Lecture 2:

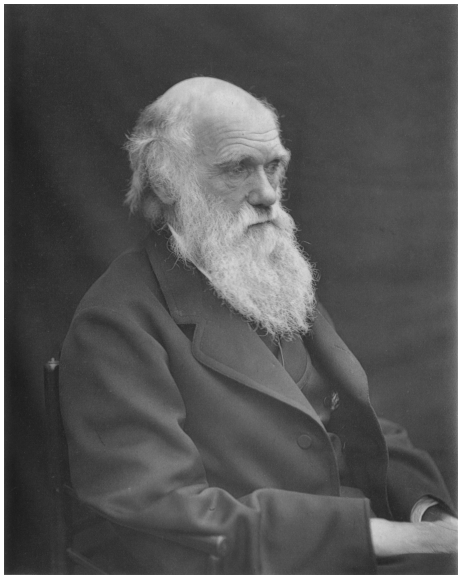
Genetics Traditions

Course 410

Molecular Evolution

Traditions of Genetics

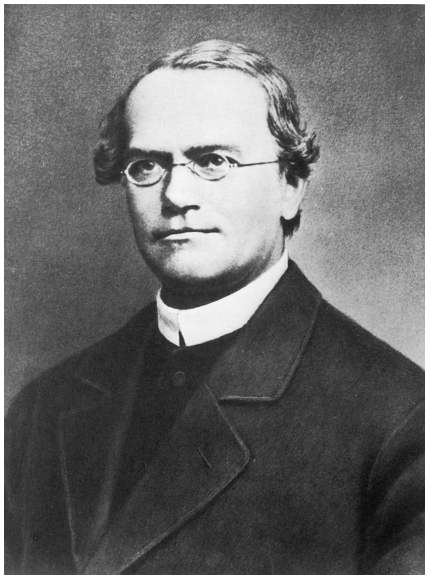
Darwin



Natural selection of small changes

Evolution and Population Genetics

Mendel



Transmission of large differences

Analytical Genetics

Traditions of Genetics

Darwin → Biometricians


Pearson


Yule

Mendel → Mendelists

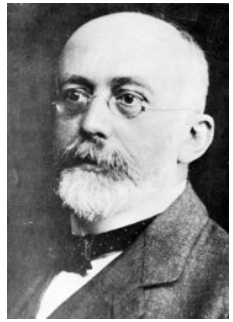

Punnett


Bateson

Yule to Bateson (1902)

"one cannot help feeling that the speculations would have had more value had he kept his emotions under better control; the style and method of the religious revivalist are ill-suited to scientific controversy" (Edwards, 2008)


Hardy

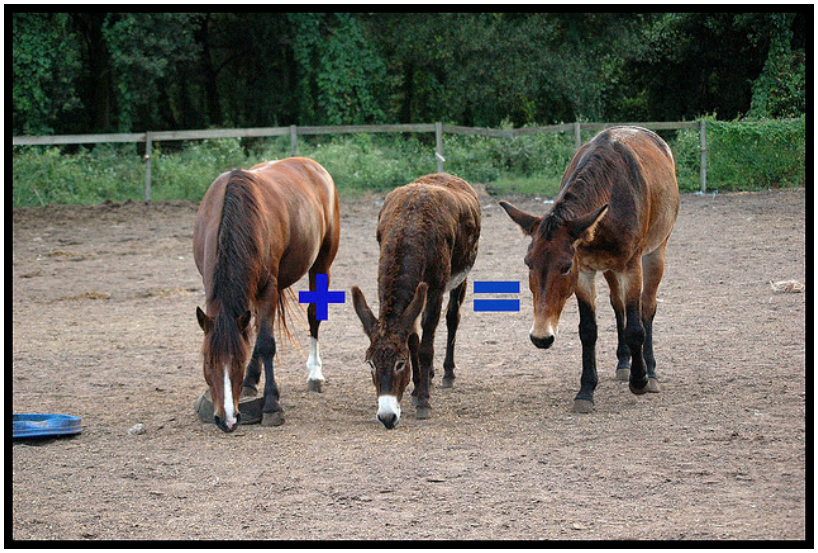

Weinberg

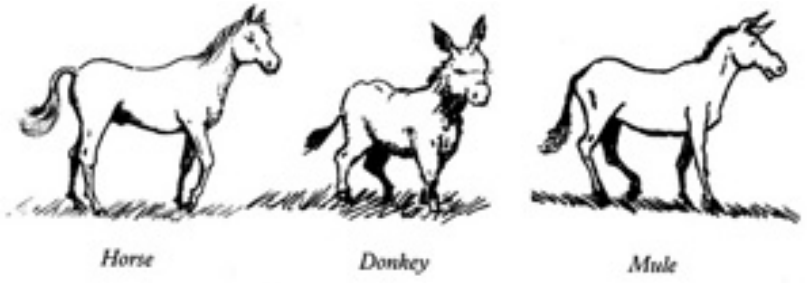
Mendel and the general theory of inheritance and basic laws

Scientific motives

The chief motives to understand heredity and the bases of it were:

1. Speciation
2. Hybridization
3. Similarities between parents and offsprings





Mare (Female Horse) *Equs caballus* 2n = 64 n = 32 X **Jackass** (Male Donkey) *Equs asinus* 2n = 62 n = 31 = **Mule** 2n = 63 n = ?

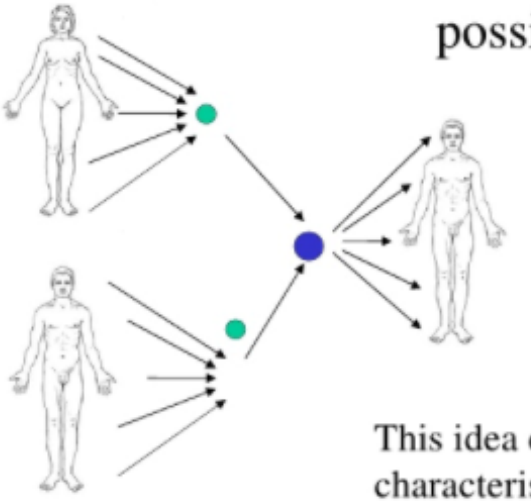
Stallion (Male Horse) X **Jennyass** (Female Donkey) = **Hinny**

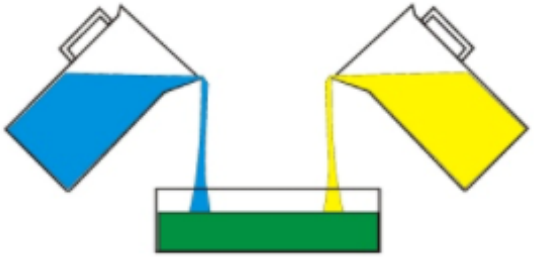
What were the theories of inheritance at the time?

Theories of inheritance

Before Mendel the only proposed theory of inheritance was "**blending inheritance**"

Pangenesis and the Blending of Bloods: An early hypothesis of how traits are passed from one generation to the next. Pre-Mendel, this was a popular idea, and Darwin even considered it a possibility!





This idea depends on the true mixing of characteristics from each parent.

Theories of inheritance

How traits are passed on?

Two hypotheses

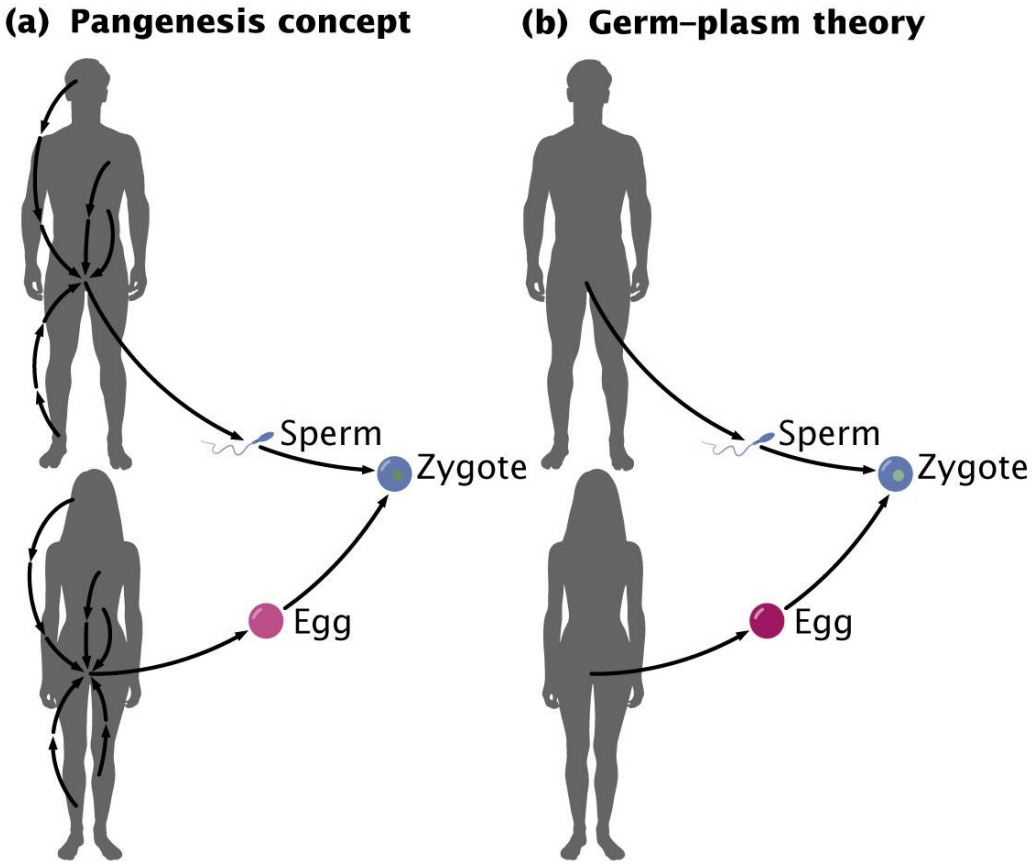
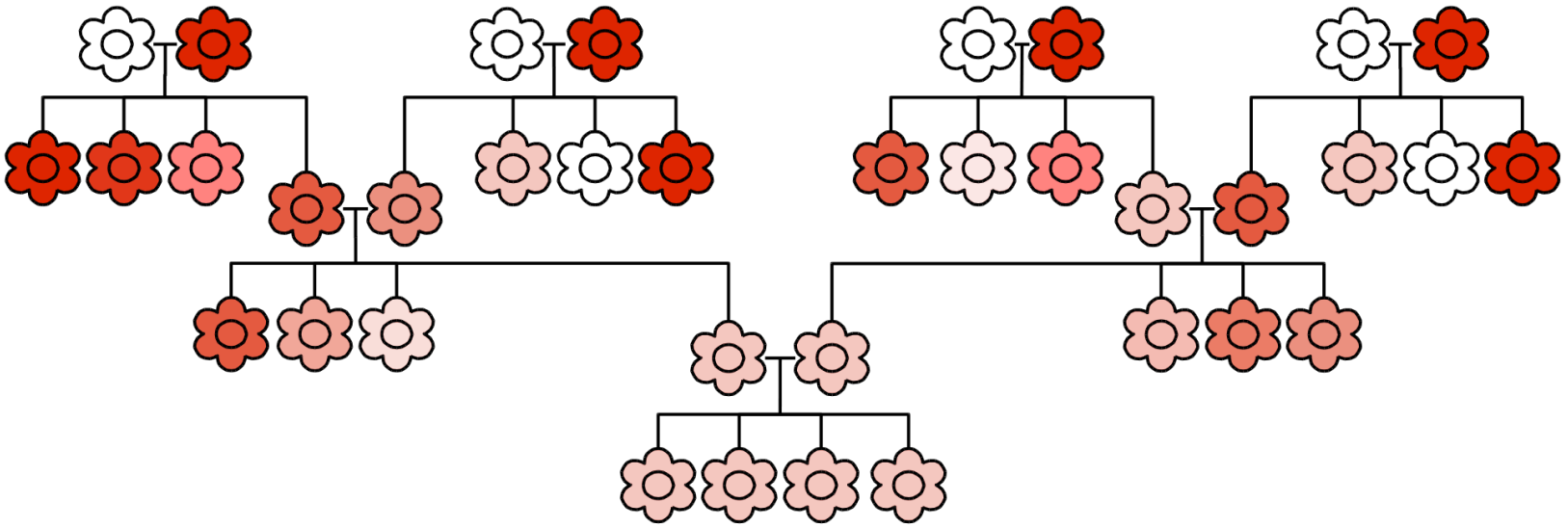


Fig_01-14 *Genetics, Second Edition* © 2005 W.H. Freeman and Company

Theories of inheritance

What happens to characters when they are blended every generation?

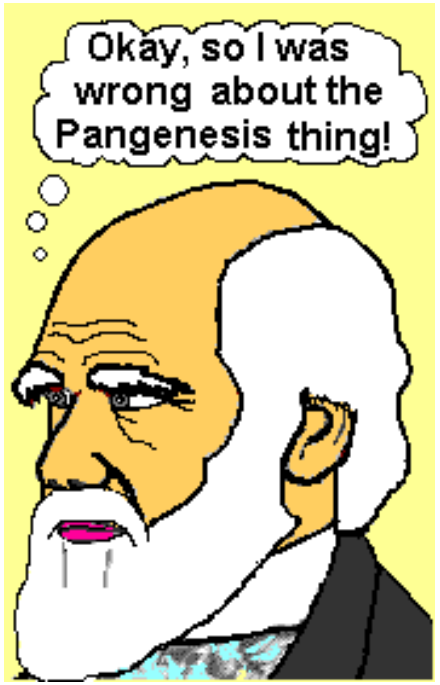


Theories of inheritance

Darwin's hypothesis of Pangenesis

"Gemmules" travel from every part of the body to the reproductive system to pass the traits to future generation.

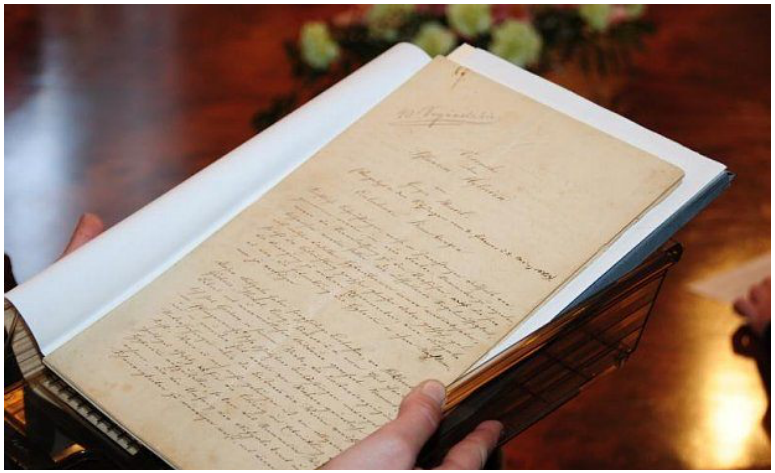
Hypothesis **NOT** supported by scientific evidence.



O'Connor, C. & Miko, I. (2008) Developing the chromosome theory. Nature Education 1(1):44

Mendel and his peas

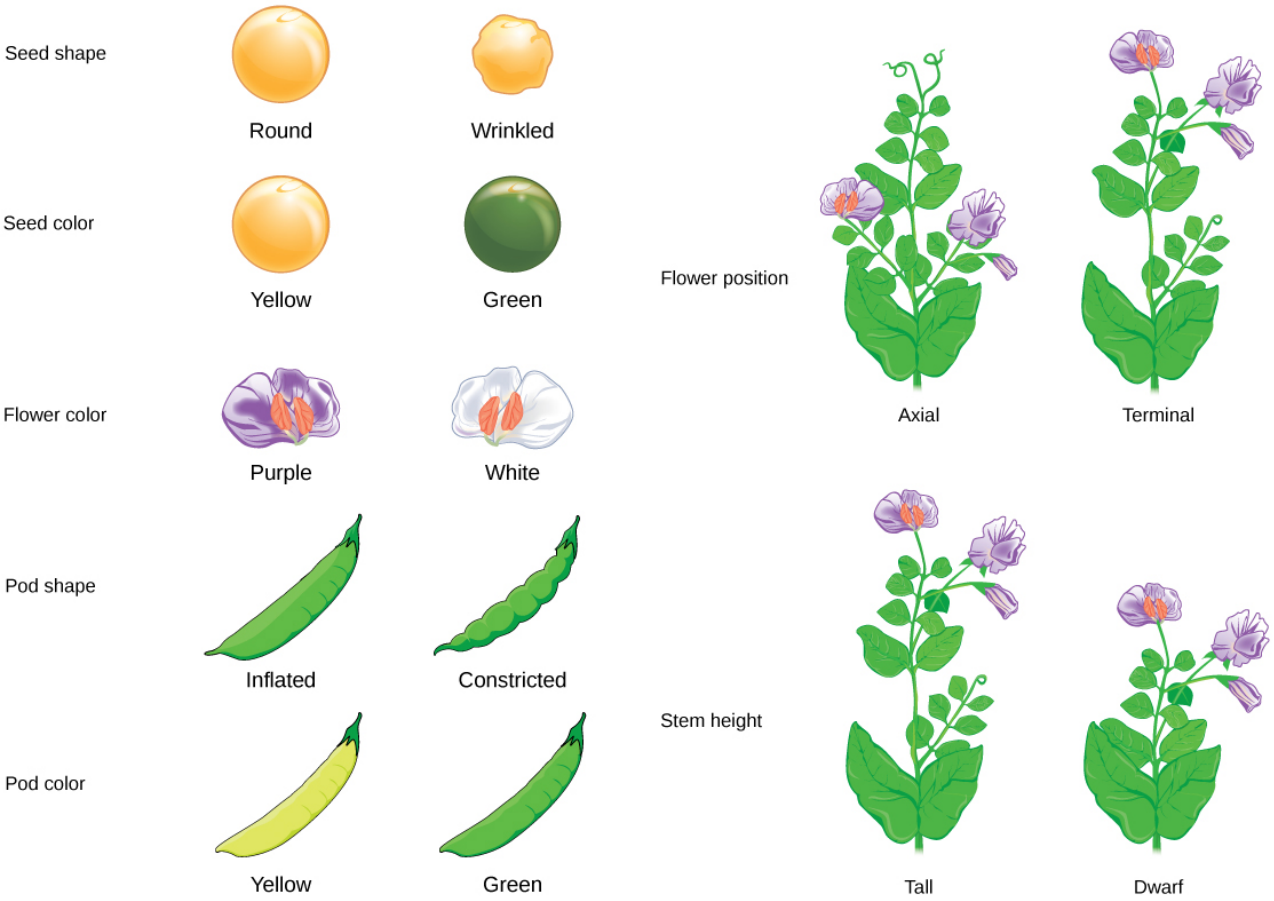
Gregor Mendel (Johann) studied heredity by the systematic breeding experiments of garden pea (*Pisum sativum*)



Mendel's Peas

Why Pea plants?

Clear and distinct visual Traits/characters

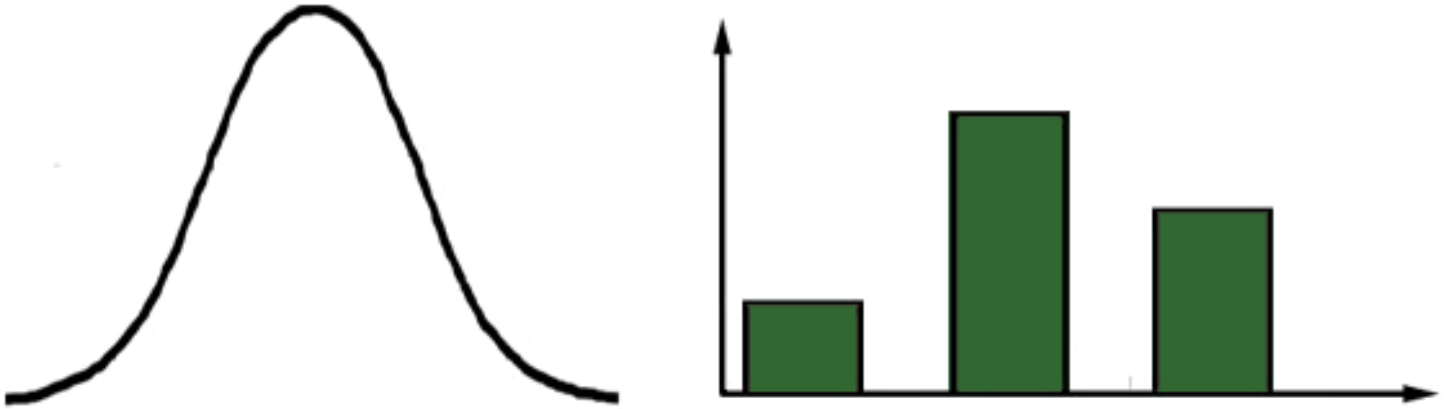


What kind of traits/characters are these?

Why should we care?

Traits and characters

Traits can be continuous or discontinuous (also called discrete)



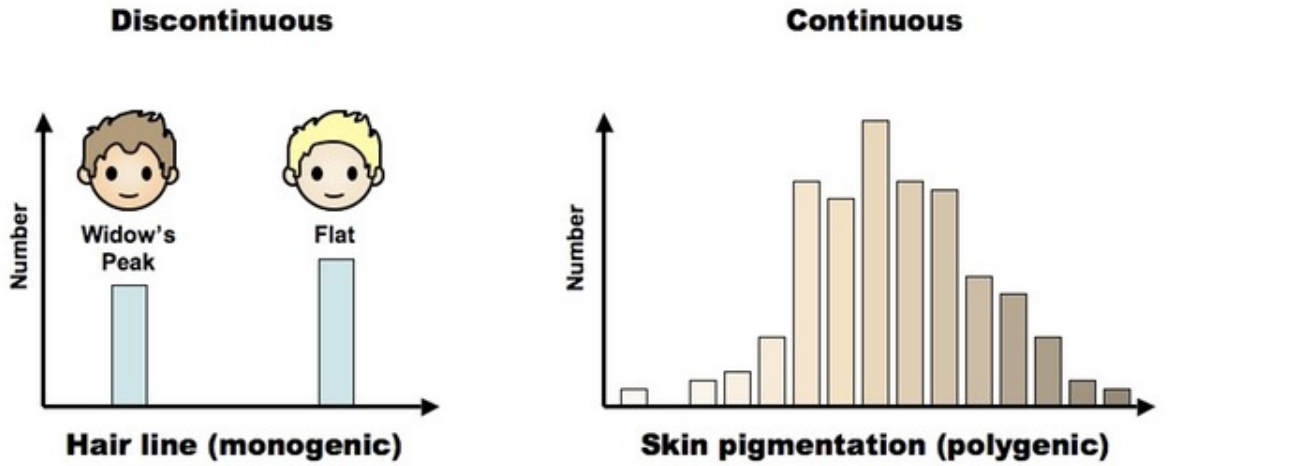
Continuous Variation

- No distinct catagories
- Tends to be quantitative
- Controlled by a lot of genes
- Strongly influenced by the environment

Discontinuous Variation

- Distinct catagories
- Tends to be qualitative
- Controlled by a few genes
- Unaffected by the environment

Traits


Discontinuous
Number
Widow's Peak
Flat
Hair line (monogenic)
Continuous
Number
Skin pigmentation (polygenic)


4:10
5:0
5:1
5:2
5:3
5:4
5:5
5:6
5:7
5:8
5:9
5:10
5:11
6:0
6:1
6:2
6:3
6:4
6:5

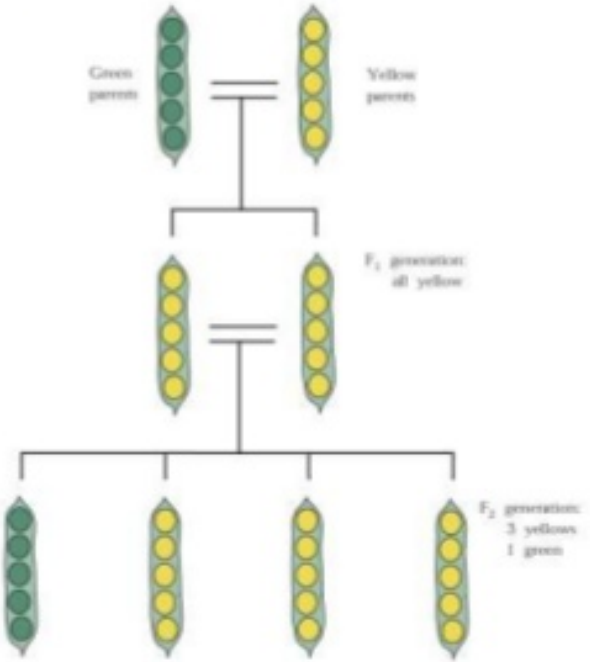
OK!?

What is the connection to Darwin and Mendel?

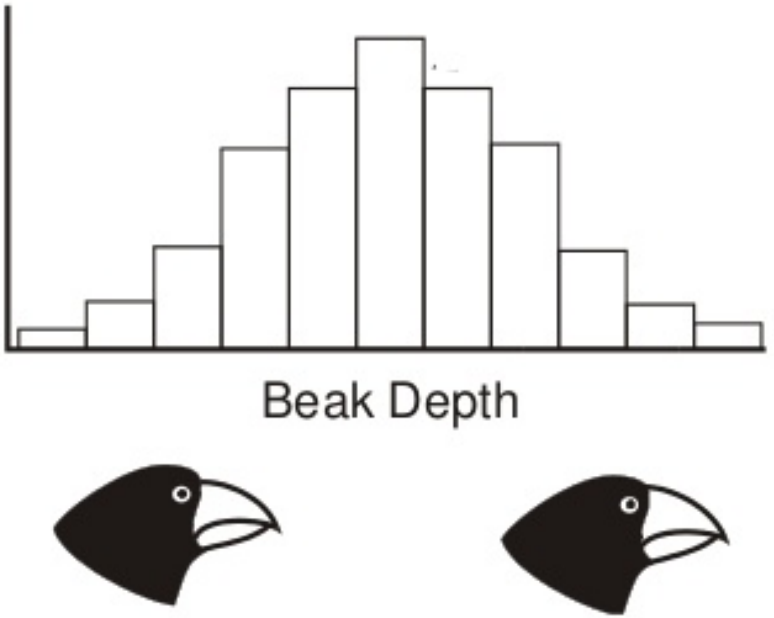
Mendel's characters vs. Darwin's

Mendel focused on variation of large effect while Darwin observed small variations that affect fitness

Mendel studied discontinuous (discrete) traits

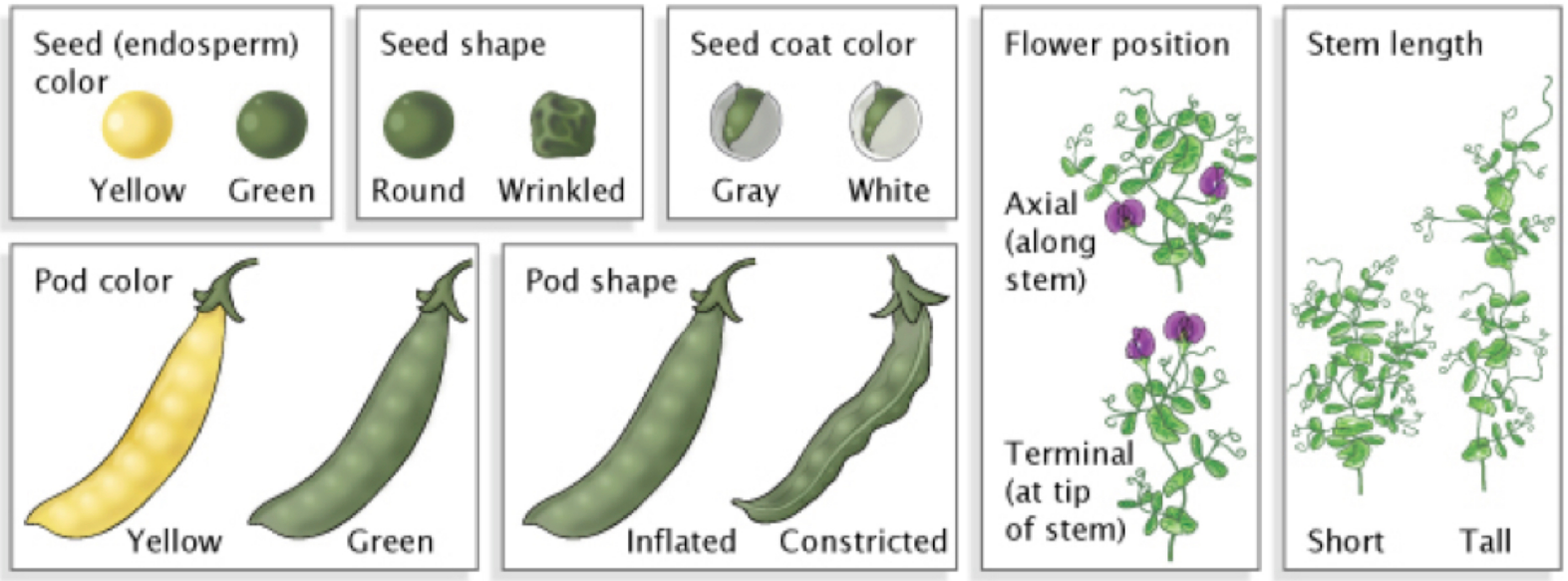


Darwin observed continuous variation



Mendel's discrete characters

Mendel chose seven discrete characters that can be easily be visualized and identified.

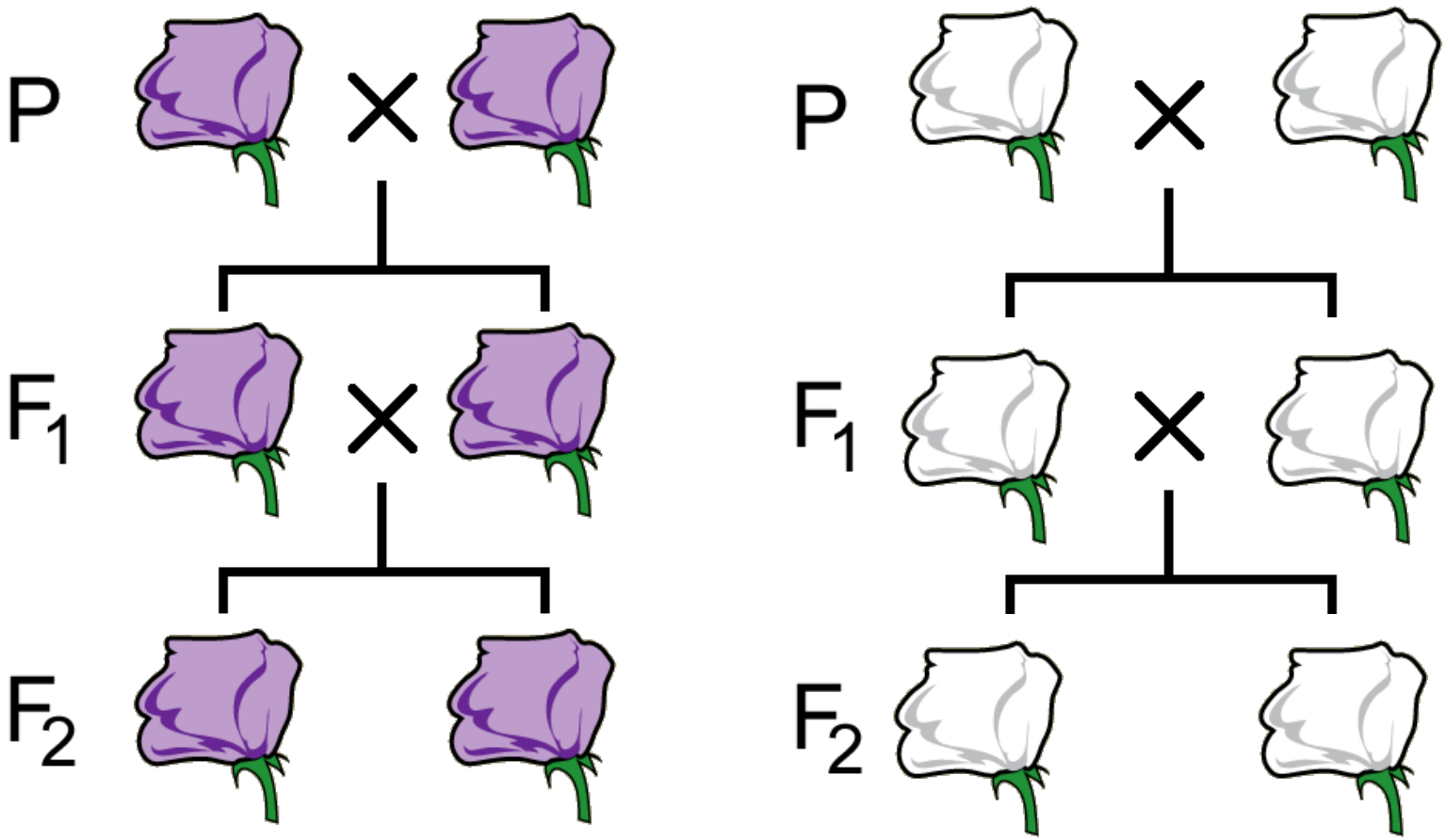


Miko, I. (2008) Gregor Mendel and the principles of inheritance. Nature Education 1(1):134

Mendel's pure single trait lines

1) Establish pure lines of each character

Which characters?



What are pure lines?

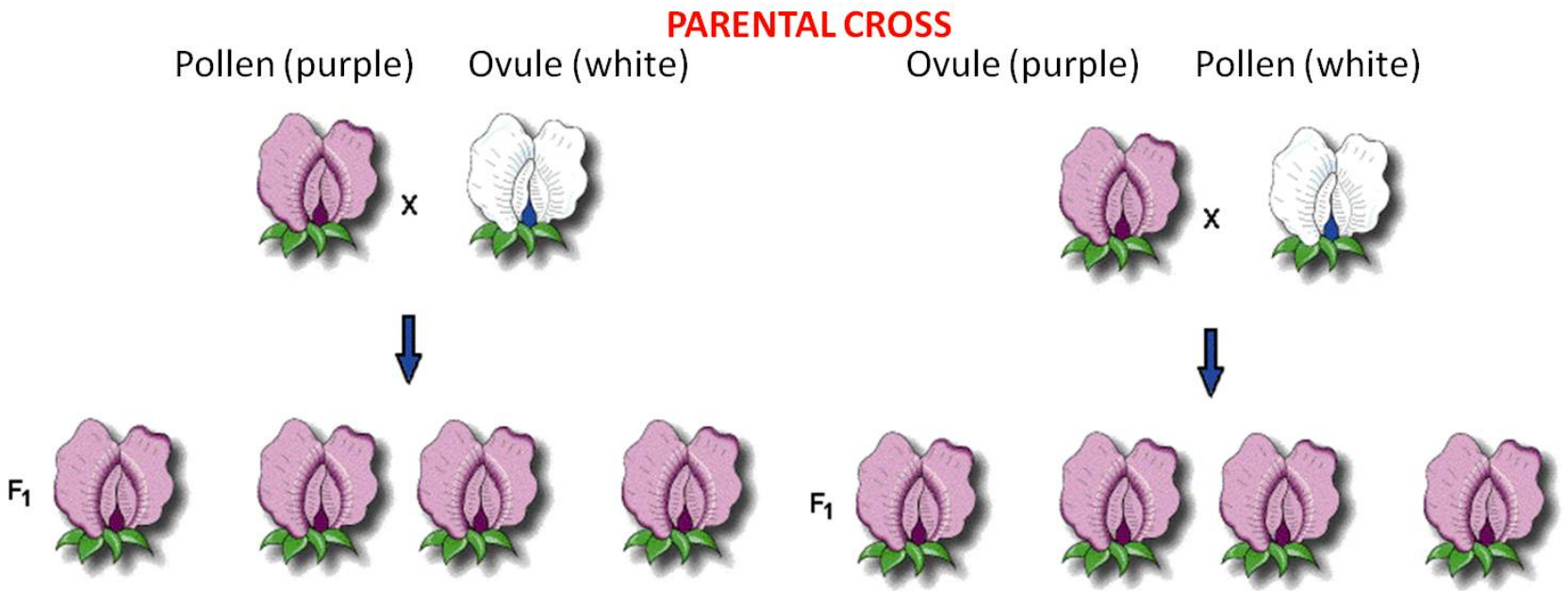
Homozygous?

Identical by state?

First generation

2) Cross breed the pure lines.
3) Resulting plants are hybrids.
4) Inspect the phenotypes of the first generation.

How do we inspect the phenotype?
Why?



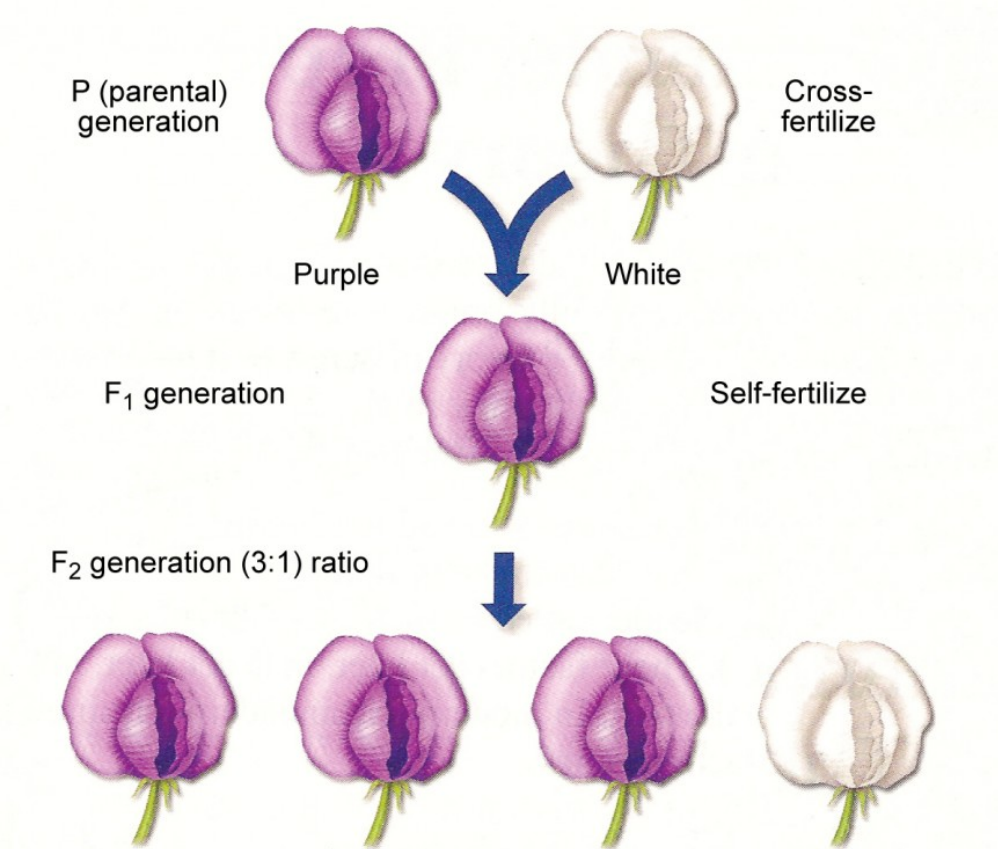
First generation

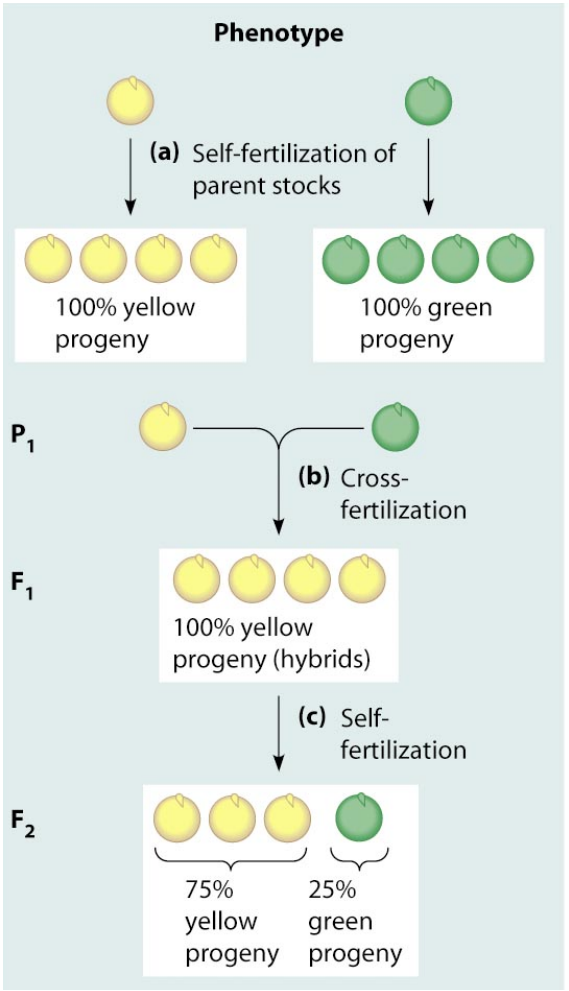
Observations and findings:

- All resulting plants exhibits the phenotype of one of the parents.
- One of the parental phenotypes disappears in the first hybrid generation.

Second generation

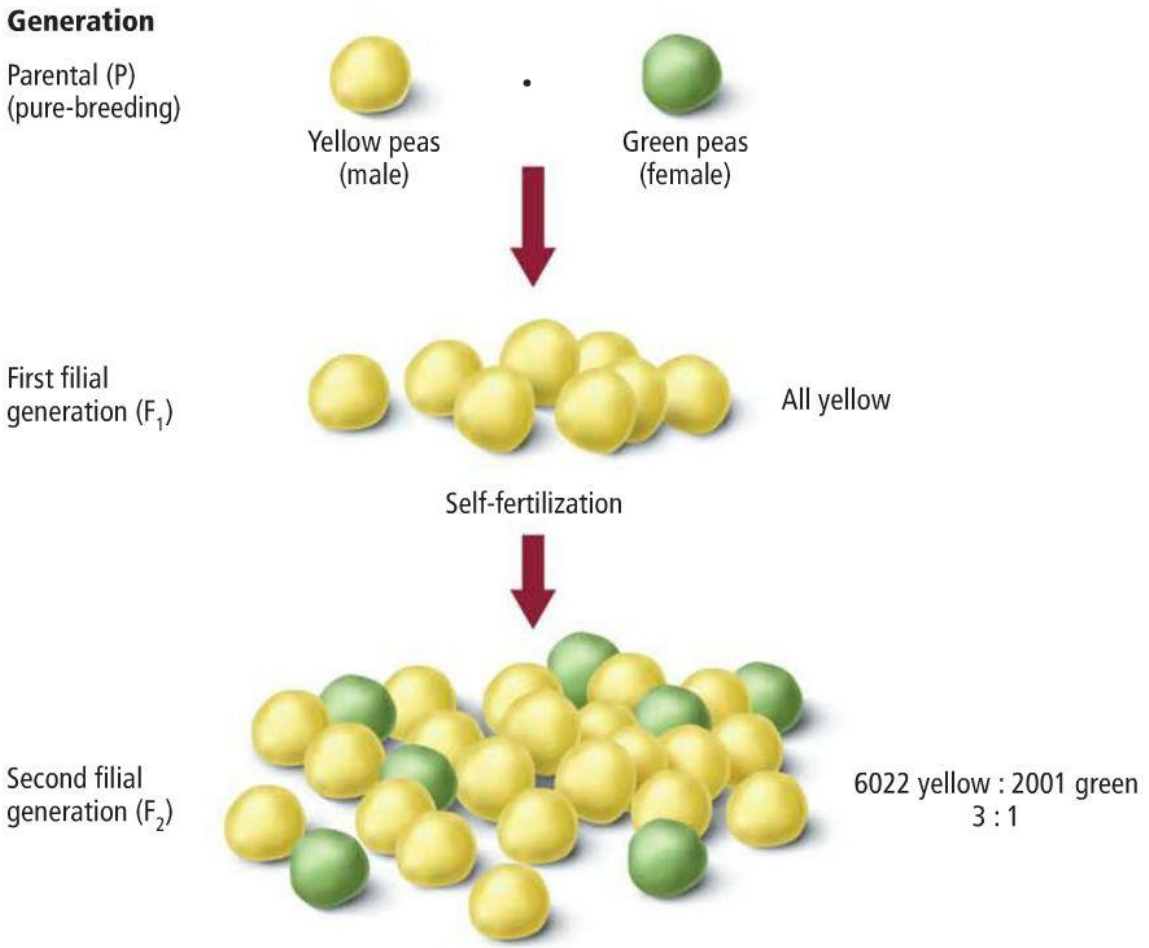
5) Self cross the F1 individuals.
6) Inspect the phenotypes of the resulting F2 generation.



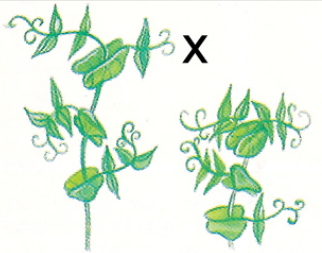


Second generation

How did Mendel inspect the phenotypes of the F2 generation?



Mendel's monohybrid results

Trait	Dominant vs. recessive	F_2 generations Dominant form	Recessive form	Ratio
Flower color	 Purple X White	705	224	3.15:1
Seed color	 Yellow X Green	6022	2001	3.01:1
Seed shape	 Round X Wrinkled	5474	1850	2.96:1
Pod color	 Green X Yellow	428	152	2.82:1
Pod shape	 Round X Constricted	882	299	2.95:1
Flower position	 Axial X Top	651	207	3.14:1
Plant height	 Tall X Dwarf	787	277	2.84:1

Mendel's Monohybrid Experiment

Observations and findings:

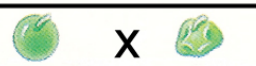
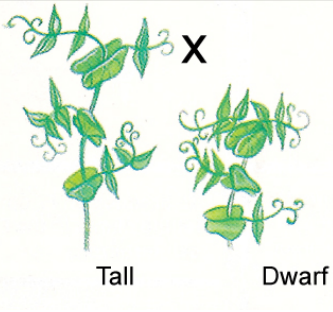
- The selfing of the first generation results in the reappearance of one of the parents' characteristics.
- A factor/particle is within the plant that results in the appearance of the plant.
- Both male and female contribute equally to the phenotype.
- The absence or appearance of a specific character depends on the combination of factors.

Factor's type

Observations and findings:

- The "factor" that appeared in all individuals of the first generation is the "**dominant**" factor.

- The "factor" that disappeared in the first generation is the "**recessive**" factor.

Trait	Dominant vs. recessive
Flower color	
Seed color	
Seed shape	
Pod color	
Pod shape	
Flower position	
Plant height	

Genotype

- The P generation is a pure bred contains each with two factors of the same type (**homozygous**).
- The F1 generation is a hybrid and as a result contains two different “factors” one from each of the parents (**heterozygous**).

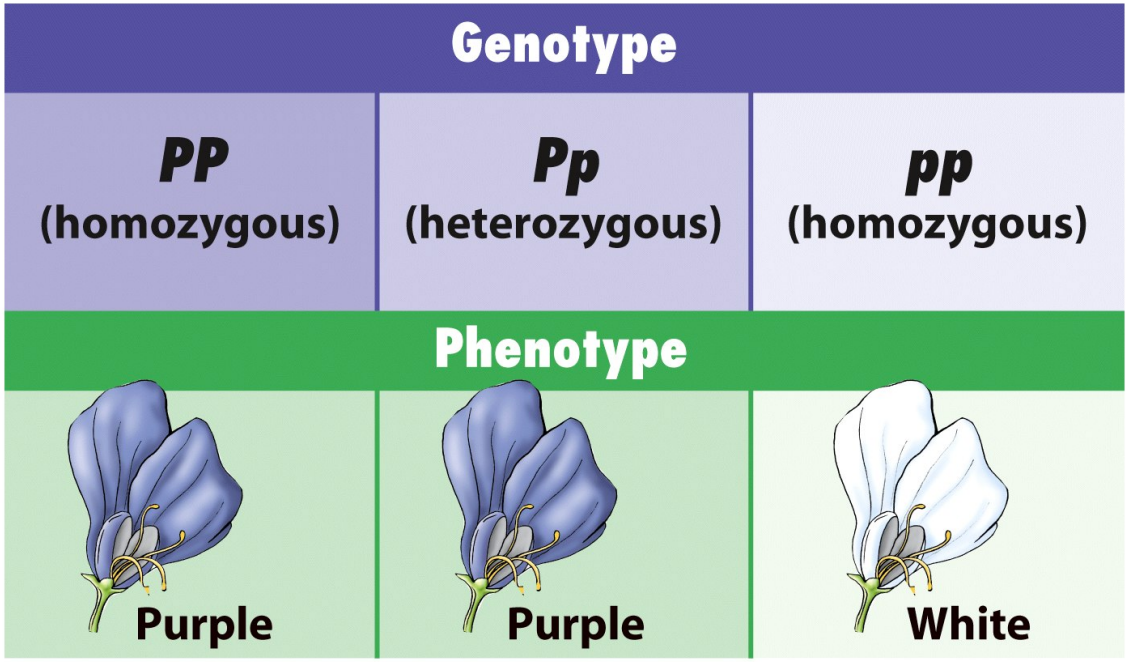
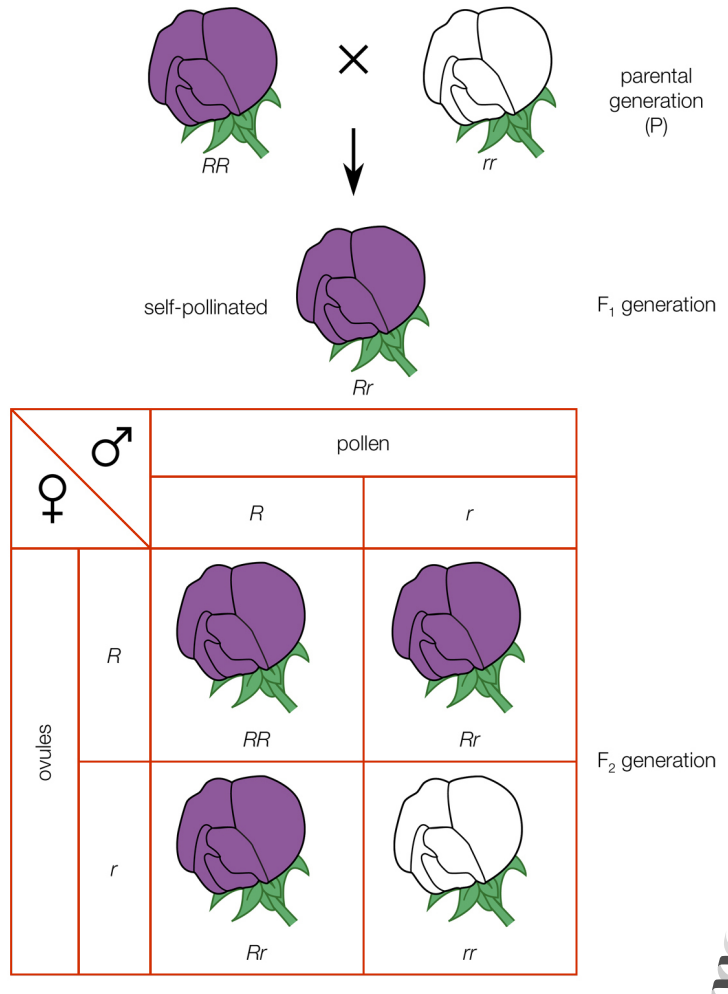


Figure 10-3 Discover Biology 3/e

Mendel's 1st law

- "Factors" within a plant separate during the formation of gametes.
- "Factors" unite during fertilization randomly.
- The phenotype of resulting union is determined by the combination of factors.

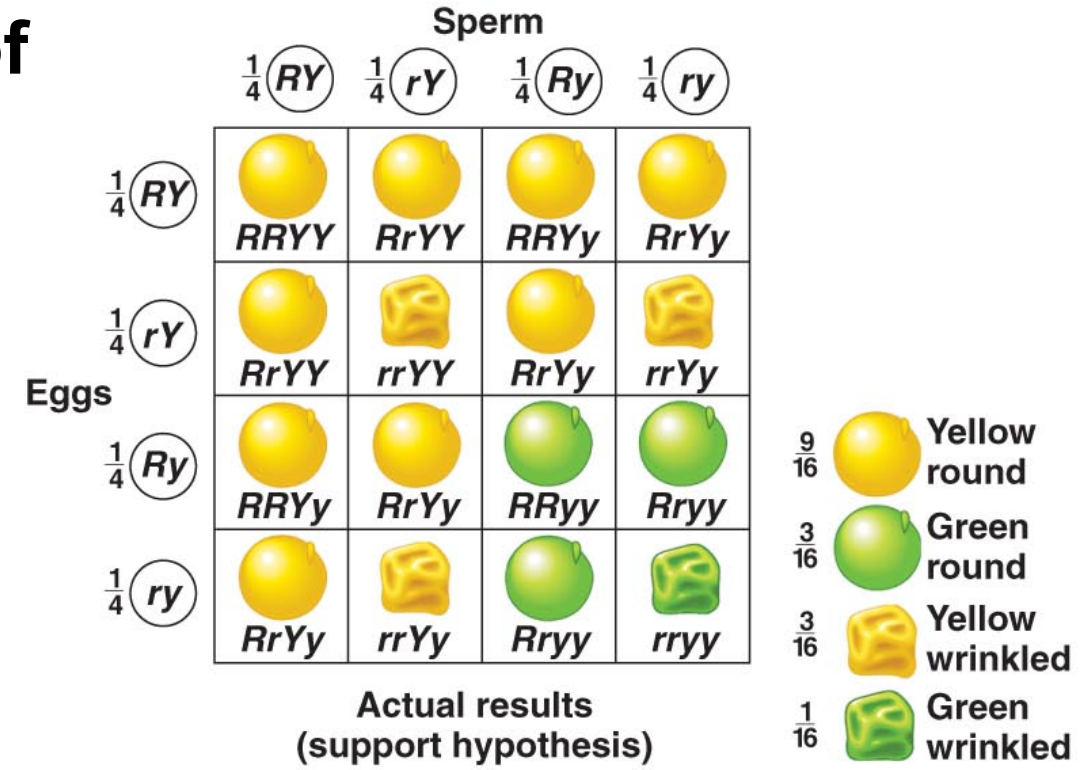
Segregation of factors (alleles)



Mendel's 2nd law

What is the ratio of each phenotype independently?

- Yellow : Green
  3:1
- Round : Wrinkled
  3:1



- Each factor segregate independently.

Independent Assortment

Review

- What is “dominant” and “recessive” a description of?
- What is a phenotype?
- What is a genotype?
- What is a homozygous?
- Identical by state?
- What is a heterozygous?
- Different by state?
- Did Mendel observe or infer genotype?
- Did Mendel observe or infer phenotype?

Genes and Genotype

It appears as most simple to use the last syllable 'gen' taken from Darwin's well-known word pangene.... Thus, we will say for 'das pangene' and 'die pangene' simply 'Das Gen' and 'Die Gene.

(Wilhelm Ludvig Johannsen)

that Johannsen's pure lines were exactly the same as his own 'elementary species'. Shull reviewed Johannsen's claims and concluded that 'if sustained by further research' they would certainly constitute an important new principle.[22]

Johannsen also introduced the term 'gene' and gave genetics two very useful terms: 'genotype', which refers to the full set of genes an organism carries; and 'phenotype' which refers to its external features, everything from size and colour to behaviour. As we saw from Mendel's original pea experiments, a pea plant with yellow peas (the yellow phenotype) might have either two copies of the yellow version of the gene (the yellow allele), or one yellow and one green; from the phenotype alone, it was impossible

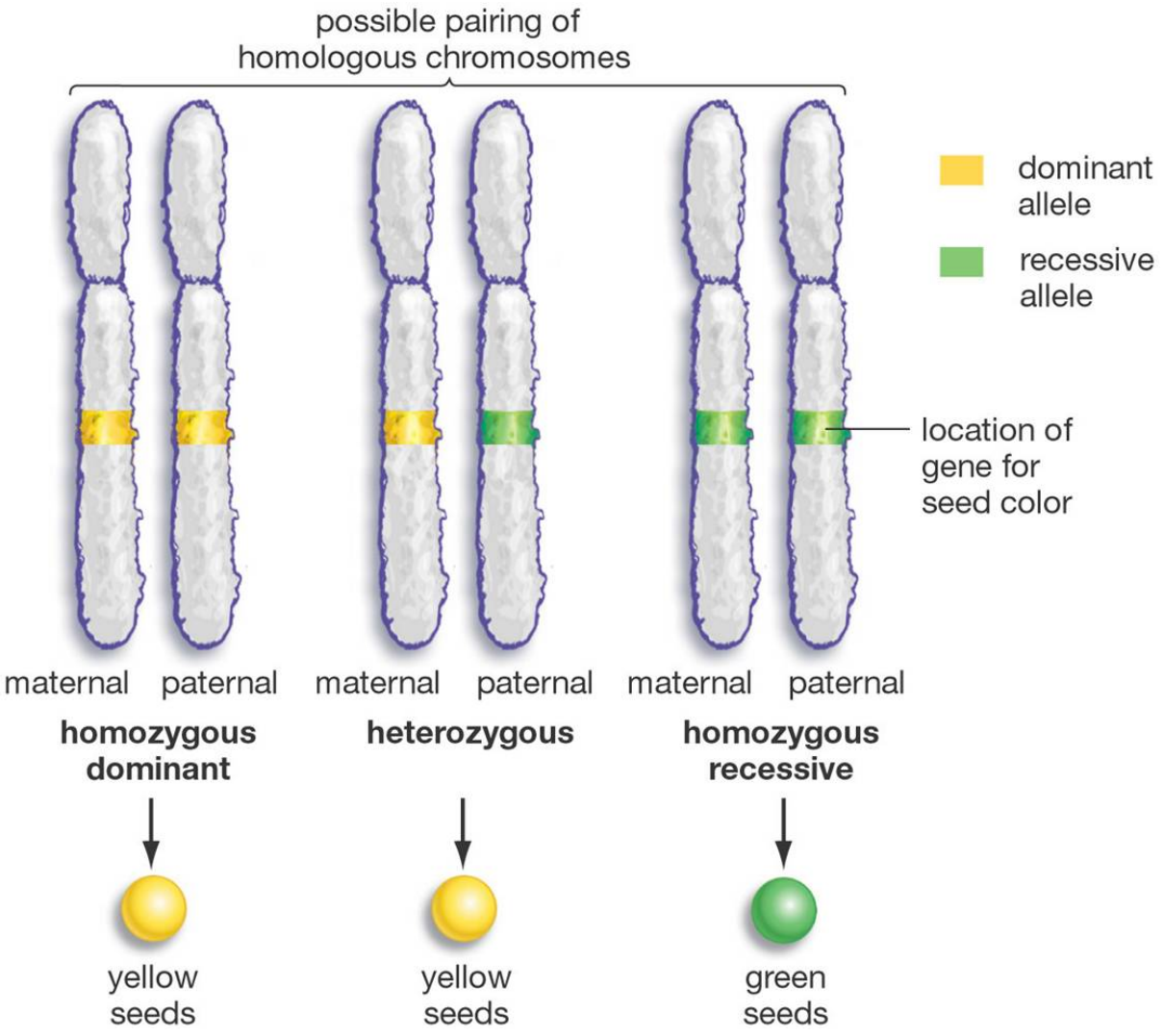
What is an allele?

Alleles are Mendel's factors that he could not see but infer by crosses

Do not get it?

They are the (A) and (a) that are being passed into gametes and unite to give the genotype of an individual.

What is an allele?



Alleles on chromosomes?

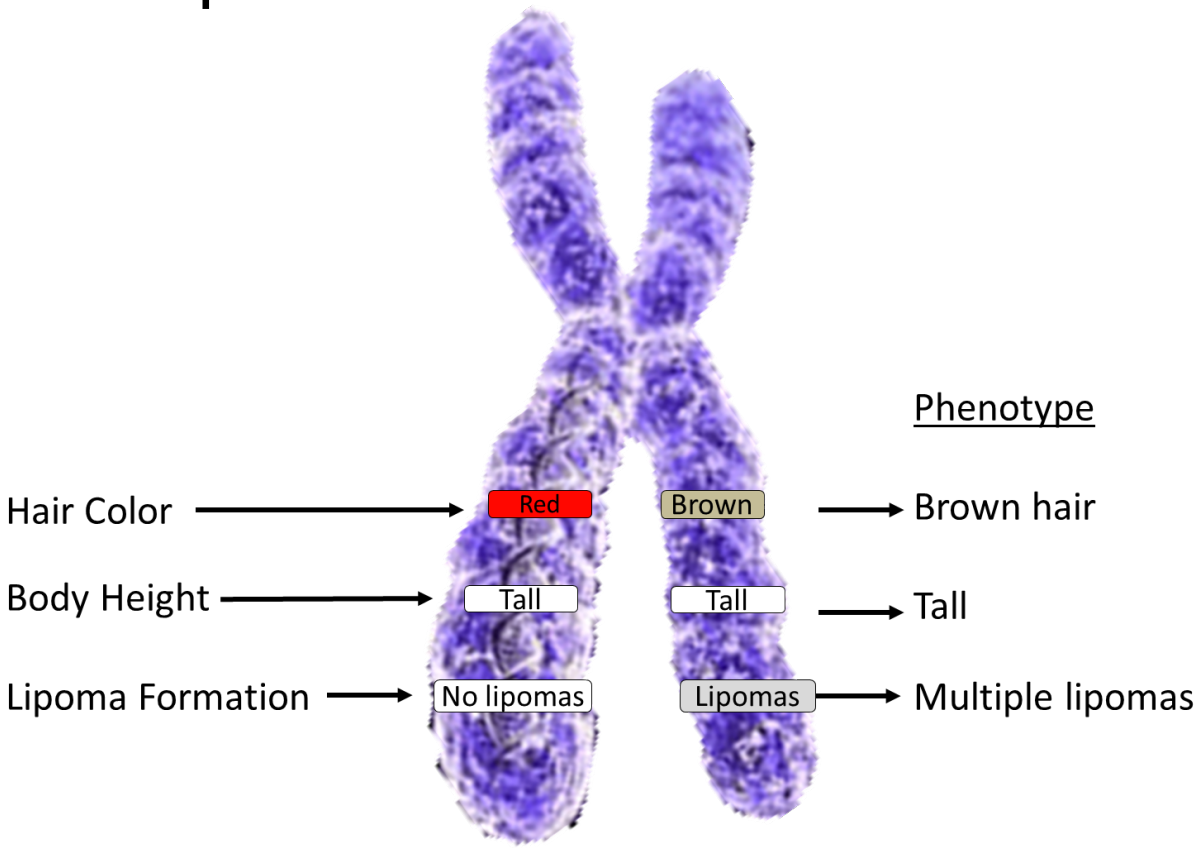
Where?

Same location?

Same chromosome?

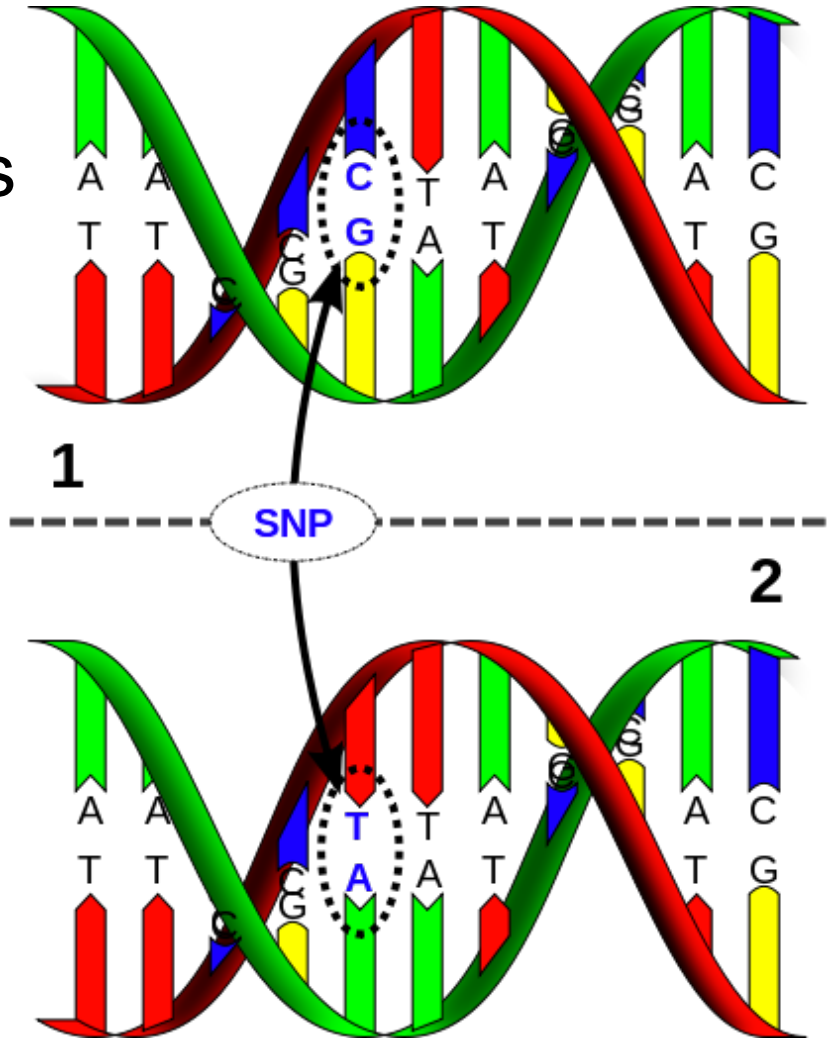
Locus

- A specific location in the genome is called **locus** (plural **loci**).
- Alleles at the same locus are inherited each from one parent.



Alleles at a locus

- DNA at a specific locus may differ in one individual.
- How?
- What are the alleles in the figure?

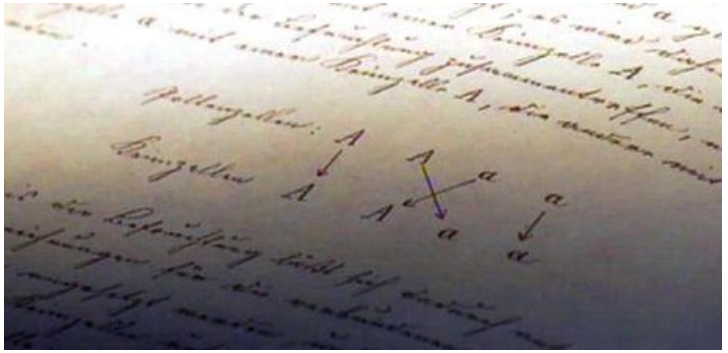


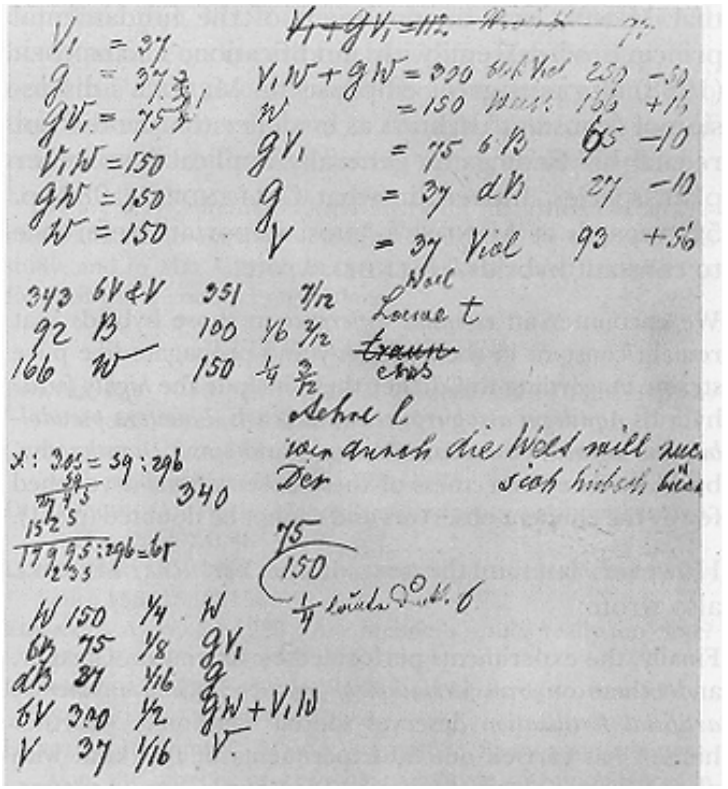
Mendel's work

Important contributions by Mendel to biology:

1. Genotypic notation.

2. Quantitative framework.





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