Mendel’s laws

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Mendel’s laws

- Mendel experiment
- Mendelian ratios and statistic
- Deviations from Mendelian ratios
- Type of inheritances
- Gene interactions

Mendel’s laws

- Gregor Mendel was born by July 1822 in Heizendorf (today Hynice in the Czech Republic)

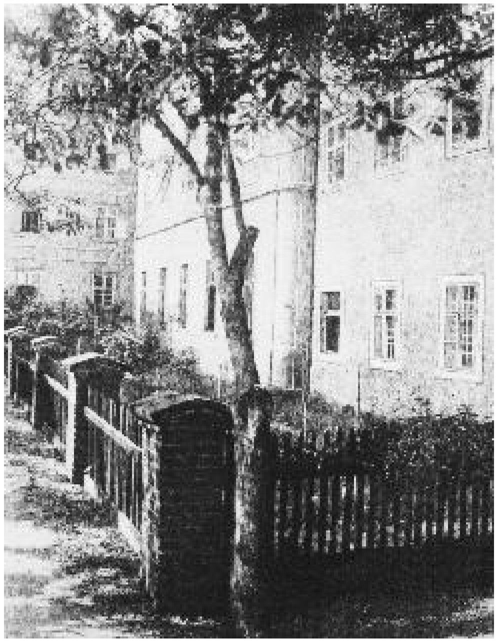
- From 1851 to 1853, studied zoology, botany, chemistry, and physics at the University of Vienna

- He became a member of the Zoologico-Botanical Society of Austria and has published two scientific treatises in the "Verhandlungen" of this scientific organization (1853 and 1854)

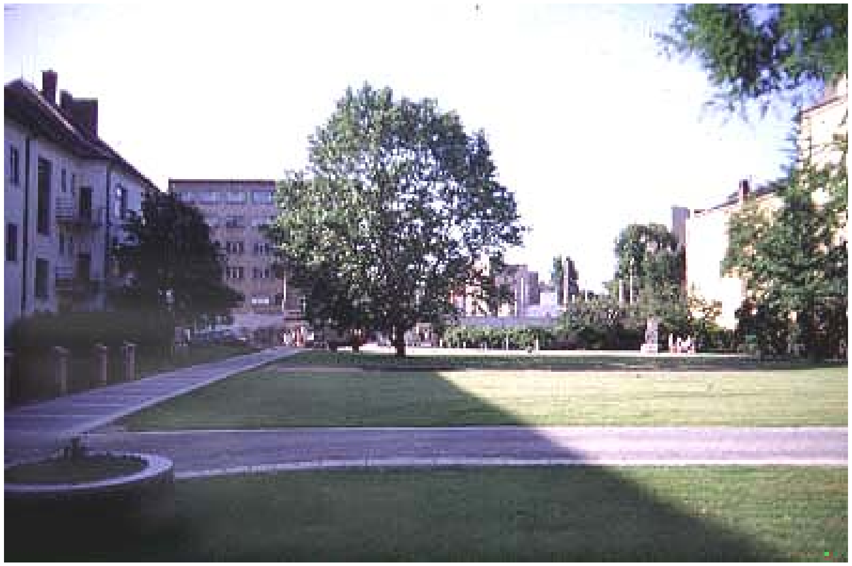
- Probably due to health reasons (epilepsia ?), Mendel has returned to Brno without formally finishing the University in Vienna.

Mendel's laws

- **Experimentation in the monastery garden results in laws of inheritance**
- **Mendel began his experiments with the hybrid cultivation of pea plants in 1856**
- **He reported on the results of his observations at the meetings of the Association for Natural Research in Brno in 1865.**
- **The Association published the written accounts of these observations in 1866, under the title Versuche über Pflanzen-Hybride.**



The garden of the Augustinian Convent in Brno.



Mendel's laws

- **There was little reaction from the scientific community**
  - **Nature of his experimentation was unconventional for his age, nobody before him had use mathematical and statistical analysis as a means of interpreting the results of biological inquiry**
  - **Mendel was known as a relatively shy person and might not have presented his results with the necessary emphasis and stress**
  - **Scientific fraternity of the day will have been the limited number of people who read the Brno Association's records**
  - **Although it in fact dealt with no fewer than 355 cross-bred strains and 12,980 resultant hybrids, his work was described as "incomplete“.**
  - **Mendel received a "fatal" advice: to continue his investigations using the hawkweed (*Hieracium*), later botanists discovered that these plants have asexual reproduction.**

Mendel's laws

- **The rediscovery of Mendel's Laws**
  - **It was to take thirty-four years before Mendel's prediction came true.**
  - **The year 1900, Carl Correns in Germany, Hugo de Vries in the Netherlands and Erich von Tschermak-Seysenegg in Austria.**
  - **Their achievement was to realize that Mendel had not merely conducted experiments in successful hybridization**
  - **But had in fact studied the heredity of specific characteristics as they were passed on from parent plants to their offspring.**

Mendel experiment

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

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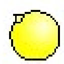
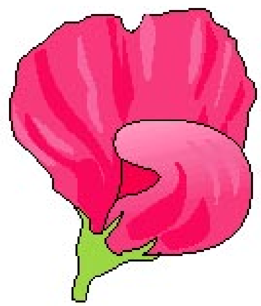
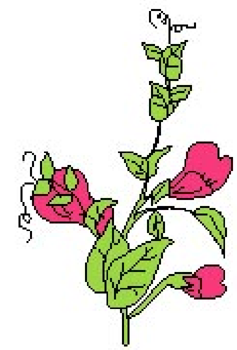
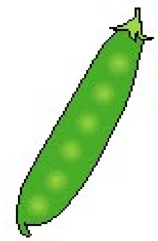
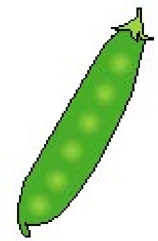
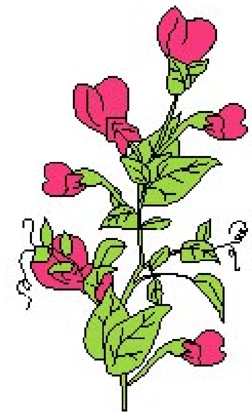
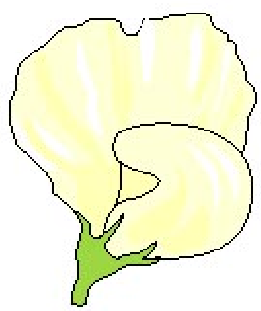
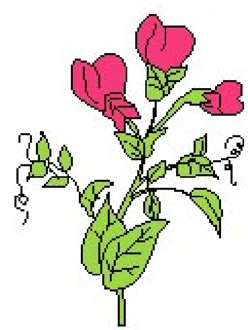
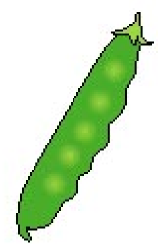
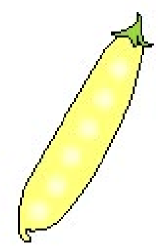
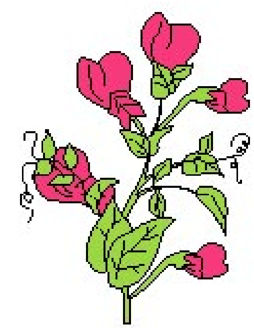
DOMINANTE	Lisa 	Amarilla 	rojo-violeta 	Flores axiales 	Inflada 	Verde 	Alta 
RECESIVO	Rugosa 	Verde 	Blanca 	Flores terminales 	Apretada 	Amarilla 	Baja 

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Mendel experiment

Pure Line - a population that breeds true for a particular trait [this was an important innovation because any non-pure (segregating) generation would and did confuse the results of genetic experiments]

Results from Mendel's Experiments

Parental Cross	**F_1 Phenotype**	**F_2 Phenotypic Ratio**	**F_2 Ratio**
Round x Wrinkled Seed	Round	5474 Round:1850 Wrinkled	2.96:1
Yellow x Green Seeds	Yellow	6022 Yellow:2001 Green	3.01:1
Red x White Flowers	Red	705 Red:224 White	3.15:1
Tall x Dwarf Plants	Tall	l787 Tall:227 Dwarf	2.84:1

Mendel experiment

Terms and Results Found in the Table

1. **Phenotype. Literally means "the form that is shown"; it is the outward, physical appearance of a particular trait**

   Mendel's pea plants exhibited the following phenotypes:
   - **round or wrinkled seed phenotype**
   - **yellow or green seed phenotype**
   - **red or white flower phenotype**
   - **tall or dwarf plant phenotype**

2. **What is seen in the F1 generation? We always see only one of the two parental phenotypes in this generation. But the F1 possesses the information needed to produce both parental phenotypes in the following generation. The phenotype of the F1 is dominant to the trait that disappears in the F1.**

Mendel experiment

3. **The F2 generation always produced a 3:1 ratio where the dominant trait is present three times as often as the recessive trait.**
4. **Mendel coined two new terms**

Dominant

- **the allele that expresses itself at the expense of an alternate allele; the phenotype that is expressed in the F1 generation from the cross of two pure lines**

Recessive

- **an allele whose expression is suppressed in the presence of a dominant allele; the phenotype that disappears in the F1 generation from the cross of two pure lines and reappears in the F2 generation**

Mendel experiment

Mendel's Conclusions

- The hereditary determinants are of a particulate nature. These determinants are called genes.

- Each parent has a gene pair in each cell for each trait studied. The F1 from a cross of two pure lines contains one allele for the dominant phenotype and one for the recessive phenotype. These two alleles comprise the gene pair.

- One member of the gene pair segregates into a gamete, thus each gamete only carries one member of the gene pair

- Gametes unite at random and irrespective of the other gene pairs involved

Other Important Genetic Terms

- **Allele**
  - **one alternative form of a given gene pair; tall and dwarf are the alleles for the height of a pea plant; more than two alleles can exist for any specific gene, but only two of them will be found within any individual**
- **Allelic pair**
  - **a combination of two alleles**
- **Homozygote**
  - **an individual which contains only one allele at the allelic pair; for example *DD* is homozygous dominant and *dd* is homozygous recessive; pure lines are homozygous for the gene of interest**

Other Important Genetic Terms

- **Heterozygote**
  - **an individual that contains two different alleles at the allelic pair; for example the *Dd* heterozygote**
- **Genotype**
  - **the specific allelic combination for a certain gene or set of genes**

Symbolic representation of the cross between tall and short pea plants

Parental Generation ***DD*** **x** ***dd***

↓ ↓

Parental Gametes ***D*** ***d***

↘ ↙

F_1 Genotype ***Dd***

The Punnett square shows the F2 generation created by selfing the F1 plants.

Union of gametes occurs at random

	D	***d***
D	***DD*** **(tall)**	***Dd*** **(tall)**
d	***Dd*** **(tall)**	***dd*** **(dwarf)**

The Punnett Square allows us to visualize specific genetic ratios.

F_2 Genotypic ratio:	1 *DD* : 2 *Dd* : 1 *dd* (or 3 *D_* : 1 *dd*)
F_2 Phenotypic ratio:	3 tall : 1 dwarf

Mendel's First Law
The law of segregation; during gamete formation each member of the allelic pair separates from the other member to form the genetic constitution of the gamete

F3 confirmation of Mendel's first law

Mendel confirmed his hypothesis by selfing the F2 plants. If his law was correct he could predict what the results would be.

F_2 **Phenotypic Classes**	Self Tall (*D_*)	Self Dwarf (*dd*)
	↓	↓
F_3 **Phenotypic Ratios**	1/3 All tall : 2/3 segregating	All dwarf
	↓	↓
	3 tall : 1 dwarf	

From these results we can now confirm the genotype of the F2 individuals.

F_2 **Tall**	1/3 *DD* (pure line homozygote dominant) 2/3 *Dd* (Heterozygotes)
F_2 **Dwarf**	all *dd* (Pure line homozygote recessive)

Thus the F_2 is genotypically: 1/4 DD : 1/2 Dd : 1/4 dd

This data was also available from the Punnett Square using the gametes from the F_1 individual. So although the phenotypic ratio is 3:1 the genotypic ratio is 1:2:1

Backcross Confirmation of Mendel's First Law of Segregation

Mendel performed one other cross to confirm the hypothesis of segregation --- the backcross. Remember, the first cross is between two pure line parents to produce an F1 heterozygote.

Parental Cross *DD* x *dd*

↓

F_1 *Dd*

At this point instead of selfing the F1, Mendel crossed it to a pure line, homozygote dwarf plant. The cross looks like this.
Backcross: *Dd* x *dd*

MaleGametes

Female Gametes		*d*
	D	*Dd*
	d	*dd*

Backcross One or (BC_1) Phenotypes: 1 Tall : 1 Dwarf
BC_1 Genotypes: 1 *Dd* : 1 *dd*

More Genetic Terms

Backcross

- **the cross of an F1 hybrid to one of the homozygous parents; for pea plant height the cross would be Dd x DD or Dd x dd; most often, though a backcross is a cross to a fully recessive parent**

Testcross

- **the cross of any individual to a homozygous recessive parent; used to determine if the individual is homozygous dominant or heterozygous**

Monohybrid cross

- **cross between parents that differ at a single allelic pair (usually AA x aa)**

Monohybrid

- **the offspring of two parents that are homozygous for alternate alleles of a gene**

Remember --- a monohybrid cross is not the cross of two monohybrids.

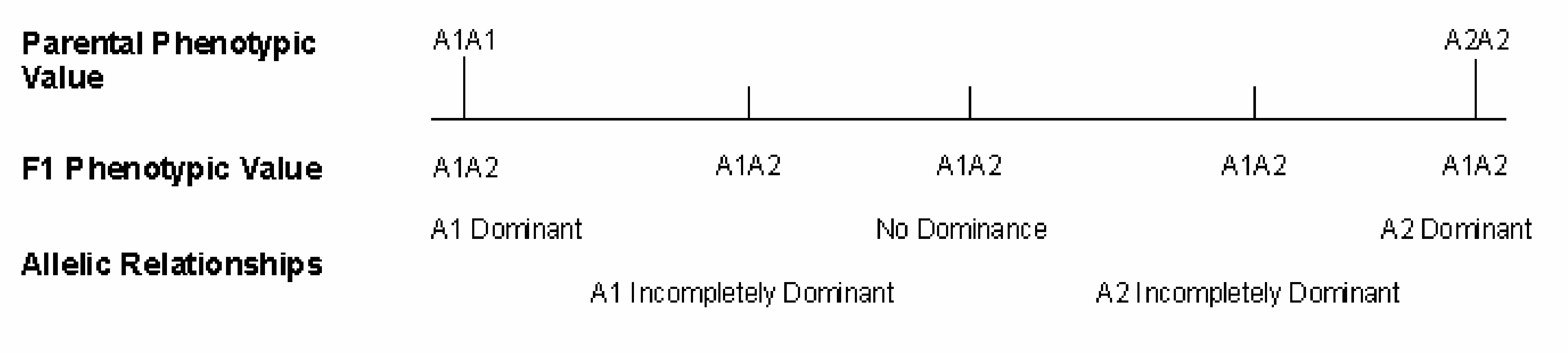
Dominance

- **the ability of one allele to express its phenotype at the expense of an alternate allele; the major form of interaction between alleles; generally the dominant allele will make a gene product that the recessive can not; therefore the dominant allele will express itself whenever it is present**

Incomplete Dominance

- **Incomplete dominance**
  - **the F1 produces a phenotype quantitatively intermediate between the two homozygous parents; if the product is exactly intermediate between the two homozygous parents the relationship is termed no dominance (although some have tried to substitute the term no dominance for codominance, it has not been widely accepted)**

- **Diagrammatic Representation of Allelic Relationships**



Mendel's Law of Independent Assortment

- **Dihybrid cross**
  - **a cross between two parents that differ by two pairs of alleles (AABB x aabb)**
- **Dihybrid**
  - **an individual heterozygous for two pairs of alleles (AaBb)**

Parental Cross	Yellow, Round Seed x Green, Wrinkled Seed
	↓
F_1 Generation	All yellow, round
	↓
F_2 Generation	9 Yellow, Round:3 Yellow, Wrinkled:3 Green, Round:1 Green, Wrinkled

Mendel's Law of Independent Assortment (cont.)

- Gene Symbols: Seed Color: Yellow = G; Green = g
- Seed Shape: Round = W; Wrinkled = w

Let's now look at a Punnett square describing the cross using our gene symbols.

Parental Genotypes *GGWW* x *ggww*

↓ ↓

Parental gametes *GW* *gw*

↘ ↙

F_1 Genotype *GgWw*

↓

F_1 Gametes *GW Gw gW gw*

Mendel's Law of Independent Assortment (cont.)

Now set up the Punnett Square for the F2 cross.

Female Gametes

Male Gametes	***GW***	***Gw***	***gW***	***gw***
GW	*GGWW* (Yellow, round)	*GGWw* (Yellow, round)	*GgWW* (Yellow, round)	*GgWw* (Yellow, round)
Gw	*GGWw* (Yellow, round)	*GGww* (Yellow, wrinkled)	*GgWw* (Yellow, round)	*Ggww* (Yellow, wrinkled)
gW	*GgWW* (Yellow, round)	*GgWw* (Yellow, round)	*ggWW* (Green, round)	*ggWw* (Green, round)
gw	*GgWw* (Yellow, round)	*Ggww* (Yellow, wrinkled)	*ggWw* (Green, round)	*ggww* (Green, wrinkled

Mendel's Law of Independent Assortment (cont.)

The phenotypes, general genotypes, and general genotypic ratios from this cross can be represented in the following manner:

Phenotype	General Genotype
9 Yellow, Round Seed	9 *G_W_*
3 Yellow, Wrinkled Seed	3 *G_ww*
3 Green, Round Seed	3 *ggW_*
1 Green, Wrinkled Seed	1 *ggww*

Mendel's Second Law

the law of independent assortment; during gamete formation the segregation of the alleles of one allelic pair is independent of the segregation of the alleles of another allelic pair

Backcross confirmation of Mendel's 2nd law

Let's use the example of the yellow, round seeded F_1.

Backcross *GgWw* x *ggww*

↓

Gametes *GW Gw gW gw*

Punnett Square for the backcross

		Female Gametes			
		GW	*Gw*	*gW*	*gw*
Male Gametes	*gw*	GgWw (Yellow, round)	Ggww (Yellow, wrinkled)	ggWw (Green, round)	Ggww (Green, wrinkled)

The phenotypic ratio of the test cross is:

1 Yellow, Round Seed; 1 Yellow, Wrinkled Seed; 1 Green, Round Seed; 1 Green, Wrinkled Seed

The Chi-Square Test (or Goodness of Fit Test)

Chi-Square Formula

$$\chi^2 = \sum \frac{(\text{Observed Value - Expected Value})^2}{(\text{Expected Value})}$$

Degrees of freedom = n-1 where n is the number of classes

The Chi-Square Test (or Goodness of Fit Test)

Let's test the following data to determine if it fits a 9:3:3:1 ratio.

315 Round, Yellow Seed
108 Round, Green Seed
101 Wrinkled, Yellow Seed
32 Wrinkled, Green
556 total seeds
Expected Values:
(9/16)(556) = 312.75 Round, Yellow Seed
(3/16)(556) = 104.25 Round, Green Seed
(3/16)(556) = 104.25 Wrinkled, Yellow
(1/16)(556) = 34.75 Wrinkled, Green

$$\chi^2 = \frac{(315 - 312.75)^2}{312.75} + \frac{(108 - 104.25)^2}{104.25} + \frac{(101 - 104.25)^2}{104.25} + \frac{(32 - 34.75)^2}{34.75}$$

number of classes (n) = 4; df = n-1 = 4-1 = 3
Enter the Chi-Square table at df = 3 and we see our chi-square value fall between 80% and 95%. Thus we accept the hypothesis that two independent genes are controlling the two traits

Extension of Mendelian analysis.

A gene can have more than two alleles

- **More than two allele are possible for any given gene**
- **Together these alleles form an allelic series**

Examples:

1. **Human Blood Types**
   - **Blood types phenotypes are - A, B, AB and O**
   - **Three alleles are possible:**
     - i^A
     - i^B
     - i

- **Alleleic series is $i^A = i^B > I$**

2. **Coat Color in Rabbits**
   - **Four phenotypes and four alleles**
   - **Allelic sereies is C > cch > ch > c**

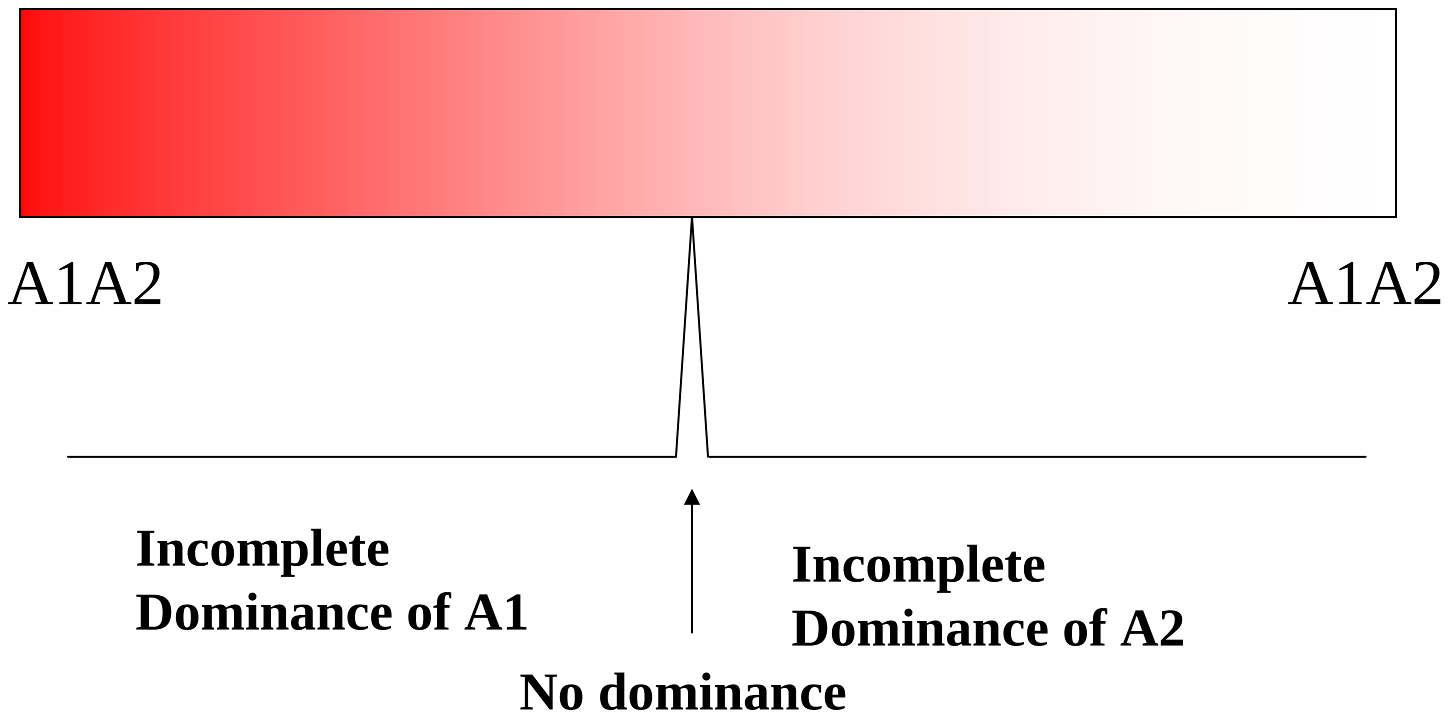
Blood Phenotype	Genotype
0	ii
A	I^AI^A or I^Ai
B	I^BI^B or I^Bi
AB	I^AI^B

Coat Color Phenotype	Genotype
Full Color	CC, Cc^{ch}, Cc^h, Cc
Chinchilla	$c^{ch}c^{ch}$, $c^{ch}c^h$, $c^{ch}c$
Himalayan	c^hc^h, c^hc
Albino	cc

Extension of Mendelian analysis

- Phenotypes of some heterozygotes reveal types of dominance other than full dominance
- Many genes have alleles that can kill the organism
- Most characters are determined by sets of genes that interact with one another and with the environment
- Modified Mendelian ratios reveal gene interaction

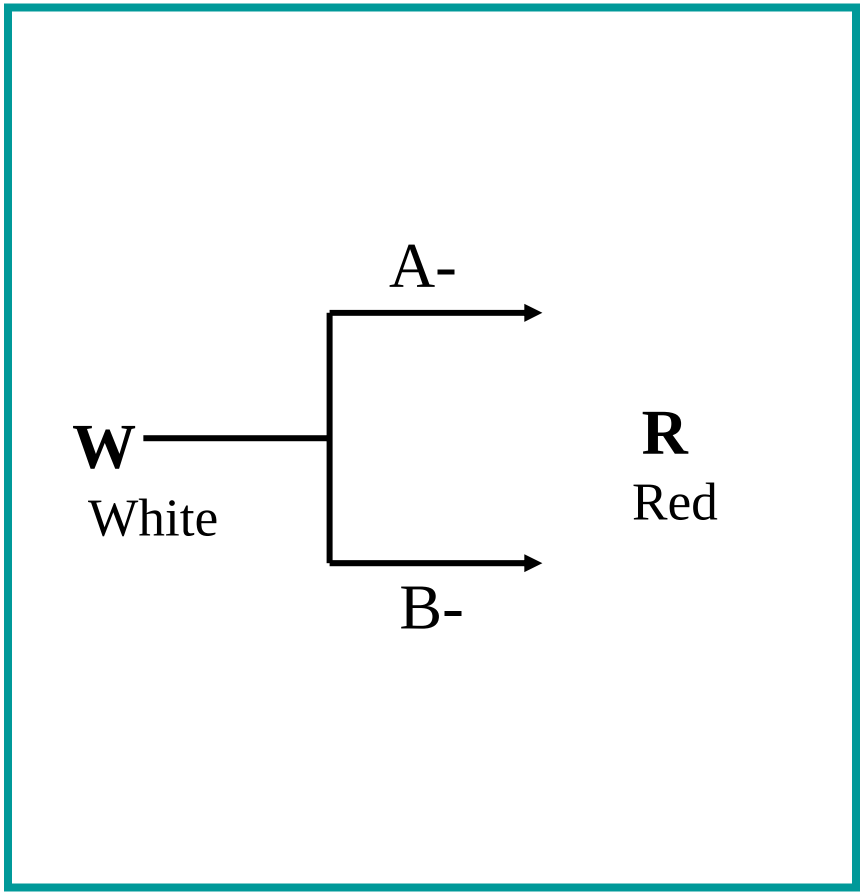
Co-dominance



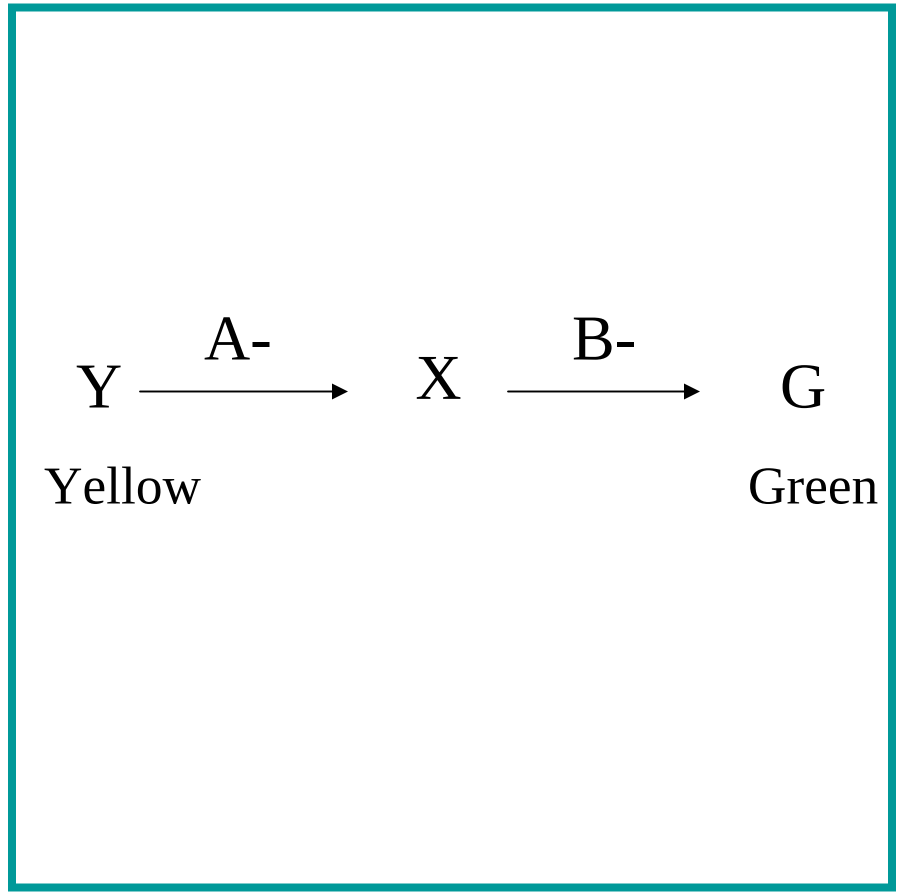
Epistasis

Type of gene interaction	9 A_B_	3 A_bb	3 aaB_	1 aabb	Phenotypic ratio
None (four distinct phenotypes)	9	3	3	1	9:3:3:1
Complementary gene action	9	7			9:7
Recessive Suppression by aa acting on bb	12		3	1	13:3
Recessive epistasis of aa acting on B and b alleles	9	3	4		9:3:4
Dominant epistasis of A acting on B and b alleles	12		3	1	12:3:1
Duplicate genes	15			1	15:1

Duplicate Genes



Complementary Genes



Extension of Mendelian analysis

Example Allelic interactions Type of epistasis

Example	**Allelic interactions**	**Type of epistasis**
Kernel Color in Wheat	*A* epistatic to *B*,*b* *B* epistatic to *A*	Duplicate genes
Sweet Pea Flower Color	*cc* epistatic to *P*,*p* *pp* epistatic to *C*,*c*	Complementary gene action
Squash Color	*W* epistatic to *G*,*g*	Dominant epistasis
Malvidin Production in *Primula*	*D* epistatic to *K*,*k*	Dominant suppression

Extension of Mendelian analysis

Modifier Genes

Example: Coat Color in Mice

- **B gene:**
  - controls coat color in mice
  - *B* allele (black coat color) is dominant to the *b* allele (brown coat color)
- **D gene**:
  - controls intensity of expression of the *B* gene
  - *D* allele (gives full color) is dominant to the *d* allele (conditions dilute color expression)

Genotype	Phenotype
9 *B_D_*	black
3 *B_dd*	dilute black
3 *bbD_*	brown
1 *bbdd*	dilute brown

The *D* gene does not mask the effect of the *B* gene, rather it modifies its expression.

Modifier gene - a gene that has small, quantitative effects on the level of expression of another gene

Extension of Mendelian analysis

Variation in Gene Expression
***P* Gene:**
Controls extra digit (toe or fingers) expression in humans
The dominant allele *P* produces polydactyly in humans

Parental Cross Normal x Normal

↓

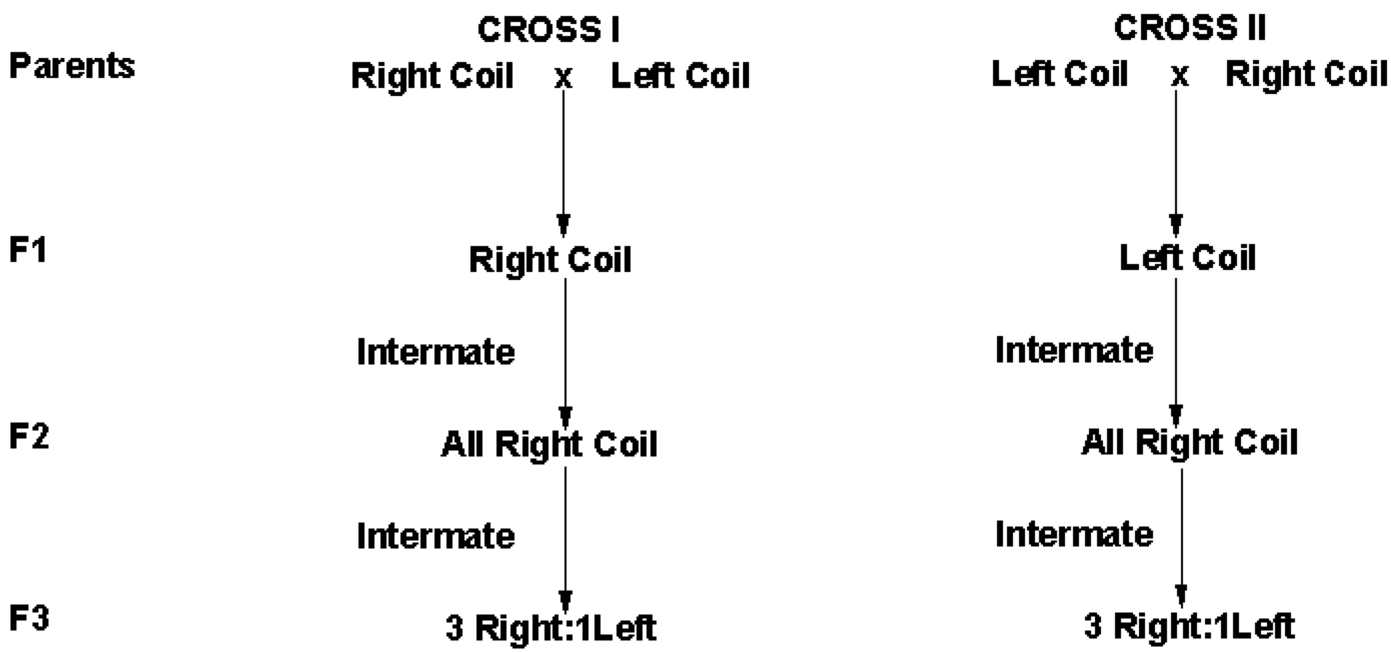
Progeny Some children with extra digits

Penetrance - the frequency of expression of an allele when it is present in the genotype of the organism

Expressivity - variation in allelic expression when the allele is penetrant

Maternal Inheritance and Maternal Effects

- **Genetics of Maternal Effects**
- **Snail Shell Coiling and Maternal Effects**
- This is an example of a pair reciprocal crosses.
- **Reciprocal Crosses** - a pair of crosses where each of the alternate phenotypes are the female in one cross



Maternal Inheritance and Maternal Effects

Variegation in four o'clock plant and maternal inheritance

- Experiments were performed by Correns on the four o'clock plant
- The plant has green, variegated (white and green) or white leaves
- Normal flowers develop at different locations on the plant
- Crosses were made among the flowers associated with each leaf color (results in table below)

Female	Male	Progeny Phenotype
Green	Green, variegated or white	Green
Variegated	Green, variegated or white	Variegated
White	Green, variegated or white	White

Maternal Inheritance and Maternal Effects

- **Results**
- The progeny cross always exhibited the color of the leaf of the female
- Trait expresses **maternal inheritance**

- **Maternal inheritance** - the female phenotype in a cross is always expressed in its offspring

- **Explanation**
- Is a result of the expression of a gene found in the genome of an organelle
- For most species, all of the organelle DNA that is found in an embryo is from the female.
- This is the biological basis of maternal inheritance.