MENDELIAN GENETICS

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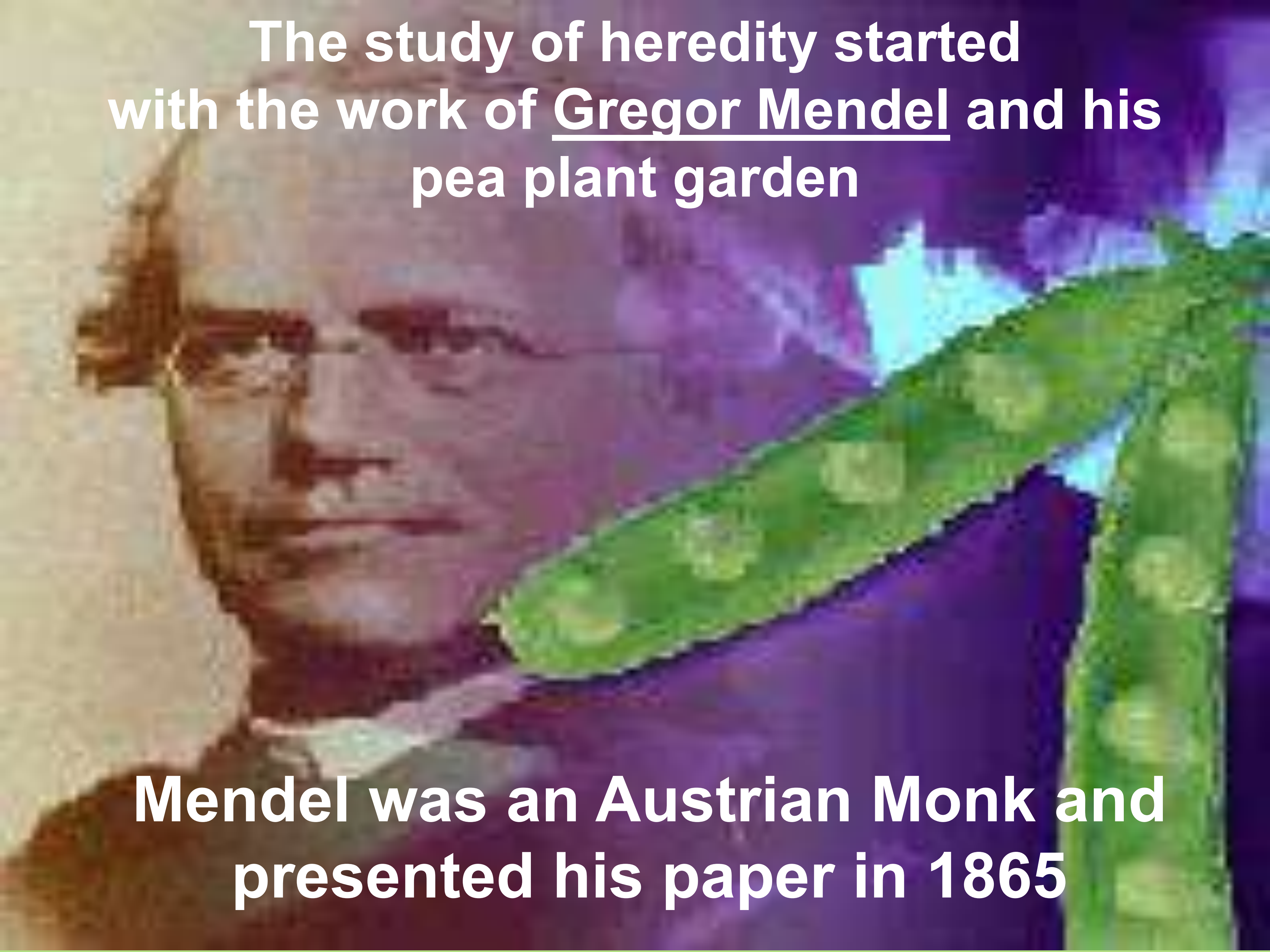
The study of heredity started with the work of Gregor Mendel and his pea plant garden



Mendel was an Austrian Monk and presented his paper in 1865

Mendel's First Law of Genetics (Law of Segregation)

- Genetic analysis predates Gregor Mendel, but Mendel's laws form the theoretical basis of our understanding of the genetics of inheritance.
- Mendel made two innovations to the science of genetics:
  - developed pure lines
  - counted his results and kept statistical notes

LANGUAGE OF GENETICS

- **Phenotype** - means "outward, physical appearance of a particular trait"
- **Genotype** - means "factor that controls or responsible for the outward physical appearance of a particular trait" (which Mendel termed as allele and now called *genes*)
- **Dominant** - the allele that expresses itself at the expense of an alternate allele;
- **Recessive** - an allele whose expression is suppressed in the presence of a dominant allele; the phenotype that disappears in the F_1 generation from the cross of two pure lines and reappears in the F_2 generation
- **Fertilization** - the uniting of male and female gametes
- **Cross** - combining gametes from parents with different traits

Genetics is the study of heredity, how traits are passed from parent to offspring

Why Mendel used peas...

- Their traits are easy to isolate and he selected seven traits.
- Easy to breed
- They reproduce sexually
- They have two distinct, male and female, sex cells called gametes
- Relatively short breeding period allowing a quick experimental turn-around
- Cross pollination was feasible

SELECTED CHARACTERS

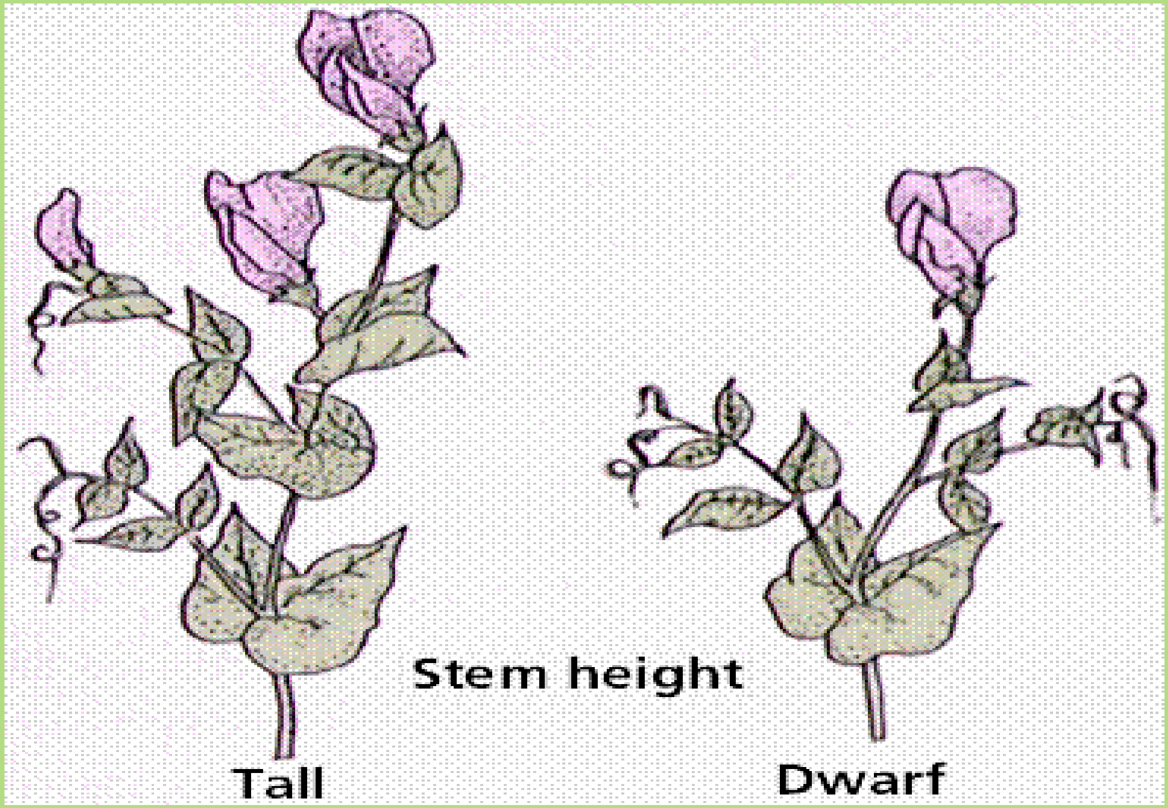
- Seed: smooth vs. wrinkled
- Seeds: yellow vs. green
- Flowers: purple vs. white
- Flowers: axial vs. terminal
- Pods: inflated vs. pinched
- Pods: green vs. yellow
- Stem: tall vs. short

Mendel’s preliminary observation & methodology

- Mendel could isolate plants that produced the same traits generation after generation
- These initial plants are called the P (Parental) generation.
- Cross between parental generation yielded offspring's called the F_1 (Filial generation)
- F1 generation were then self pollinated to produce the F_2 generation.

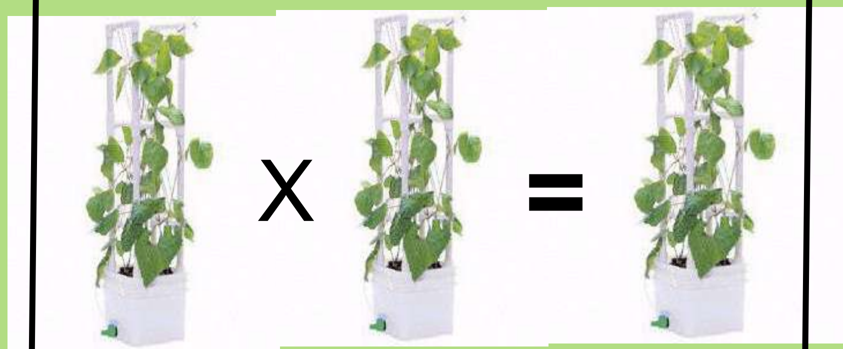
MONOHYBRID CROSS: MENDELS FIRST SET OF EXPERIMENTS

- **Mendel noted that the size of pea plants varied. He cross-bred these pea plants to find some surprising results.**



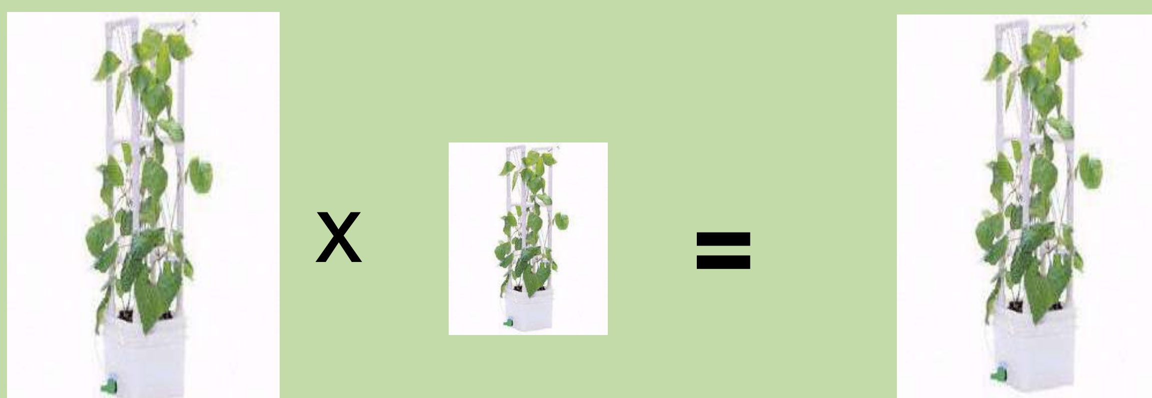
Parental Generation = “P”

Mendel’s cross between tall pea plants yielded all tall pea plants.

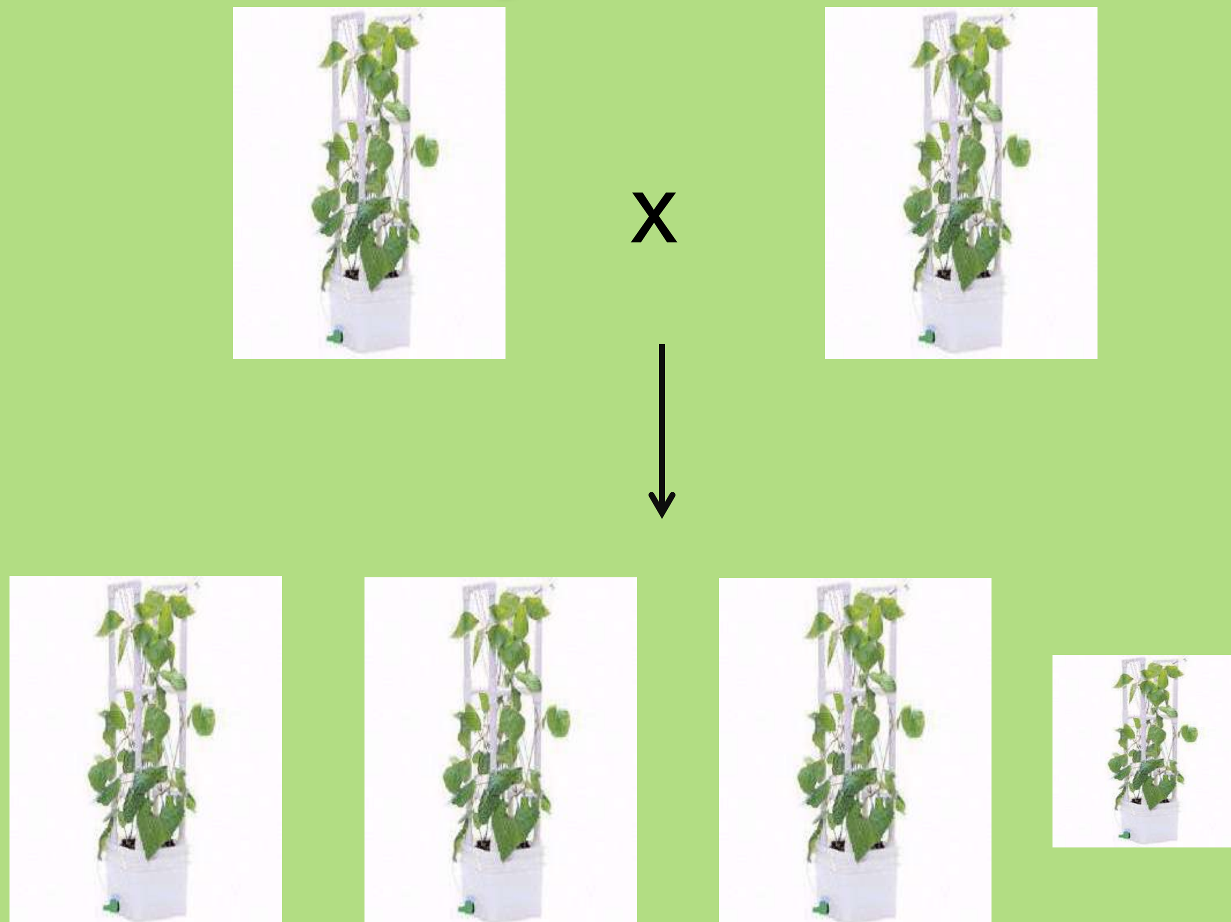


His cross between small pea plants yielded all small pea plants.

Mendels’ cross between tall pea plants and small pea plants yielded all tall pea plants. (F1 generation)



Mendel then crossed these second generation tall pea plants (**F1 generation**) and ended up with 1 out 4 being small.



Summary of Mendel’s Results

Character	Phenotype of F1	Number of F2 Individuals		
		Dominant	Recessive	Total
Seed: smooth vs. wrinkled	All smooth	5474	1850	7324
Seeds: yellow vs. green	All yellow	6022	2001	8023
Flowers: purple vs. white	All purple	705	224	929
Flowers: axial vs. terminal	All axial	651	207	858
Pods: inflated vs. pinched	All inflated	882	299	1181
Pods: green vs. yellow	All green	428	152	580
Stem: tall vs. short	All tall	787	177	1064

OBSERVATION

- How a trait present in one of the parents in P1 generation disappear in the F1 generation
- Trait which disappeared in the F1 generation reappeared in the F2 generation
- F1 generation did not breed true
- F1 generation on self fertilization produced F2 progeny that resembled the parents or P1 generation

Mendel’s Conclusion

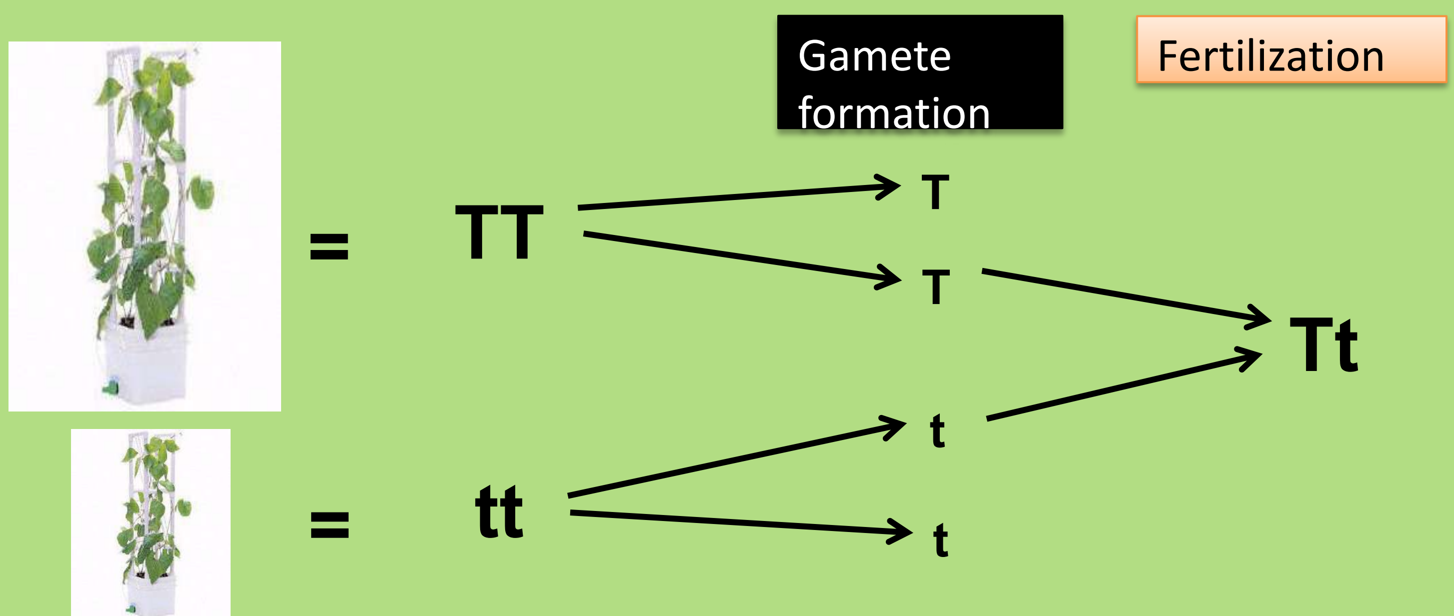
- Each one of the alternate traits i.e. tallness or dwarfness, is determined by a factor.
- Mendel named the factors as alleles which we now call genes. Genes carry hereditary information
- Mendel reasoned that the factors got transmitted from parents to progeny through the gametes
- Since, two contrasting characters were being compared, it was concluded that the alleles exists as in alternate forms
- Moreover, as F1 individuals were able to produce both the phenotype of the parents, it was concluded that both the factors were present in the F1 generation.
- Since, one character disappeared in F1 generation, one of the two factors must be dominant and the other recessive
- Lastly, as two factors were present in F1 generation, it was concluded that the parents also carried two factor.

Summary of the result

- There are alternative forms of genes, the units determining heritable characteristics. This is now known as an **allele**
- An organism inherits one allele from each parent. The F1 generation inherited one green and one yellow pod allele from the parental generation.
- A sperm or egg carries only one allele for each characteristic which pair upon fertilization.

Gamete formation & Fertilization

- Each individual has two factors for each trait
- The factors segregate during gamete formation
- Each gamete contains only one factor from each pair of factors
- Since each parent is true breeding and each contains two copies of the same allele, the genotype of the parental plant may be represented as follows
- Fertilization gives each new individual two factors for each trait
- Since each parent is true breeding and each contains two copies of the same allele, the genotype of the parental plant may be represented as follows



Punnett Square

- The best way to express what happens in the F_1 and F_2 generation is with a type of diagram known as a Punnett Square

female \ male	T	T
t	Tt	Tt
t	Tt	Tt

Result: All tall plants observed in F1 generation

Tall = dominant

short = recessive

RECIPROCAL CROSSES ALSO PRODUCED THE SAME RESULT

F2 Generation (F1 X F1)

Tt → **T**, **t**

Tt → **T**, **t**

Gamete formation

Fertilization

male / female	T	t
T	TT Tall	Tt Tall
t	Tt Tall	tt Dwarf

LAW OF SEGREGATION

- Based on the results of the monohybrid cross, Mendel proposed his First Law of Segregation

Mendel's first law states that each phenotype is determined by a pair of factors which we now call genes. These pair of factors segregate from each other while forming gametes. At the time of fertilization, the segregated gametes pair again reunite to form new combination.

DIHIBRID CROSS & MENDEL'S LAW OF INDEPENDENT ASSORTMENT

What happens when more than one pair of characters is simultaneously involved in the crosses?

States that allele pairs separate and assort independently during the formation of gametes. The alleles or traits are transmitted to offspring independently of one another.

DIHYBRID EXPERIMENT

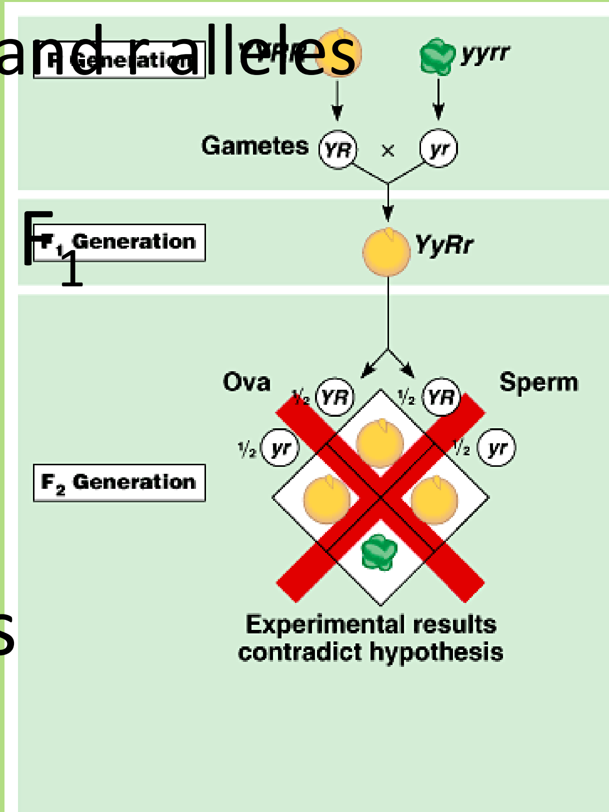
- In one **dihybrid** cross experiment, Mendel studied the inheritance of seed color and seed shape.
  - The allele for yellow seeds **(Y)** is dominant to the allele for green seeds **(y).**
  - The allele for round seeds **(R)** is dominant to the allele for wrinkled seeds **(r).**
- Mendel crossed true-breeding plants that had **yellow, round** seeds **(YYRR)** with true-breeding plants that has **green, wrinkled** seeds **(yyrr).**
- The resulting F1 dihybrid progeny were heterozygous for both traits **(RrYy)** and had **round yellow seeds**, the dominant phenotypes.
- From the F1 generation, Mendel could not tell if the two characters were inherited independently or not, so he allowed the **F1** progeny to **self-pollinate.**

Mendel considered two alternate hypothesis:

- **Hypothesis 1**: If the two characters segregate together, the F1 hybrids can only produce the same two classes of gametes
- **(RY** and **ry)** that they received from the parents, and the F2 progeny will show a 3:1 phenotypic ratio.
- **Hypothesis 2**: If the two characters segregate independently, the F1 hybrids will produce four classes of gametes **(RY, Ry, rY, ry)**
- And the F2 progeny will show a 9:3:3:1 ratio
- **Experiment:** Mendel performed a dihybrid cross by allowing self-pollination of the F1 plants (RrYy X RrYy)
- **Results:** Mendel categorized the F2 progeny and determined a ratio of 315:108:101:32 which approximates 9:3:3:1.
- **Conclusion:** The experimental results supported the hypothesis that each allele pair segregates independently during gamete formation

POSSIBLE OUTCOME

- 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_1 offspring would produce yellow, round seeds.
- The F_2 offspring would produce two phenotypes in a 3:1 ratio, just like a monohybrid cross.

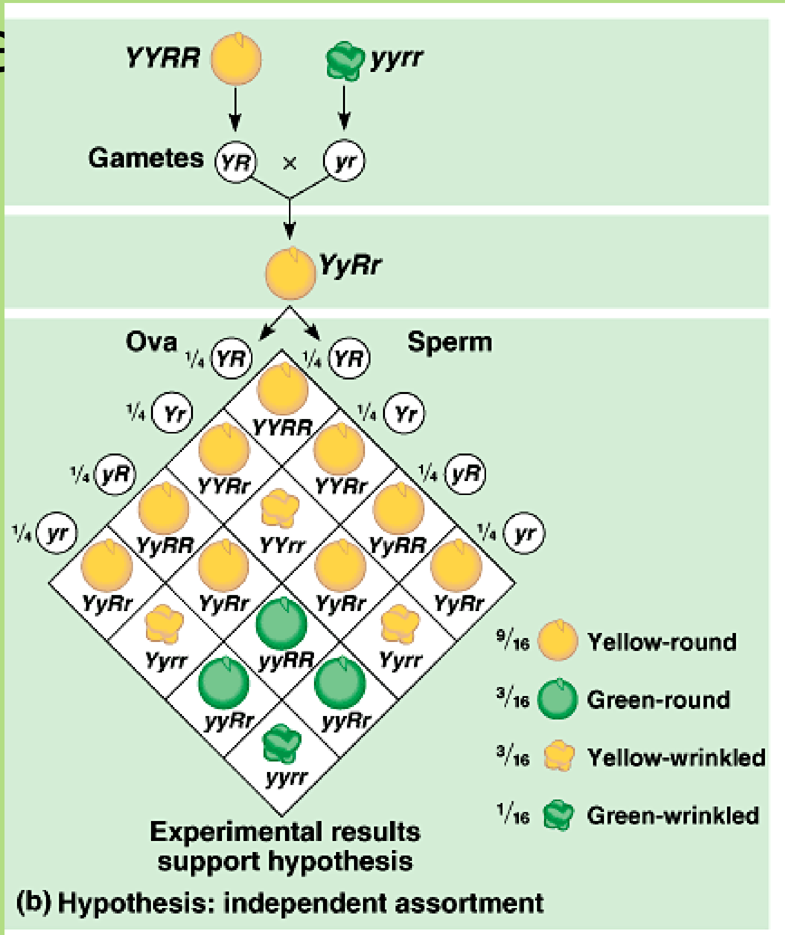


(a) Hypothesis: dependent assortment

- 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_1 offspring would still produce yellow, round seeds.
- However, when the F_1's produced gametes, genes would be packaged into gametes with all possible allelic

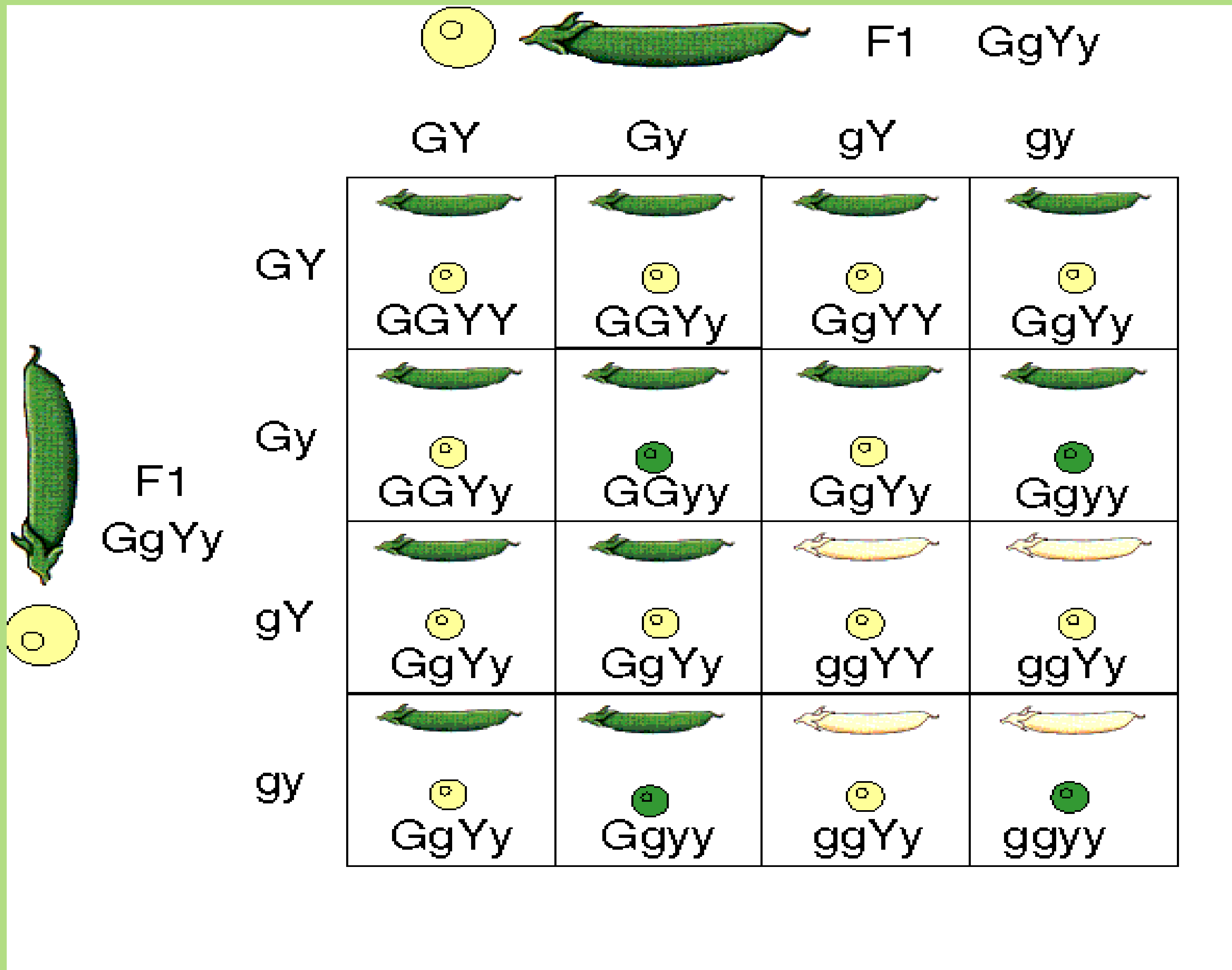
DIHIBRID CROSS

- When sperm with four classes of alleles and ova with four classes of alleles combined, the 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.



- Mendel repeated the dihybrid cross experiment for other pairs of characters and always observed a 9:3:3:1 phenotypic ration in the F_2 generation.
- Each character appeared to be 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

Dihybrid cross



Mendel's two laws explain inheritance in terms of discrete factors (genes) which as passed from generation to generation according to simple rules of chance.

- **These principles apply to all sexually reproducing organisms for simple patterns of inheritance.**
- **Experiments with many that more complicorganisms indicate ated patterns of inheritance exist.**
- **The more complicated patterns of inheritance include situations where one allele is not completely dominant over another allele, there are more than two alleles for a trait, or the genotype does not always dictate the phenotype in a rigid manner.**

TEST CROSS & BACK CROSS

- It is not always possible to determine what genes an organism is carrying by simply looking at its appearance.
- After all, gene expression is a complex process that is dependent on many environmental and hereditary factors.
- Today, scientists use the word "phenotype" to refer to what Mendel termed "external resemblance" and the word "genotype" to refer to an organism's "internal nature."

TEST CROSS & BACK CROSS

- Mendel showed that phenotypic traits can be hidden in one generation, yet reemerge in subsequent generations. (MONOHIBRID CROSS)
- This occurs because some alleles are dominant over others, which means that their phenotype will mask the phenotype associated with the recessive alleles as shown in the table below.

Table 1: Male Fly Is Homozygous (EE –black eye) X **Female Gametes *(EE –brown eye)***

		Female Gametes *(EE –brown eye)*	
	GAMETES	**E**	**E**
Male Gametes *(EE –black eye)*	**E**	*Ee* ***(BLACK EYE)***	*Ee* ***(BLACK EYE)***
	E	*Ee* ***(BLACK EYE)***	*Ee* ***(BLACK EYE)***

TEST CROSS

- Gregor Mendel's experiments with pea plants showed how dominant traits can mask recessive ones (Mendel, 1866). To comprehend whether an individual is homozygous or heterozygous for an allele expressing dominant phenotype, Mendel devised the test cross.

Definition of Test Cross: a test cross is an experimental cross of an individual organism of dominant phenotype but unknown genotype and an organism with a homozygous recessive genotype (and phenotype).

Definition of Back Cross: in an experimental if the F1 generation is crossed with any of the parental generation that it is called BACK Cross.

Test Cross: heterozygous gamets

- ***Table 1: Outcome if Male Fly Is Heterozygous (Ee)***

	Female Gametes		
Male Gametes	gametes	**e**	**e**
	E	***Ee***	***Ee***
	e	***ee***	***ee***

If the individual is heterozygous, the phenotypic ration of the F1 generation of a test cross will always be 1: *1*

Test Cross: homozygous gamets

- ***Table 2: Outcome if Male Fly Is Homozygous (EE)***

		Female Gametes	
	gametes	**e**	**e**
Male Gametes	**E**	*Ee*	*Ee*
	E	*Ee*	*Ee*

If the individual is homozygous, the phenotype of all the F1 individuals will same irrespective of the allele in consideration is dominant or recessive.

Conclusion

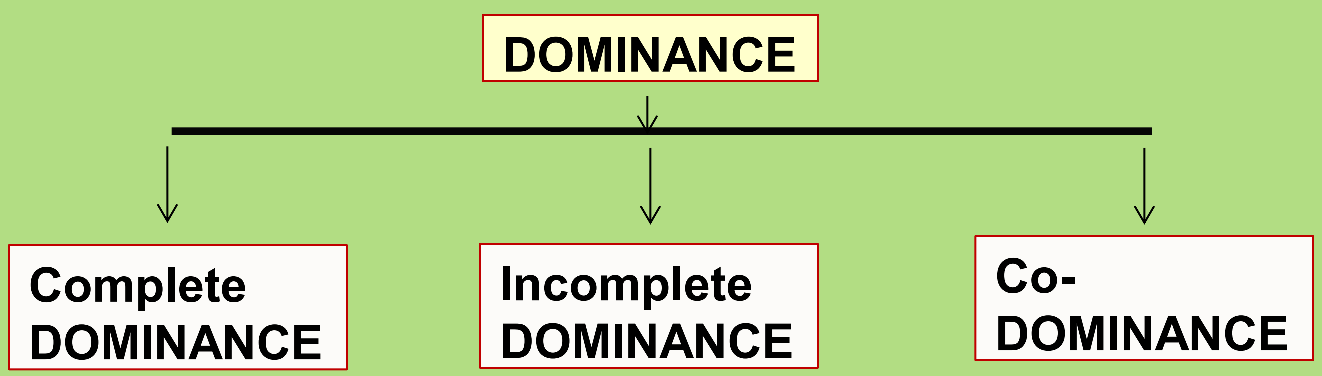
- Test crosses operate under the same principle no matter whether you are considering one gene or multiple genes; in all cases, you are crossing an individual of a dominant phenotype but unknown genotype to an individual that is homozygous recessive for all relevant genes.

EXTENSIONS AND MODIFICATION OF MENDELIAN GENETIC ANALYSIS

- Subsequent analysis of genes revealed that there are exceptions to and extensions to Mendel's laws
- Such findings do not invalidate Mendel's findings but reveal the complexity of inheritance of traits

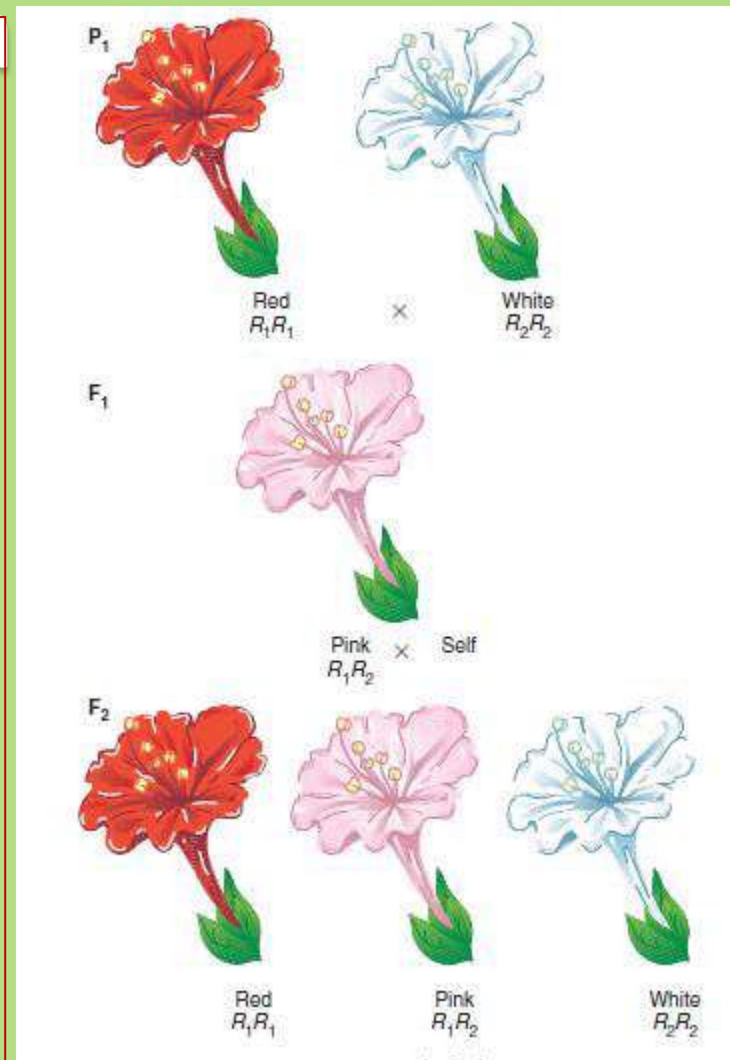
DOMINANCE

- Dominant genes are not always universal
- If all the genes were either dominant or recessive, then all the heterozygotes would look like the dominant parent and we would always see the 3:1 ratio when heterozygotes are crossed.
- If, however, the heterozygote were distinctly different from both homozygotes, we would see a 1:2:1 ratio of phenotypes when heterozygotes are crossed.



Partial dominance or Incomplete domina

- Carl Correns crossed red flower coloured four-o'clock plants (*Mirabilis jalapa)* with white flower coloured plants
- F1 generation plants produced all pink coloured flowers
- When the F1 plants were self fertilized,



Conclusion

- In this case, one allele (*R1) specifies* red pigment color, and another allele specifies no color
- Flowers in heterozygotes (*R1R2) have about half* the red pigment of the flowers in red homozygotes(*R1R1) because the heterozygotes have only one copy of* the allele that produces color, whereas the homozygotes have two copies
- Such phenomenon where the

Codominance

- **Codominance** in which the phenotype of the heterozygote is not intermediate between the phenotypes of the homozygotes; rather, the heterozygote simultaneously expresses the phenotypes of both homozygotes.

  Best example is the MN blood group

- MN locus codes for one of the types of antigens on red blood cells.
- Foreign MN antigens do not elicit a

$L^M L^N$ Blood Group

- At the MN locus, there are two alleles:
  - the *L^M allele, which codes for the M antigen;*
  - the *L^N allele, which codes for the N antigen.*
- *Homozygotes* with genotype *$L^M L^M$ express the M antigen on their red blood* cells and have the M blood type.
- Homozygotes with genotype *$L^N L^N$ express the N antigen and have the N blood type.*

Differences between dominance, incomplete dominance, and codominance

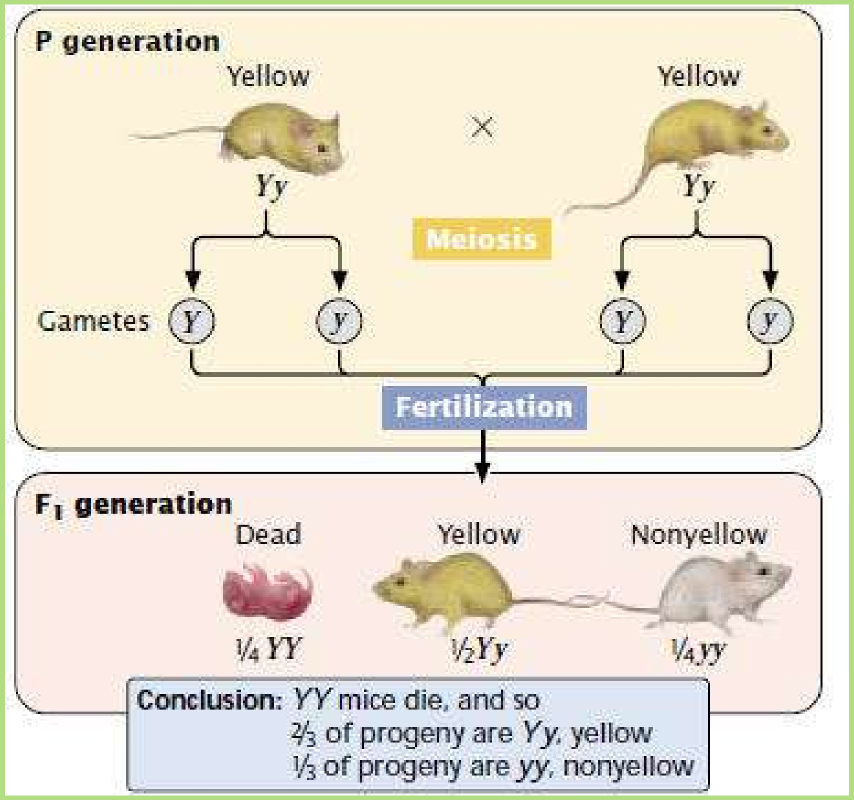
Type of Dominance	Definition
Dominance	Phenotype of the heterozygote is the same as the phenotype of one of the Homozygotes
Incomplete dominance	Phenotype of the heterozygote is intermediate (falls within the range) between the phenotypes of the two homozygotes
Codominance	Phenotype of the heterozygote includes the phenotypes of both Homozygotes

Lethal Alleles

- In 1905, when Lucien Cuenot mated two yellow mice,
- approximately ⅔of their offspring were yellow and ⅓were nonyellow.
- When he test-crossed the yellow mice, he found that all were heterozygous;
- he was never able to obtain a yellow mouse that bred true.
- it was eventually realized that the yellow allele must be lethal when homozygous

Lucien Cuenot's experiment

- Cuenot originally crossed two mice heterozygous for yellow: *Yy Yy. Normally, this cross would be expected to* produce *YY, Yy, and yy.*
- *The* homozygous *YY mice are conceived but never complete* development, which leaves a 2 : 1 ratio of *Yy (yellow) to yy (nonyellow) in the observed offspring; all yellow mice are* heterozygous *(Yy).*



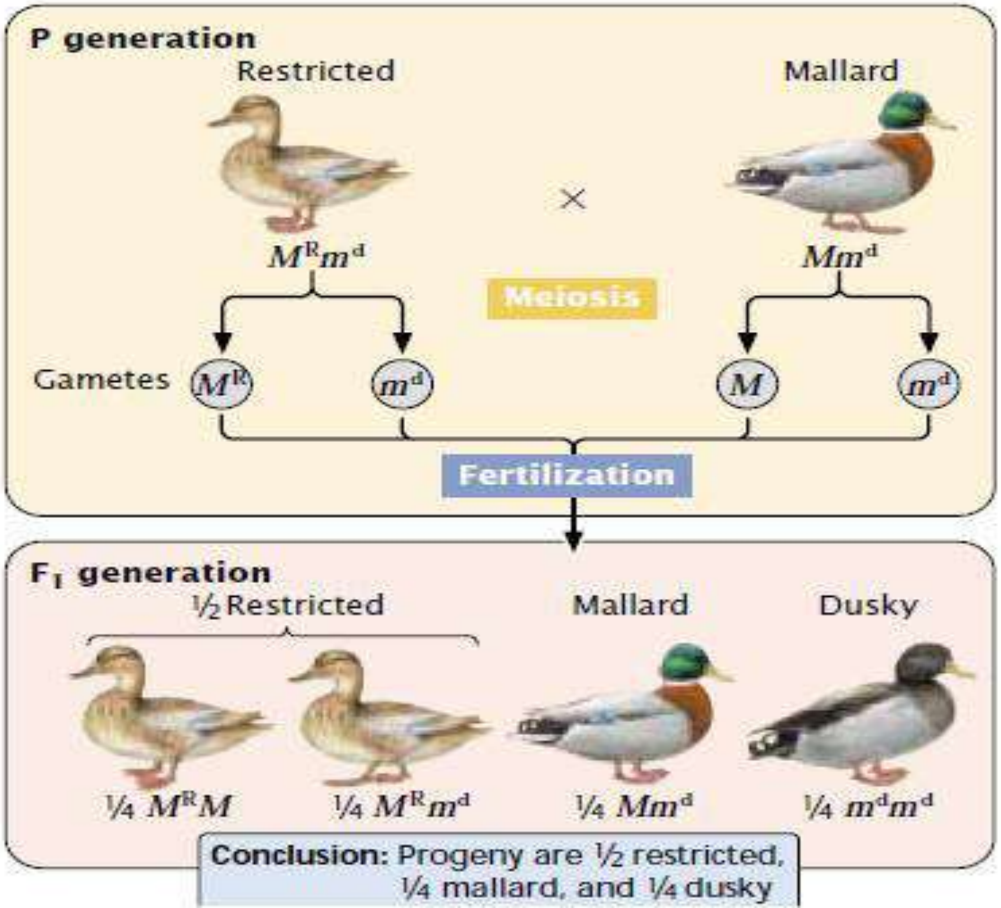
Multiple Alleles

- In every individual, each loci on chromosome is occupied by a gene.
- the genotype of each diploid *individual have* only two alleles at each loci.
- Now, in a outbred population, there may be more than two variety of the same gene or allele.
- When more than two variety of the same allele exists in the population, the phenomenon is referred to as multiple allelic

Duck-Feather Patterns

- The feather pattern of mallard ducks is determined multiple alleles at the locus
- One allele, ***M***, produces the **wild-type *mallard pattern***. *A second allele, **M^R**,* produces a different pattern called ***restricted****, and a third* allele, ***md****, produces a pattern termed* ***dusky****.*
- *In this allelic* series, **restricted is dominant over mallard and dusky**, and **mallard is dominant over dusky**: *M^R >M >md. The six* genotypes possible with these three alleles and their resulting phenotypes are:

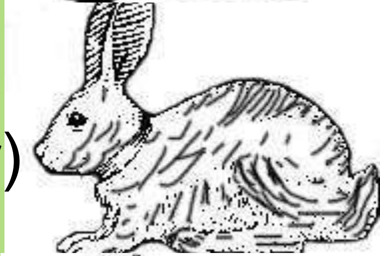
Genotype	Phenotype
M^RM^R	*restricted*
M^RM	*Restricted*
M^RM^d	*Restricted*
MM	*Mallard*
MM^d	*Mallard*
m^dm^d	*dusky*



Coat Color of Rabbits ($c^+ > c^{ch} > c^h > c$)

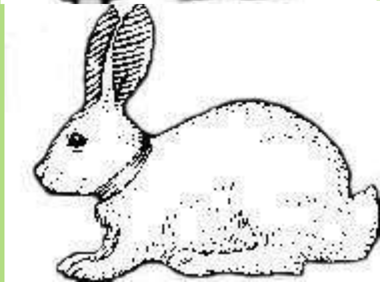
Four alleles determine the coat color "**c**":

c^+ = wild type (fully coloured)
(c^+/c^+ ; c^+/c^h; c^+/c^{ch} ; c^+/c)



c^{ch} = chinchilla (entire body is grey color)
(c^{ch} / c^{ch}; c^{ch} /c^h; c^{ch} /c)

c^h = hymalayan type (white tips at the extremes)
(c^h /c^h; c^h /c)



c = albino (c/c)

The ABO Blood Group in humans

- *IA, which codes for the A antigen;*
- *IB, which codes* for the B antigen;

Dominance relations among the ABO alleles as follows:

IA >i, IB >i, IA = IB.

The *IA and IB alleles are* both dominant over *i and are codominant with each other*

GENE INTERACTIONS AND MODIFIED MENDELIAN RATIOS

- No gene acts by itself in determining the phenotype of an individual
- The phenotype is the result of integrated pattern of molecular reactions
- The molecules that are responsible for the phenotypic expression is under the direct control of genes

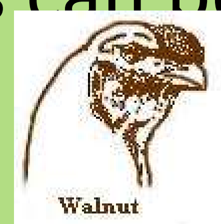
Any alteration from the standard 9:3:3:1 indicates that the phenotype is the product of the interaction of two or more genes. This altered genotypic ratios are referred to as modified mendelian ratios

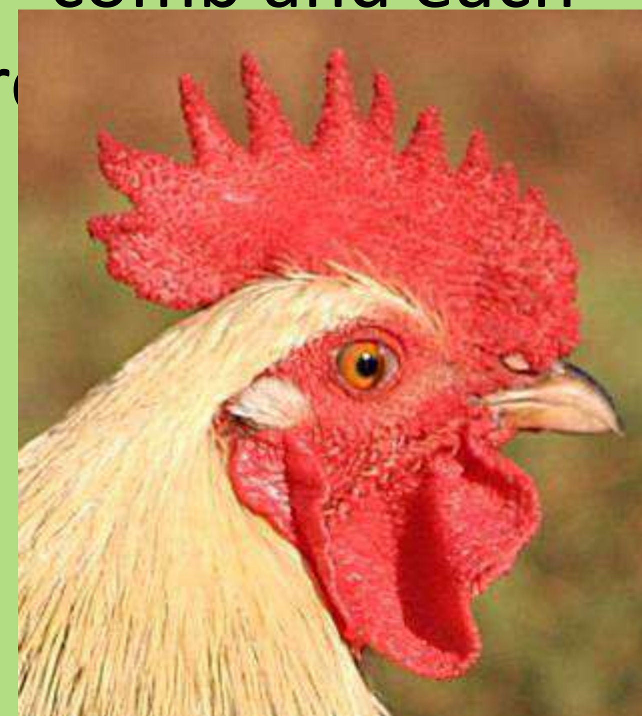
GENE INTERACTION AND NEW PHENOTYPE

- British geneticists William Bateson and R. C. Punnett showed that a specific phenotype (for e.g. Comb Shape in Chickens) is under the influence of several gene pairs
- A variety of phenotypic ratios can result depending on the type and the extent of interaction between the products of the nonallelic genes.

Epistasis

- For instance, Wyandottes have a "rose" comb, Brahmas have a "pea" comb, and Leghorns have a "single" comb and each of these types can be br



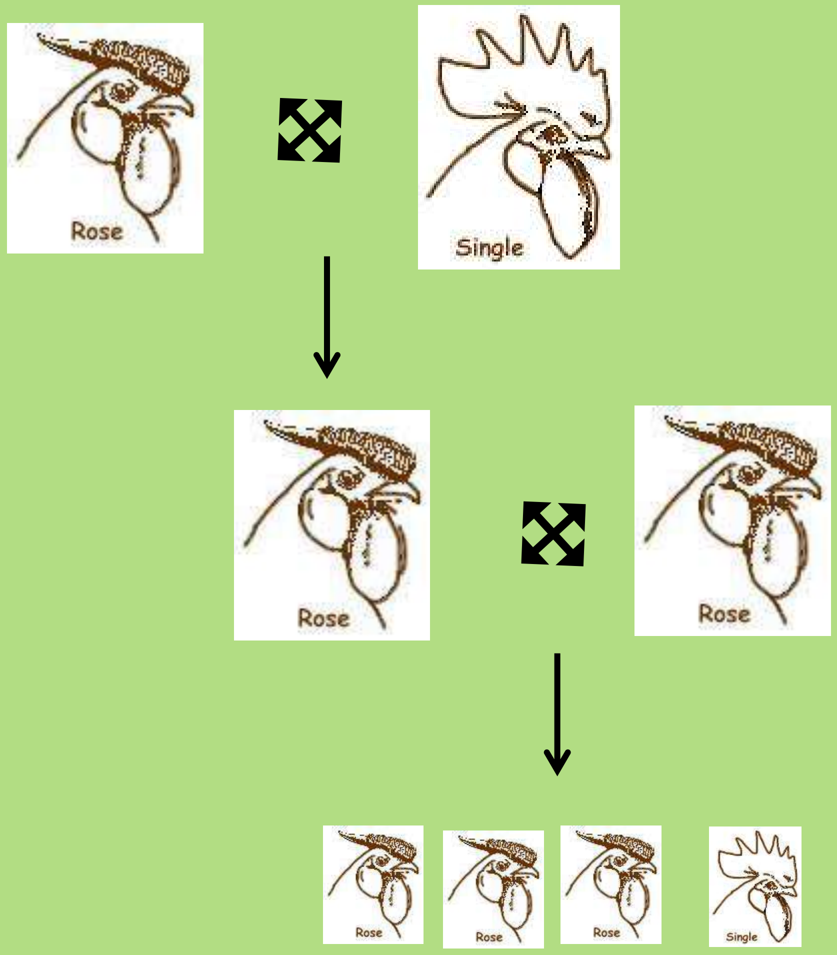


ROSE dominant over SINGLE

Parents

F1 generation

F2 generation

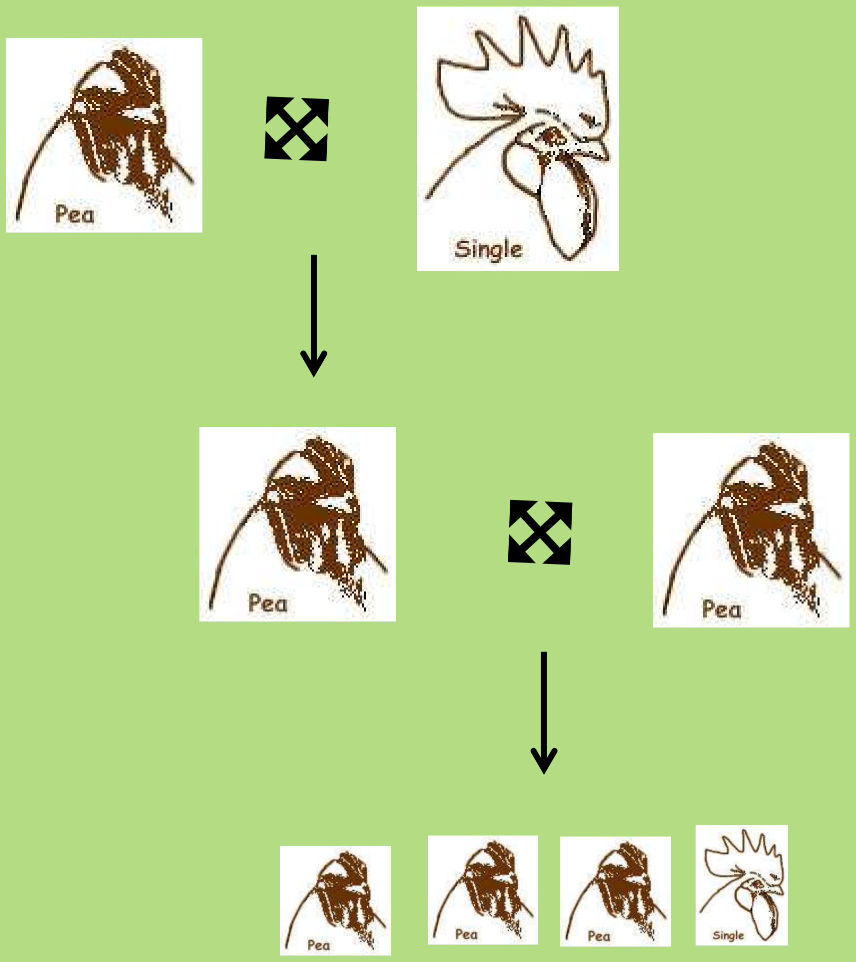


PEA dominant over SINGLE

Parents

F1 generation

F2 generation

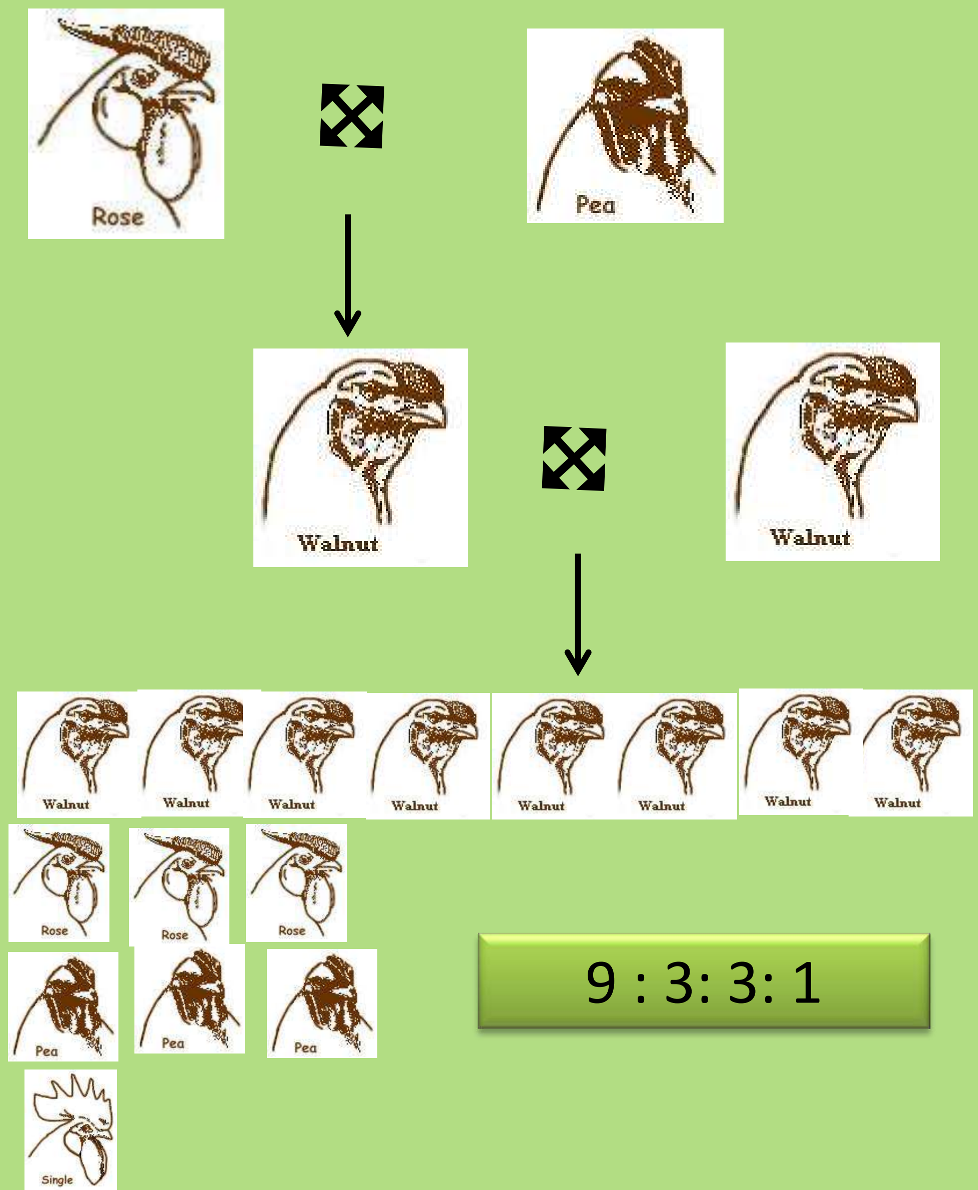


Rose crossed with Pea

Parents

F1 generation

F2 generation



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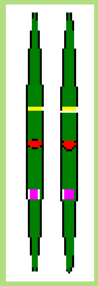
ANALYSIS OF Bateson and Punnett's experiment

- The walnut comb depends on the presence of two dominant alleles R & P
- R & P are two independently assorting gene loci



- RRPP; RRPp RrPP RrPp all gives walnut comb
- RRpp Rrpp all gives rose comb
- rrPP; rrPp all gives pea comb
- rrpp combination produces single comb

Parents genotype	Rose comb RRpp	Pea comb rrPP
gametes	Rp	rP
F1 genotype	RrPp	

RrPp

All Walnut comb

male / female	**RP**	**Rp**	**rP**	**rp**
RP	RRPP Walnut	RRPp Walnut	RrPP Walnut	RrPp Walnut
Rp	RRPp Walnut	RRpp Rose	RrPp Walnut	Rrpp Rose
rP	RrPP Walnut	RrPp Walnut	rrPP Pea	rrPp Pea
rp	RrPp Walnut	Rrpp Rose	rrPp Pea	rrpp Single

THANK YOU