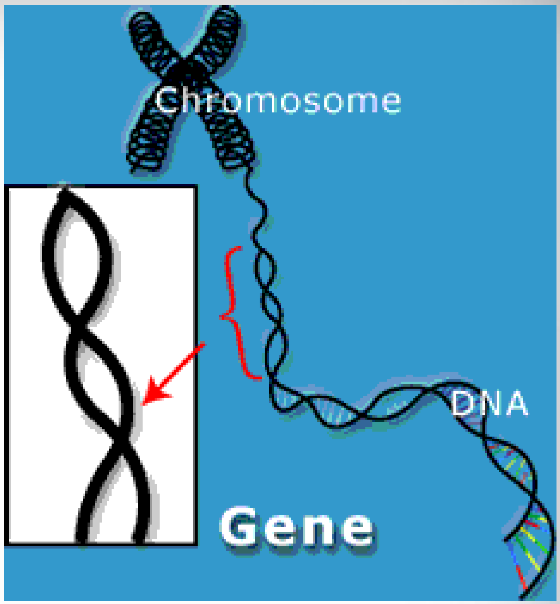
  The concept of genes

  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.

Genetics terms you need to know:

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

Genetics terms you need to know:

- **<u>Genotype</u>** – the genetic makeup of an organisms
- **<u>Phenotype</u>** – 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
- **F_1** = First filial generation; offspring from a genetic cross.
- **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

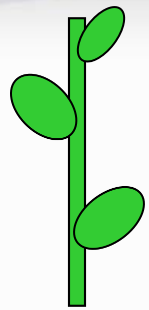
t = allele for dwarf

TT = homozygous tall plant

t t = homozygous dwarf plant

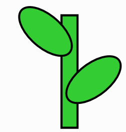
T T $\times$ t t

Monohybrid cross for stem length:



T T × *t t*

(tall) (dwarf)



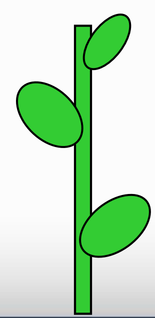
P = parentals
true breeding,
homozygous plants:

↓

F_1 generation
is heterozygous:

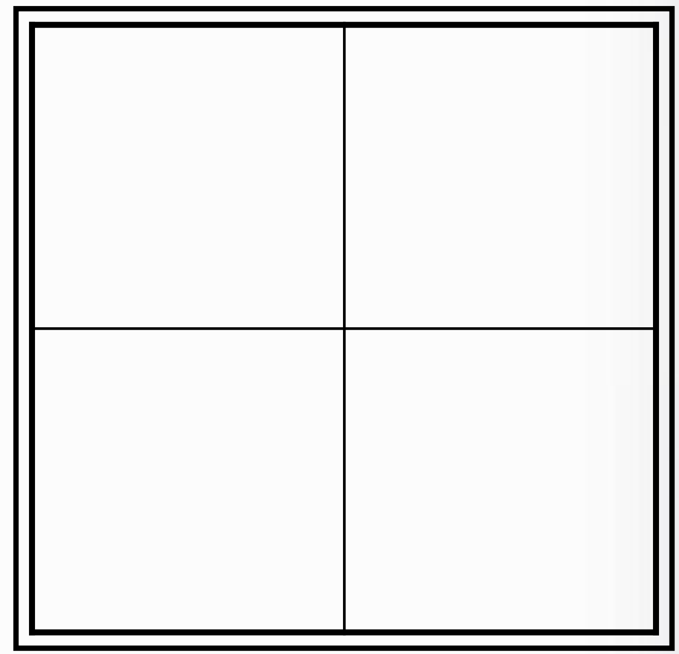
T *t*

(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:

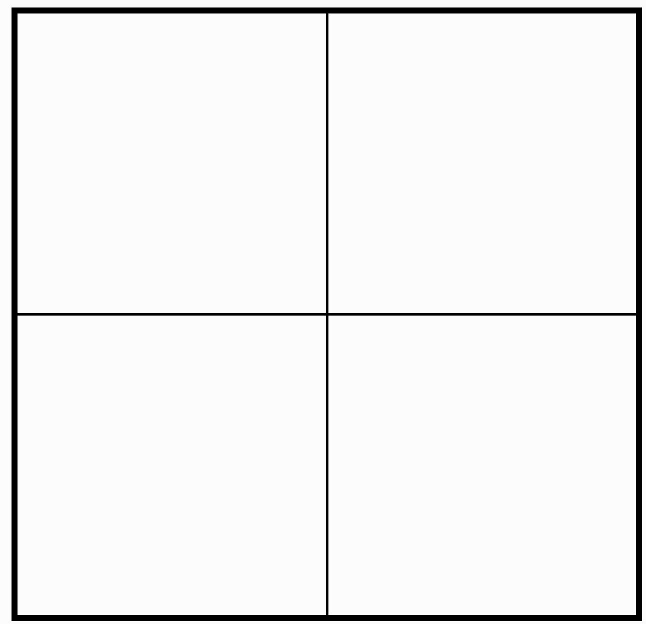
1. determine the genotypes of the parent organisms
2. write down your "cross" (mating)
3. draw a p-square

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

Sample Problem

- Black is dominant (B) and red is recessive (b). A homozygous black seed is crossed by heterozygous seeds. Do the monohybrid cross.

Seatwork

- Green (G) is dominant over yellow (g). If a homozygous green parent is crossed with heterozygous parent. Do the monohybrid cross.

- Genotypes:
- Genotypic Ratio:
- Genotypic %:
- Phenotypes:
- Phenotypic Ratio:
- Phenotypic %:

Seatwork

Round (D) is dominant over wrinkled (d). Do a monohybrid cross between the homozygous dominant parents.

- Genotypes:
- Ratio:
- Genotypic Ratio:
- Phenotypes:
- Ratio:
- Phenotypic Ratio:

Seatwork

Pink (X) flowers are dominant while red (x) flowers are recessive. Do a monohybrid cross of both heterozygous parents.

- Genotypes:
- Ratio:
- Genotypic Ratio:
- Phenotypes:
- Ratio:
- Phenotypic Ratio:

Seatwork

Homozygous tall (T) parent is crossed with homozygous short (t) parents. Do a monohybrid cross.

- Genotypes:
- Ratio:
- Genotypic Ratio:
- Phenotypes:
- Ratio:
- Phenotypic Ratio:

Punnett square

B B × *B b*

	B	**B**
B	**BB**	**BB**
b	**Bb**	**Bb**

Genotypes:
50% BB; 50% Bb

Phenotypes:
100% black seeds

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***

<u>Genotypes:</u>
1 TT= Tall
2 T*t* = Tall
1 *tt* = dwarf
Genotypic ratio= 1:2:1

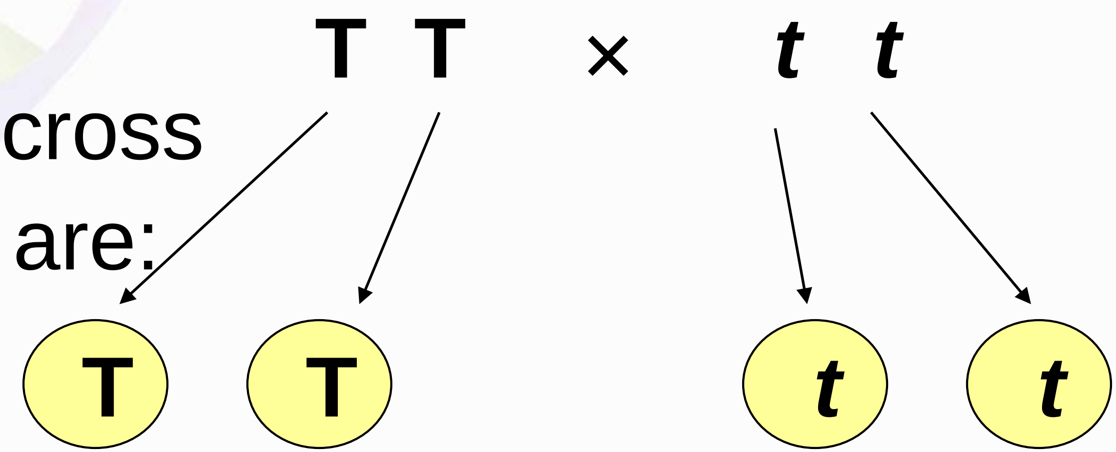
<u>Phenotype:</u>
3 Tall
1 dwarf
Phenotypic ratio= 3:1

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

The gametes are:



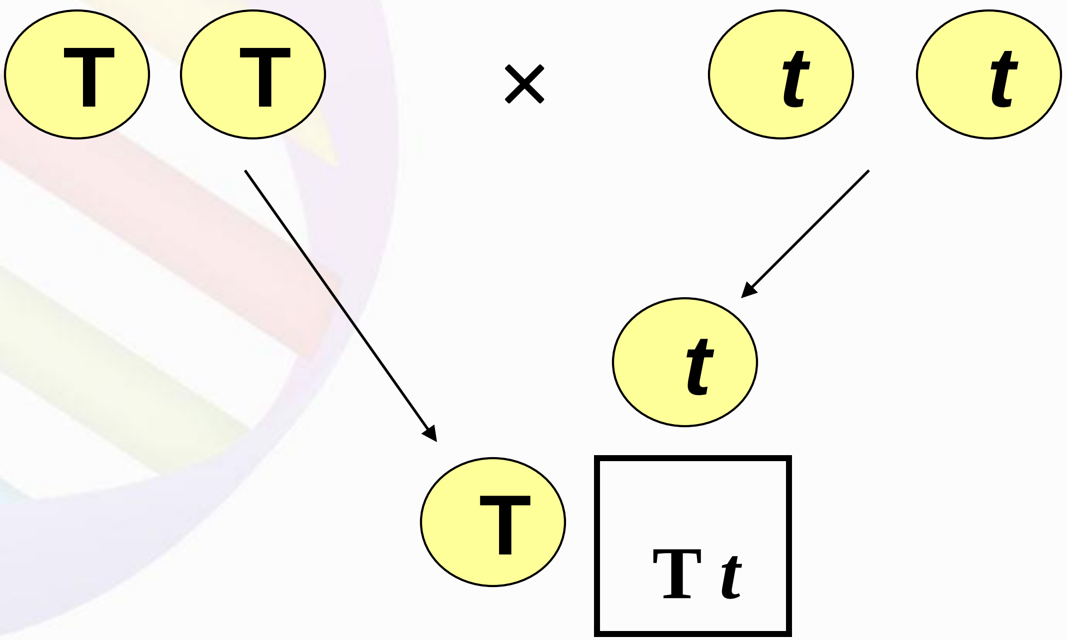
Once you have the gametes…

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

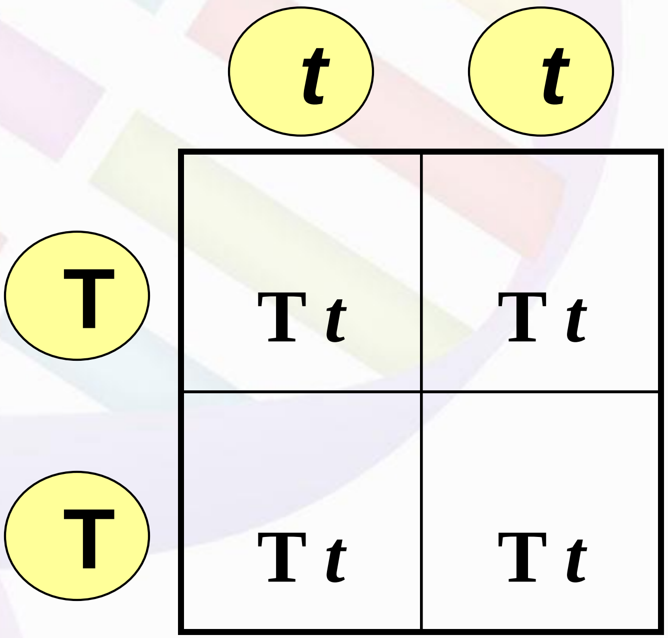


Genotypes:
100% T t

Phenotypes:
100% Tall plants

- **You only need one box!**

Understanding the shortcut…



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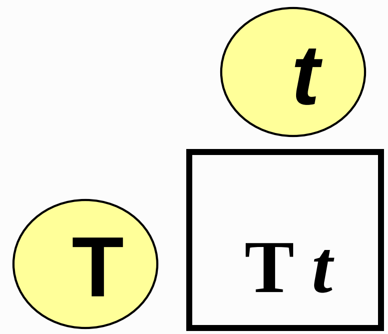


Genotypes:
100% T t

Phenotypes:
100% Tall plants

If you have another cross…

- A heterozygous with a homozygous

T *t* × *t* *t*

	t
T	T*t*
t	*t t*

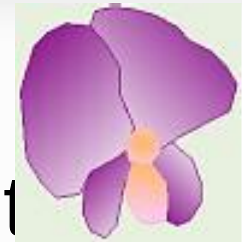
You can still use the shortcut!

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

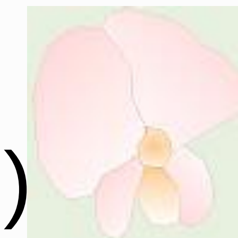
Phenotypes:
50% Tall plants
50% Dwarf plants

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**:

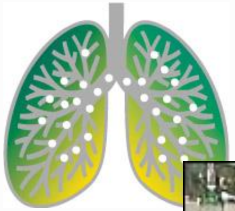
  One allele masked another, one allele was dominant over the other in the F_1 generation.

- **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

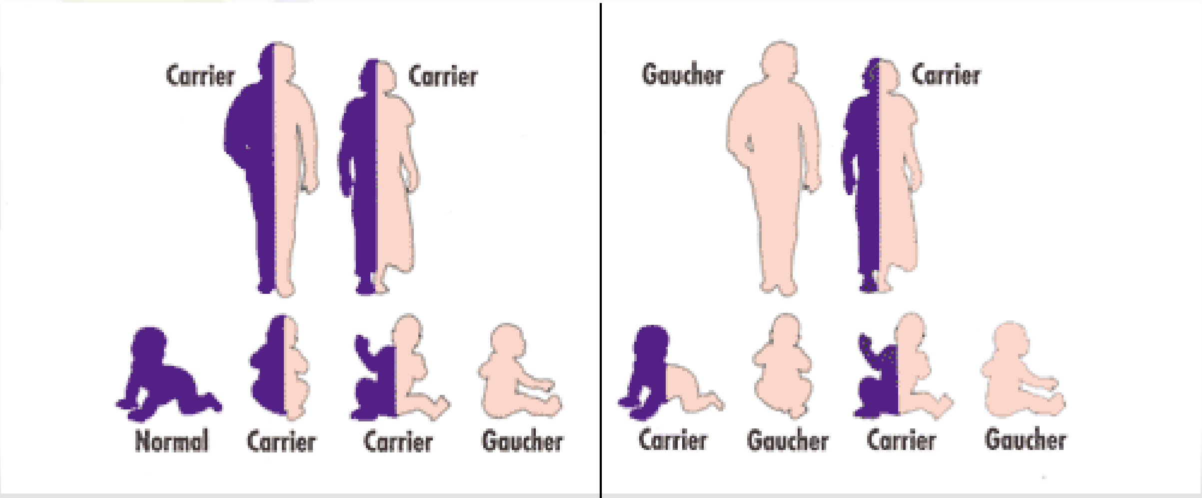
	C	*c*
C	C C	C *c*
c	C *c*	*c c*

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

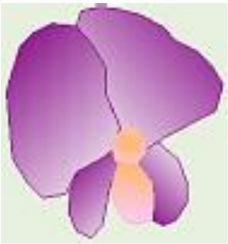
- **Gaucher Disease** is a rare, genetic disease. It causes lipid-storage disorder (lipids accumulate in spleen, liver, bone marrow)
- It is the most common genetic **disease** affecting Jewish people of Eastern European ancestry (1 in 500 incidence; rest of pop. 1 in 100,000)



Dihybrid crosses

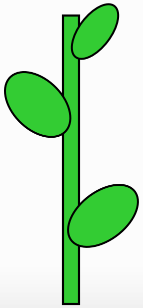
- Matings that involve parents that differ in **<u>two</u>** genes (two independent traits)

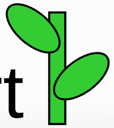
For example, flower color:

P = purple (dominant) 

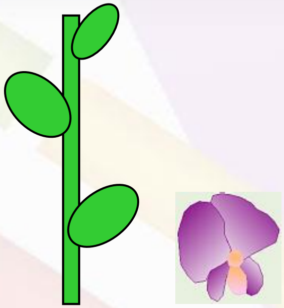
p = white (recessive) 

and stem length:

T = tall 

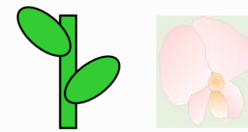
t = short 

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:

$$Tt\ Pp \times Tt\ Pp$$

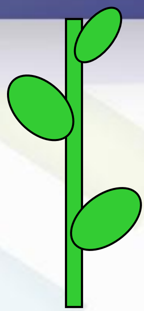
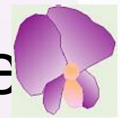
(tall, purple) (tall, purple)

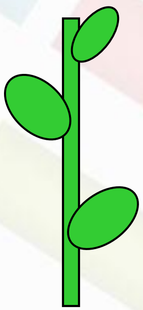
Possible gametes:
TP T*p* *t*P *tp*

	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*

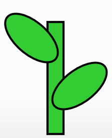
Four phenotypes observed
Tall, purple (9); Tall, white (3); Short, purple (3); Short white (1)

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:

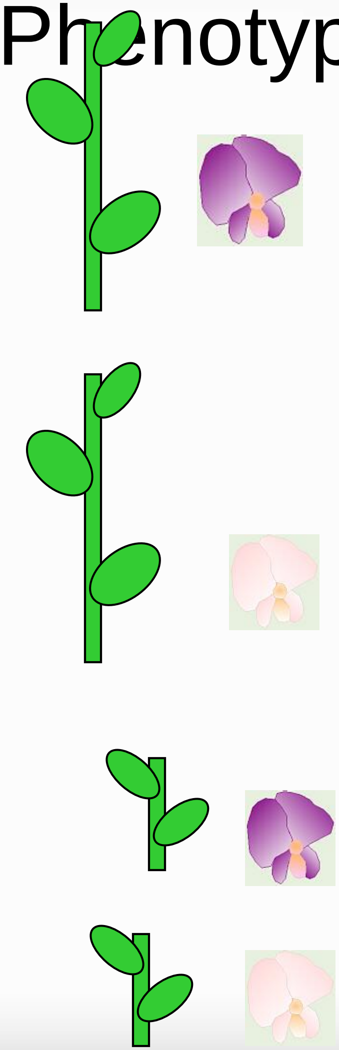
1 TTPP
2 TTP*p*
2 T*t*PP
4 T*t*P*p*
1 TT*pp*
2 T*tpp*
1 *tt*PP
2 *tt*P*p*
1 *ttpp*

Tall, purple (9)

Tall, white (3)

Short, purple (3)

Short, white (1)



Problem No. 1

- Do a dihybrid cross of the F1 generation which self-pollinated.

GGRr × GGRr

(green, round) (green, round)

Problem No. 2

- Do a dihybrid cross of the F1 generation which self-pollinated.

Llgg × Llgg

(long, yellow) (long, yellow)

Problem 1 Solution

GGRr × GGRr

(green, round) (green, round)

Possible gametes:
GR, GR, Gr, Gr

	GR	GR	Gr	Gr
GR	GGRR	GGRR	GGRr	GGRr
GR	GGRR	GGRR	GGRr	GGRr
Gr	GGRr	GGRr	GGrr	GGrr
Gr	GGRr	GGRr	GGrr	GGrr

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

Problem 1 Solution

Genotypes:

GGRR – 4

GGRr – 8

GGrr – 4

GR:4:8:4 or 1:2:1

Phenotypes: Green Round (12)

Green wrinkled (4)

PR: 12: 4 or 3:1

GGRR	GGRR	GGRr	GGRr
GGRR	GGRR	GGRr	GGRr
GGRr	GGRr	GGrr	GGrr
GGRr	GGRr	GGrr	GGrr

GGRR	GGRR	GGRr	GGRr
GGRR	GGRR	GGRr	GGRr
GGRr	GGRr	GGrr	GGrr
GGRr	GGRr	GGrr	GGrr

Problem 2 Solution

Llgg × Llgg

(long, yellow) (long, yellow)

Possible gametes:
Lg, Lg, lg, lg

	Lg	Lg	lg	lg
Lg	LLgg	LLgg	Llgg	Llgg
Lg	LLgg	LLgg	Llgg	Llgg
lg	Llgg	Llgg	llgg	llgg
lg	Llgg	Llgg	llgg	llgg

Problem 1 Solution

Genotypes:

LLgg – 4

Llgg – 8

Llgg – 4

GR:4:8:4 or 1:2:1

LLgg	LLgg	Llgg	Llgg
LLgg	LLgg	Llgg	Llgg
Llgg	Llgg	llgg	llgg
Llgg	Llgg	llgg	llgg

Phenotypes: Long yellow (12)

Short yellow (4)

PR: 12: 4 or 3:1

LLgg	LLgg	Llgg	Llgg
LLgg	LLgg	Llgg	Llgg
Llgg	Llgg	llgg	llgg
Llgg	Llgg	llgg	llgg

Principle of Independent Assortment

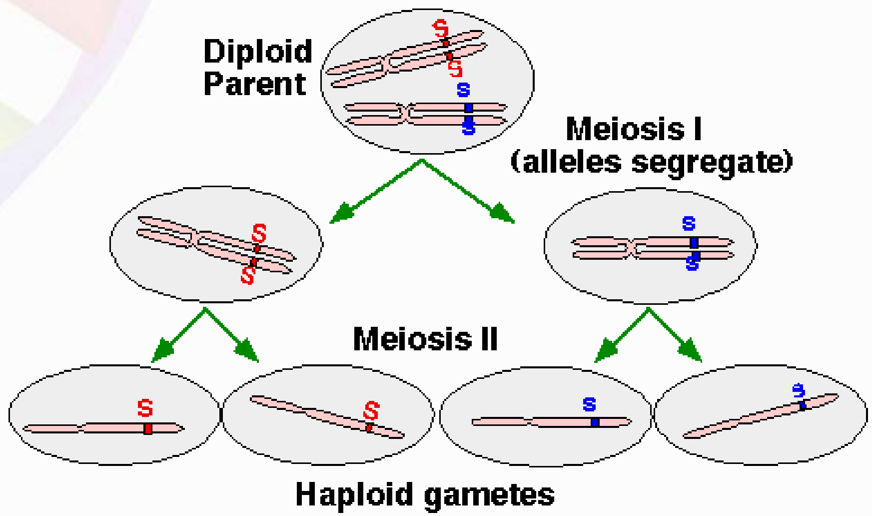
- Based on these results, Mendel postulated the **<u>3. Principle of Independent Assortment</u>**:

  "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



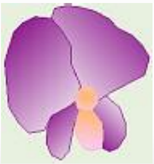
Test cross

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,

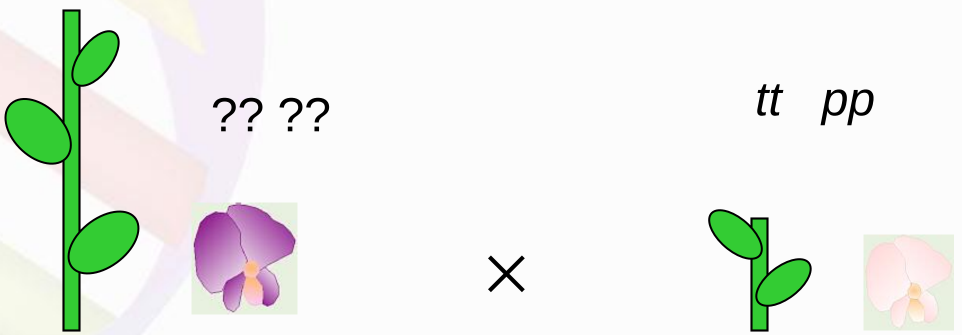
  then the unknown

  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.

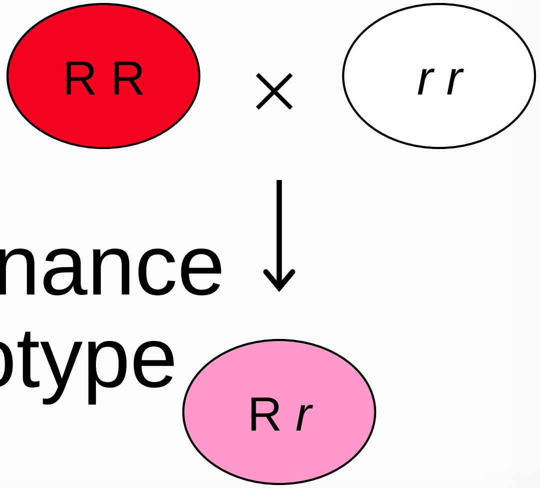
But sometimes phenotypes are not very obvious…

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)!

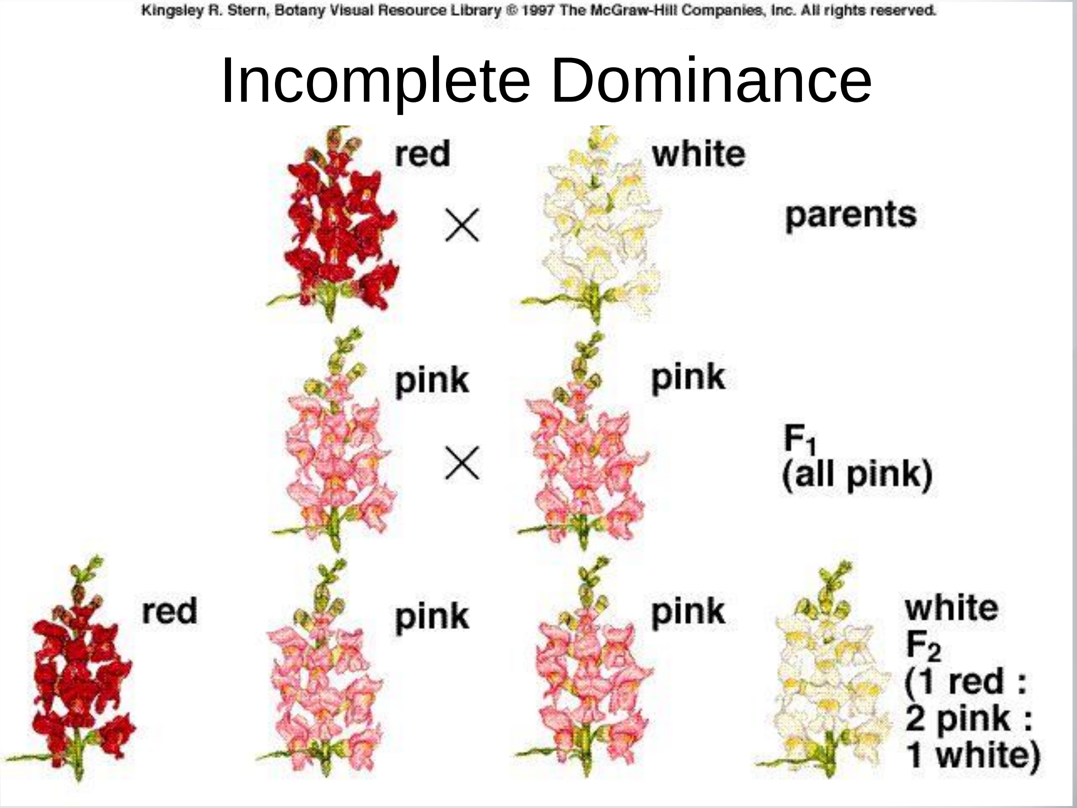


Genes show incomplete dominance when the heterozygous phenotype is intermediate.

Incomplete dominance

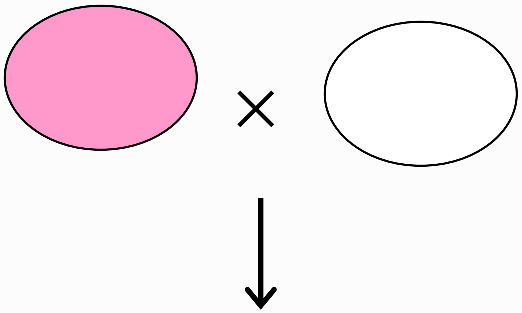
When F1 generation (all pink flowers) is self pollinated, the F2 generation is 1:2:1 red, pink, white

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

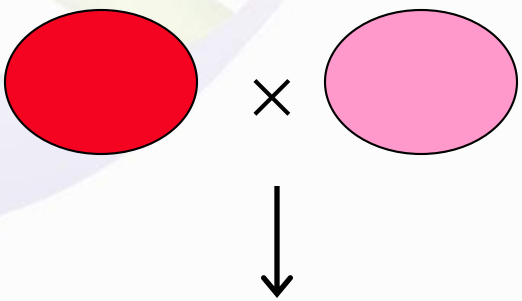


Incomplete dominance

What happens if you cross a pink with a white?



A pink with a red?



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

Lakô hã salamát!

Maraming salamat!