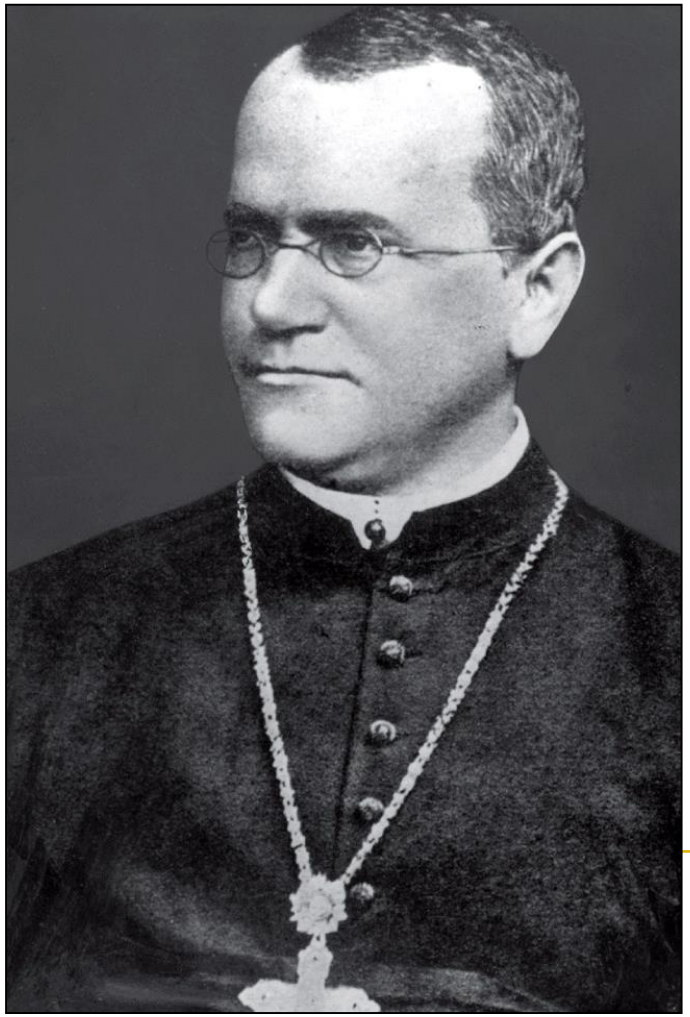
Mendelian Genetics I



Dr. Heng Chooi

Suggested Reading:
Pierce Genetics: A Conceptual Approach (4th edition 2012; 5th edition 2014)
Chapters 1 (background reading), 3

Slides adapted from Assoc. Prof Martha Ludwig's lecture

Learning Outcomes

Following this lecture you should:

- know the major factors responsible for phenotypic variation within a population of organisms.

- understand the significance of Mendel using pure lines in his experiments.

- know the relationship between the P, F_1 and F_2 generations.

- know how the 3:1 phenotypic and 1:2:1 genotypic ratios observed from a monohybrid cross are related.

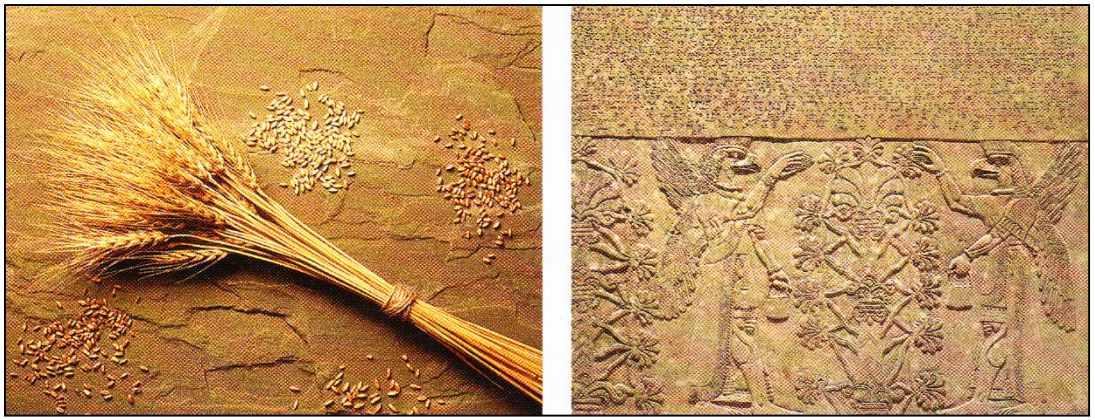
- be able to describe a testcross and why it is used.

- know and understand Mendel's First Law.

- be able to define genetic dissection, homozygote, heterozygote, recessive, dominant.

Genetics - the discipline of biology that studies heredity and variation

- practiced long before there was any knowledge of chromosomes or genes
- domestication of plants and animals ≥10,000 years ago, e.g. wheat
- sophisticated breeding techniques ~4000 years ago, e.g. dates



modern wheat

- result of inbreeding at least three different wild species
- seeds larger and do not scattered before harvest

date palms

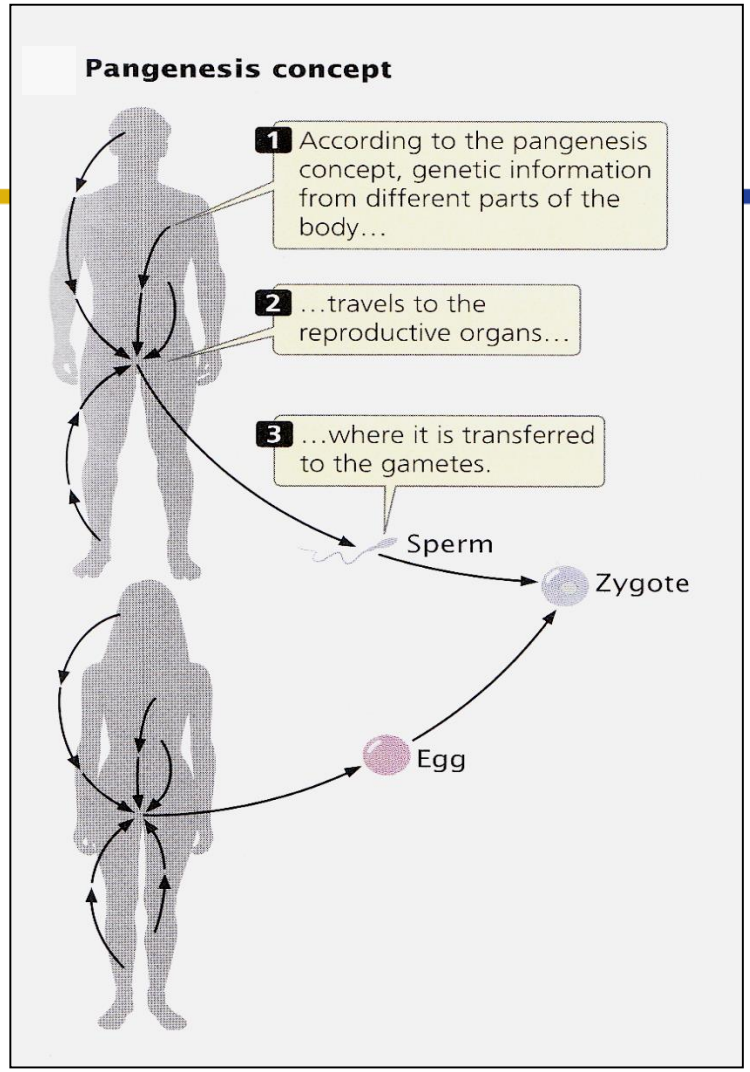
- Assyrians and Babylonians developed several hundred varieties using artificial pollination
- differed in fruit size, colour, taste, time of harvest

Pierce (2012) *Genetics: A Conceptual Approach*, p 7

Human heredity

-pangenesis

- •520 B.C. until late 1800s
- •specific particles (gemmules)
  - •carried information from various parts of the body to the reproductive organs
  - •information then passed into the embryo
- •inheritance of acquired characteristics, e.g. musical ability

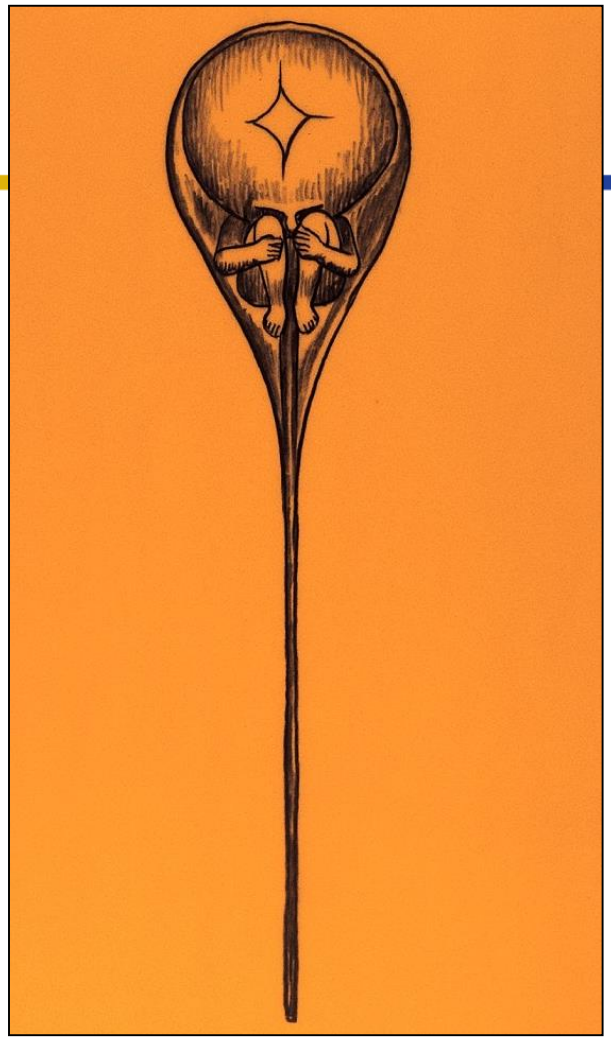


Pierce (2012) *Genetics: A Conceptual Approach*, p 8

Human heredity

-preformationism

- •17th and 18th centuries
- •homunculus: tiny miniature adult inside the sperm (spermists) or egg (ovists), enlarges during development
- •inheritance from only one parent



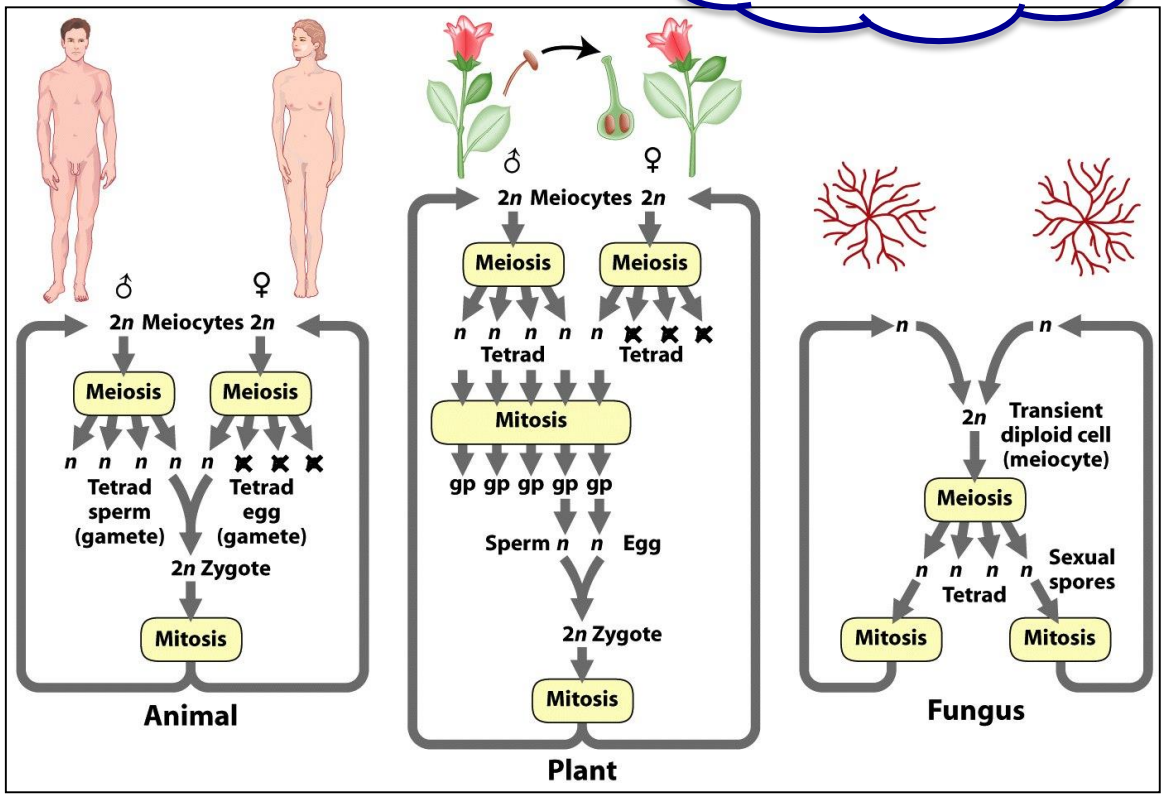
Pierce (2012) *Genetics: A Conceptual Approach*, p 9

Inheritance in eukaryotes
- hereditary material (genes encoded by DNA) is passed to progeny via gametes or sexual spores, which result from meiosis in specific parental cells

Practical 1B!

Determining which genes are responsible for a biological process or trait is called genetic dissection

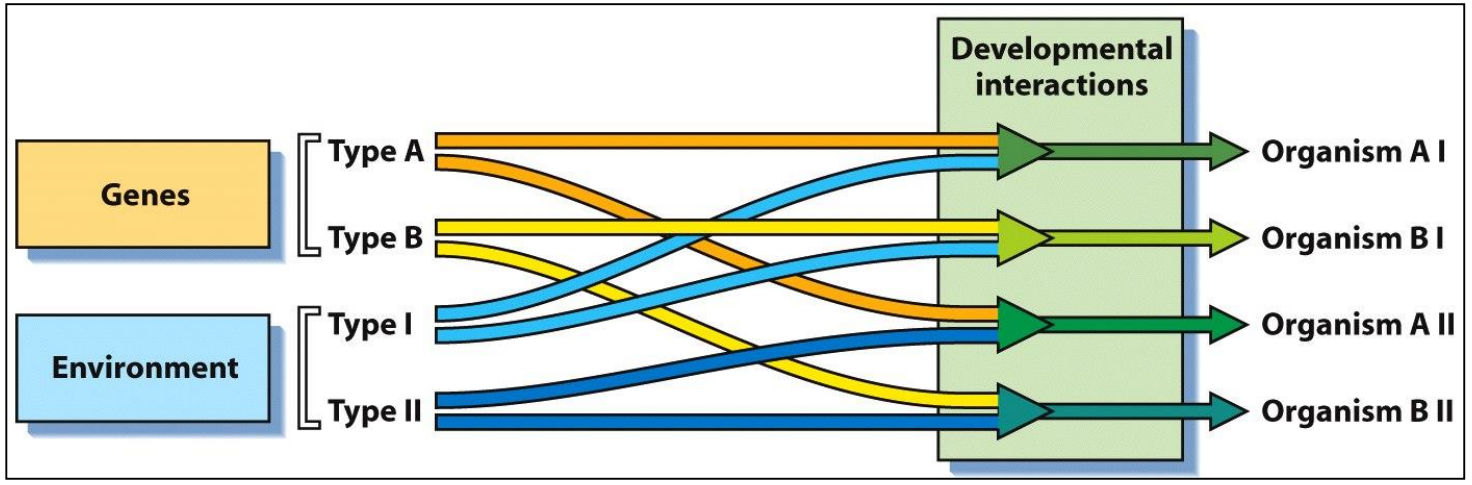
However, genes are not the only factor determining the characteristics of an organism....



Griffiths *et al*. (2012) *Introduction to Genetic Analysis*, p 35

An organism's environment also influences its characteristics

G X E (genes by environment)



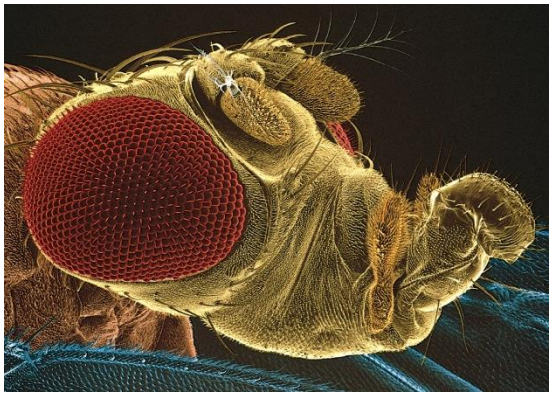
phenotype - an organism's morphology, physiology, behaviour, ecology; changes throughout lifetime of an organism

genotype - an organism's complete set of genes; essentially fixed throughout lifetime of an organism

NB we also use pheno- and genotype when we talk about a subset of characteristics or genes, respectively

Griffiths *et al*. (2008) *Introduction to Genetic Analysis*, p 23

An example of G x E: Drosophila eye size

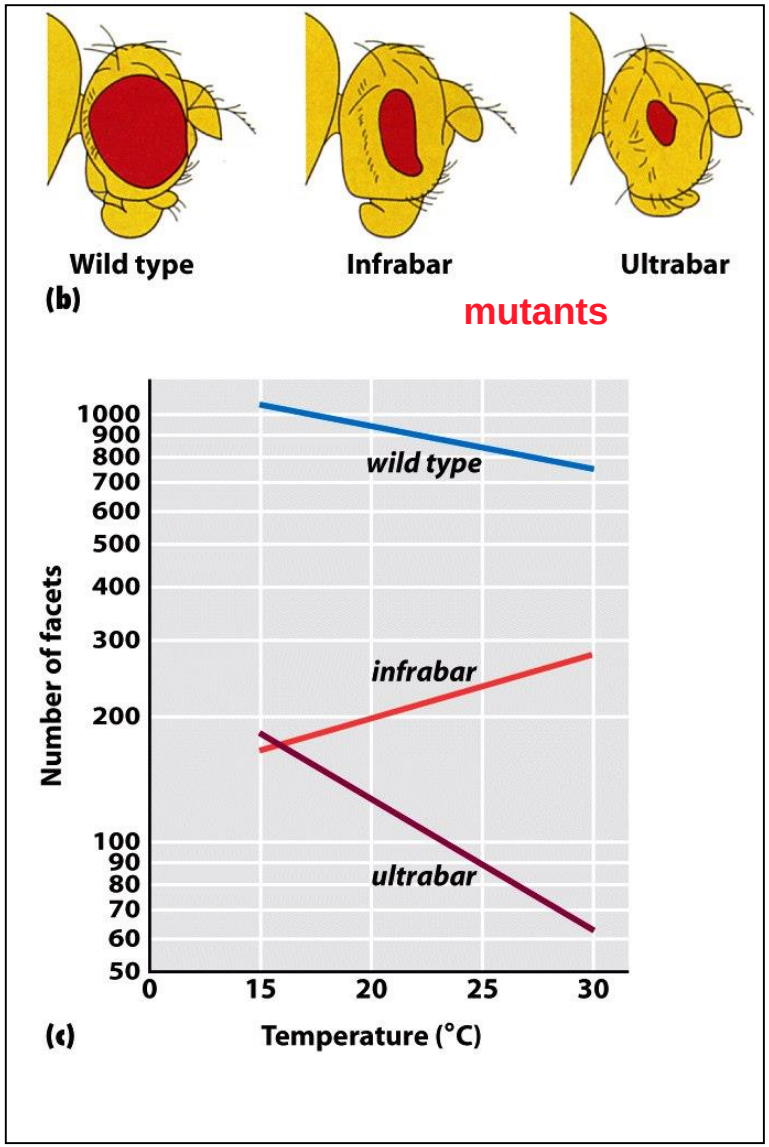


The number of facets (light receptors making up the compound eye) is influenced by genotype and temperature

wild type - the normal form of any characteristic of an organism (found in nature or a lab stock)

mutant - an altered form of a wild type characteristic

Practical 1A!

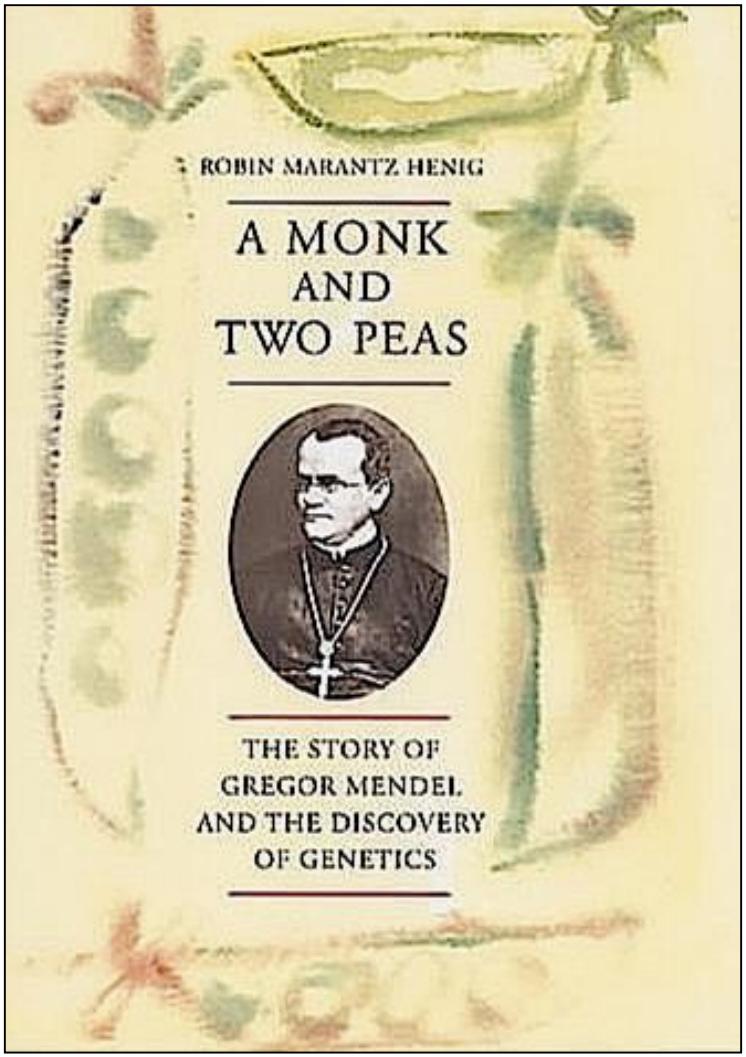


Griffiths *et al*. (2008) *Introduction to Genetic Analysis*, p 25

(Huge) Advances in genetics in less than a century

- identification of DNA as the hereditary material – Avery, MacLeod & McCarty, 1944
- structure of DNA determined – Watson & Crick (& Franklin!), 1953
- cloning of a DNA fragment – Cohen & Boyer, 1972
- genetic engineering – from 1970s
  - pharmaceuticals and non-medical products
  - insect- and herbicide-resistant crops
- whole genome sequences, including that of humans, facilitating:
  - "designer" disease treatment
  - directed animal and plant breeding
  - comparative genomics
    - conservation biology
    - botany
    - zoology
    - agricultural science – breeding, veterinary sciences
    - medical sciences

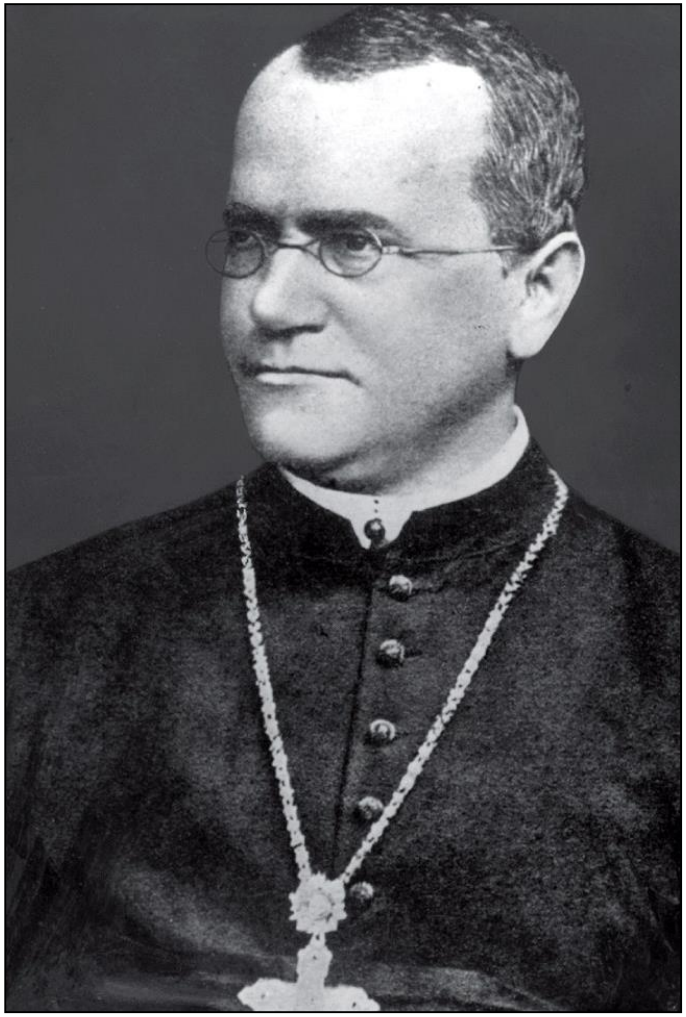
Single-Gene Inheritance



- first analysed by Gregor Mendel
  - Augustinian monk
  - garden peas
  - interested in inheritance of traits (properties, characteristics), not the properties of the peas
  - 10 years of crosses (controlled matings), counting the resulting progeny, and testing hypotheses in subsequent experiments
  - results published in 1866, but were not known or ignored until 1900
  - concluded: traits controlled by discrete units of inheritance (still knew nothing about genes or even chromosomes at that time!)
    - units existed in pairs, with the two members separating when gametes formed

8th March is International Mendel Day!

Single-Gene Inheritance



The thinking at the time of Mendel's experiments...

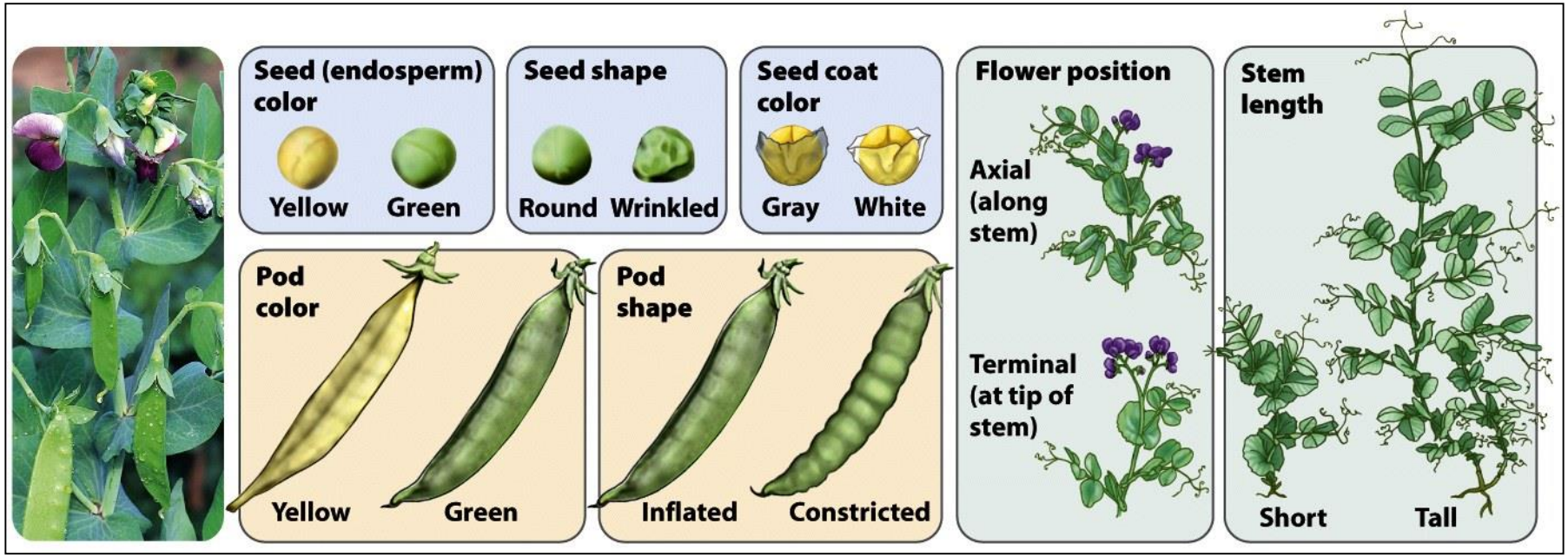
Darwin's On the Origin of Species had been published in 1859

Blending of inheritance - progeny have traits midway between those of mother and father

Mendel showed - traits were inherited randomly and independently as discrete units (no blending)

Pierce (2012) *Genetics: A Conceptual Approach*, p 9

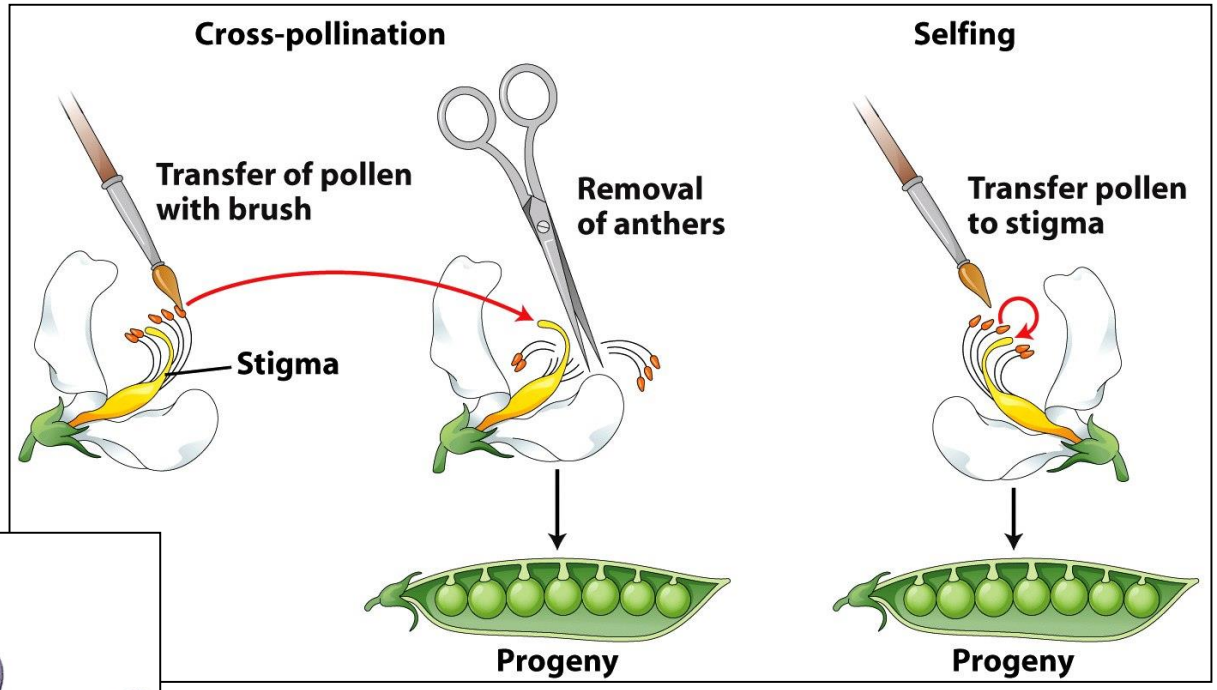
Mendel's Experiments

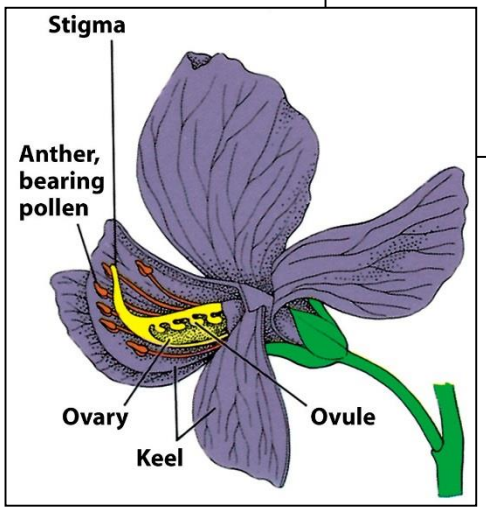


Mendel examined - 7 traits of peas
- contrasting phenotypes (easy to count and categorise)

Pierce (2012) *Genetics: A Conceptual Approach*, p 45

Mendel's Experiments





Mendel did - cross pollination crosses
- self crosses

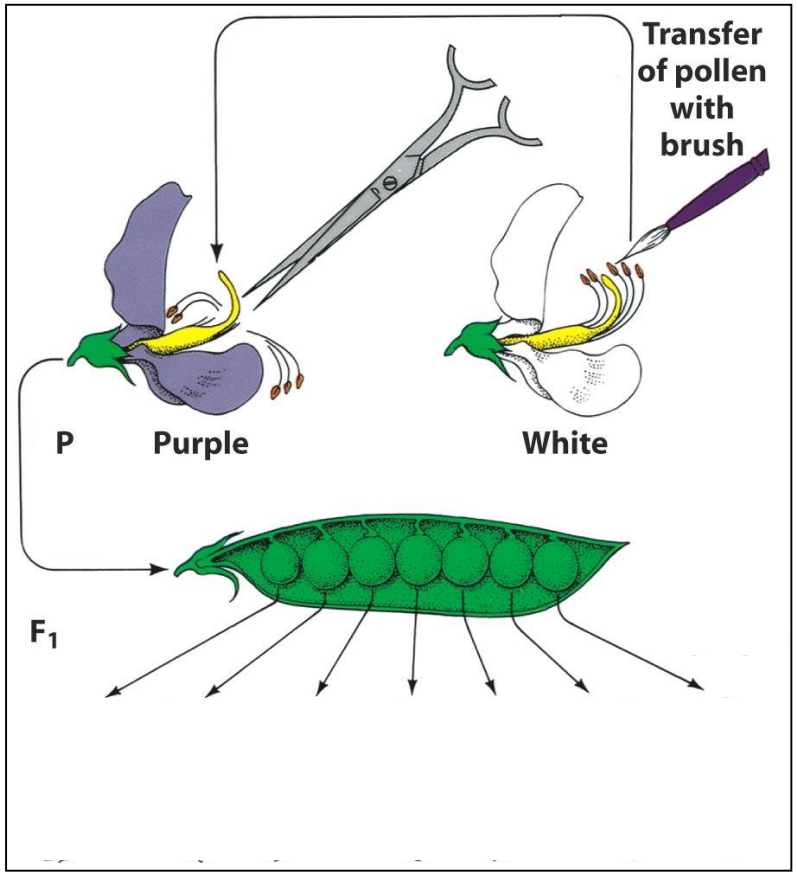
pollen - contains male gamete
- made in the anthers (male part of a flower)

stigma - receptive surface for pollen grain on female part of a flower

Griffiths *et al*. (2012) *Introduction to Genetic Analysis*, p 31

Mendel's Experiments

Mendel used - pure lines for particular traits (all progeny resulting from crosses within a line are identical for a particular trait, e.g. purple flowers)
- parental (P) generation



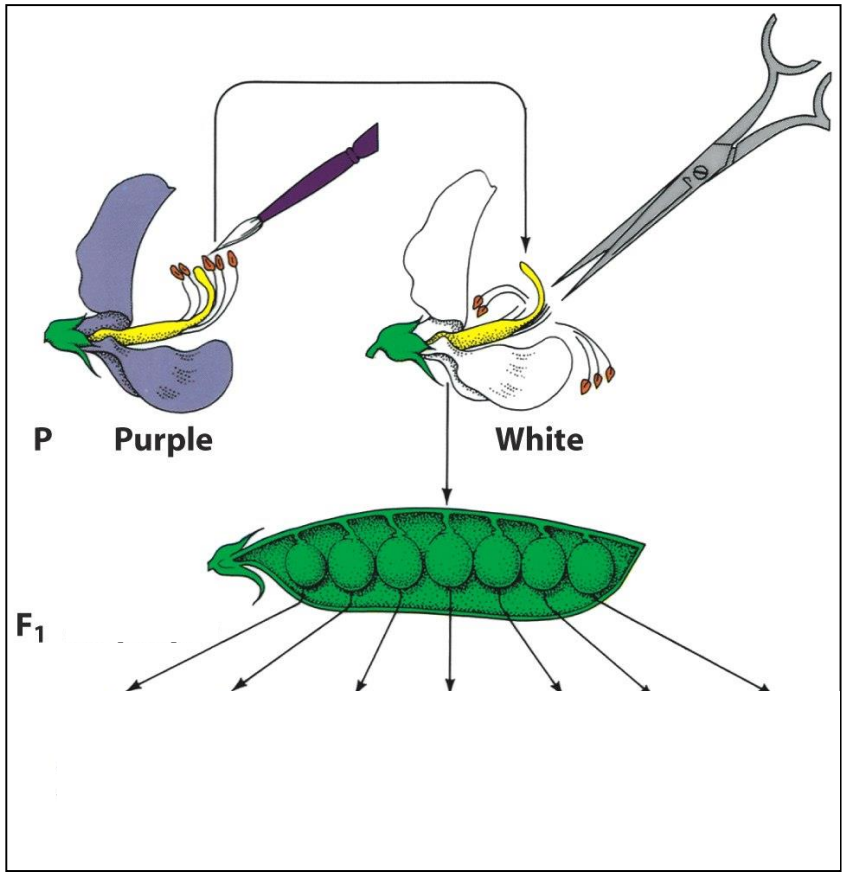
Progeny resulting from crosses of P generation are called first filial (F_1) generation

In an early experiment:

pollinated purple-flowered plant with pollen from white-flowered plant

Griffiths *et al*. (05) *Introduction to Genetic Analysis*, p 31

Mendel's Experiments



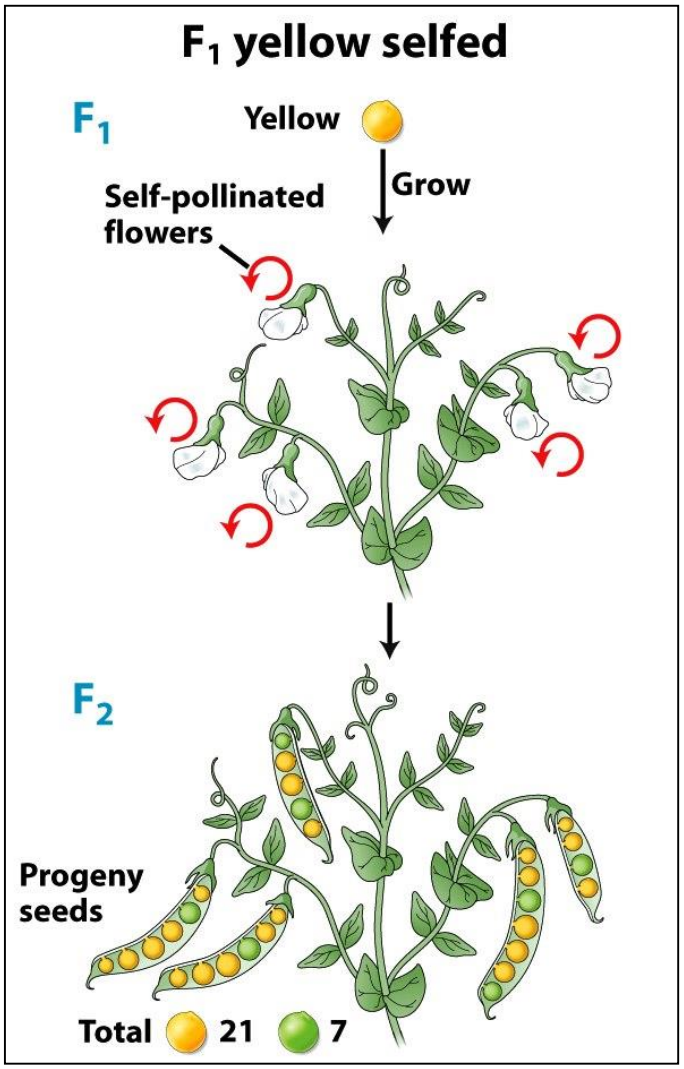
Did the reciprocal cross (pollen from purple-flowered plant used to pollinate white-flowered plant):

For both crosses - saw no blending of inheritance

Hypothesis that "discrete units of inheritance existed" was supported

Griffiths *et al*. (05) *Introduction to Genetic Analysis*, p 31

Mendel's Experiments



Experiments with the F_1 generation:

Grew F_1 yellow seeds (resulting from a cross between a yellow-seeded plant and a green-seeded plant) into plants and self crossed (monohybrid cross)

Resulting progeny represent the second filial (F_2) generation

Found 3/4 of the F_2 generation seed were yellow, 1/4 were green $\rightarrow$ 3:1 phenotypic ratio

Griffiths *et al*. (2012) *Introduction to Genetic Analysis*, p 31

Mendel's Experiments

Table 2-1 Results of All Mendel's Crosses in Which Parents Differed in One Character

Parental phenotype	F_1	F_2	F_2 ratio
1. round × wrinkled seeds	All round	5474 round; 1850 wrinkled	2.96 : 1
2. yellow × green seeds	All yellow	6022 yellow; 2001 green	3.01 : 1
3. purple × white petals	All purple	705 purple; 224 white	3.15 : 1
4. inflated × pinched pods	All inflated	882 inflated; 299 pinched	2.95 : 1
5. green × yellow pods	All green	428 green; 152 yellow	2.82 : 1
6. axial × terminal flowers	All axial	651 axial; 207 terminal	3.14 : 1
7. long × short stems	All long	787 long; 277 short	2.84 : 1

3:1 phenotypic ratio held in the F_2 generation for each trait Mendel studied

For each cross

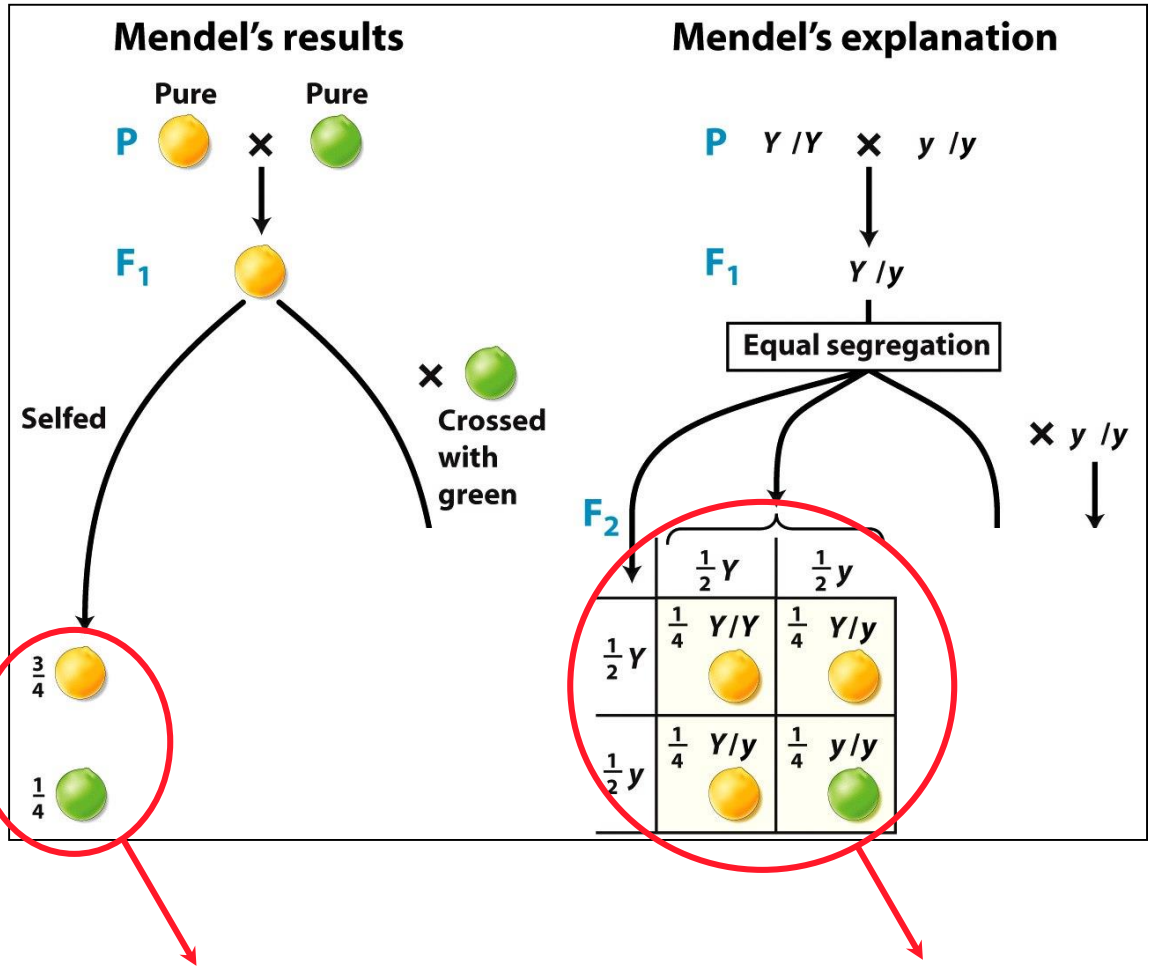
-one parental trait disappeared in the F_1 generation, but reappeared in the F_2 generation

- •unit of inheritance present in F_1 but not expressed
- •recessive allele (form) of the gene responsible for the trait (recessive phenotype)

- parental trait expressed in the F_1 represents the dominate phenotype (dominate allele of the gene)

Griffiths *et al*. (08) *Introduction to Genetic Analysis*, p 40

Mendel's Experiments - The Explanation



At meiosis, the two alleles of a gene separate (segregate) equally into the male and female gametes.

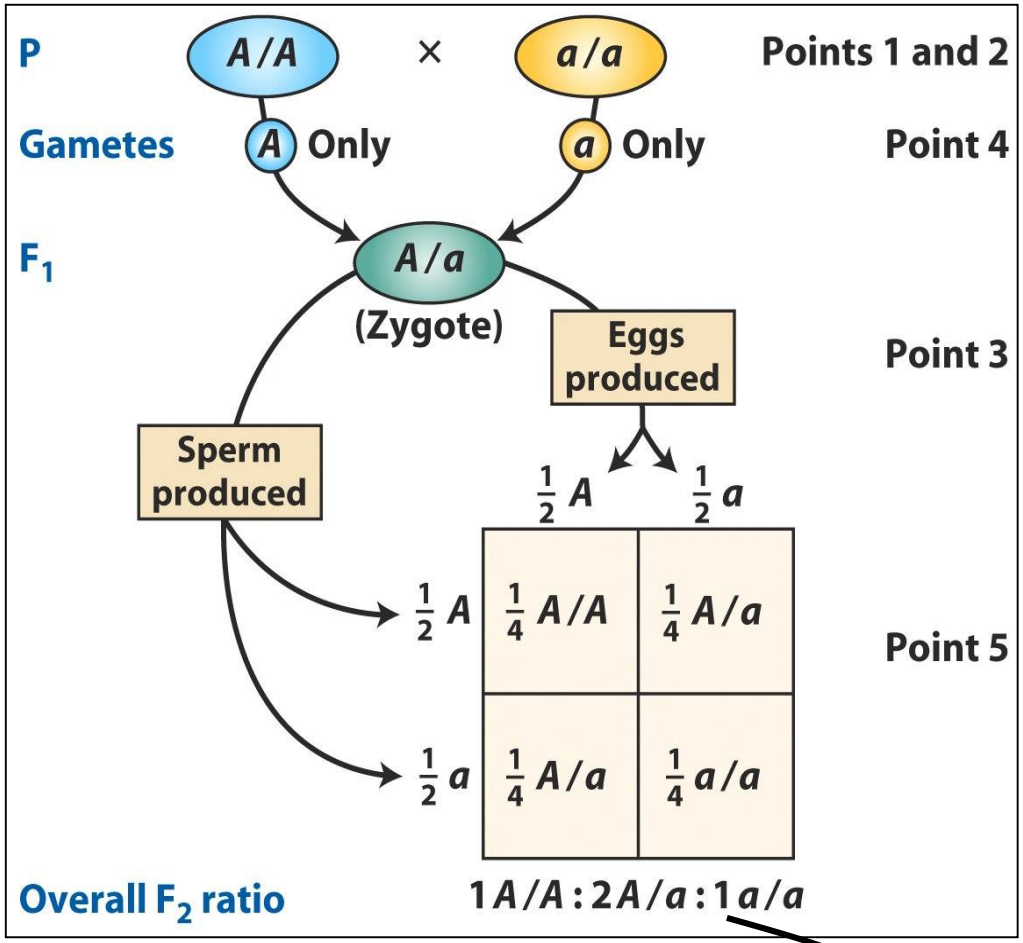
At fertilisation, the gametes combine at random.

Phenotypic ratio of 3:1
(3 yellow : 1 green)

Genotypic ratio of 1:2:1
(1Y/Y : 2Y/y : 1y/y)

Griffiths *et al*. (2012) *Introduction to Genetic Analysis*, p 33

Mendel's Experiments - The Explanation



1. Discrete units of inheritance (genes), no blending

2. Each plant has two units (alleles)

3. Each gamete carries only one allele

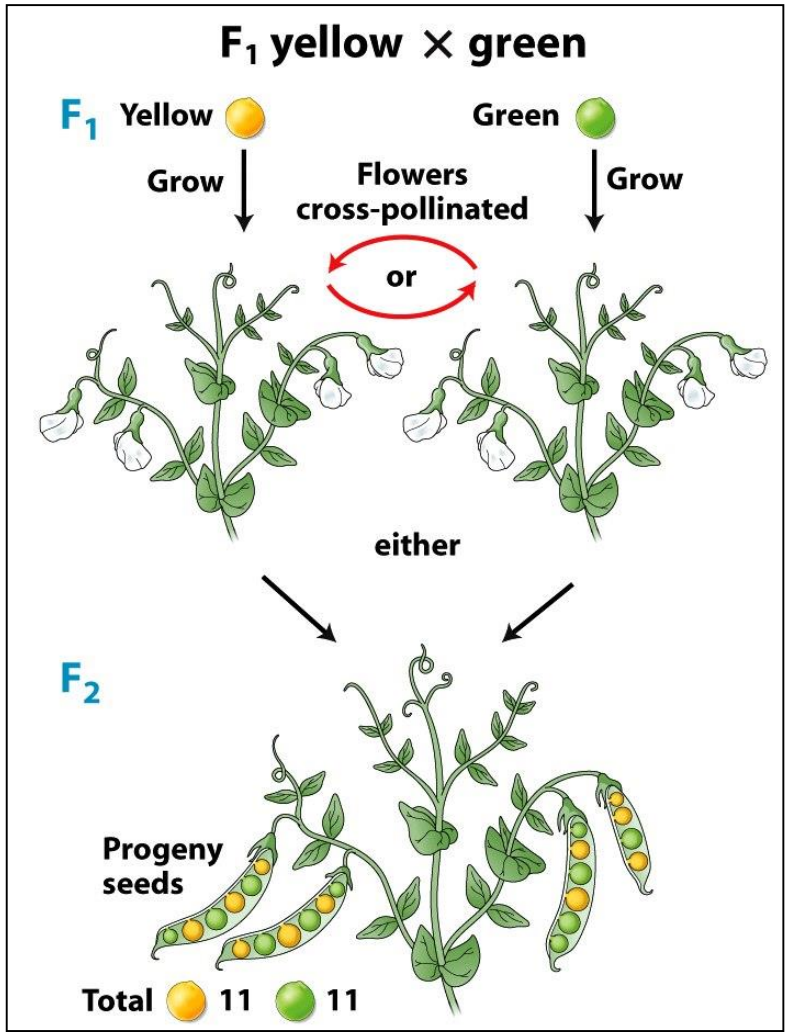
4. Each unit segregates equally and randomly into the gametes

5. Gametes combine randomly to form a zygote

A/A and a/a - homozygotes (pure lines)
A/a - heterozygotes (also called hybrids)

Griffiths *et al*. (05) *Introduction to Genetic Analysis*, p 33

Mendel's Experiments - The Test



Crossed F_1 yellow-seeded plant with a pure line green-seeded plant

- homozygous recessive
- also called a tester

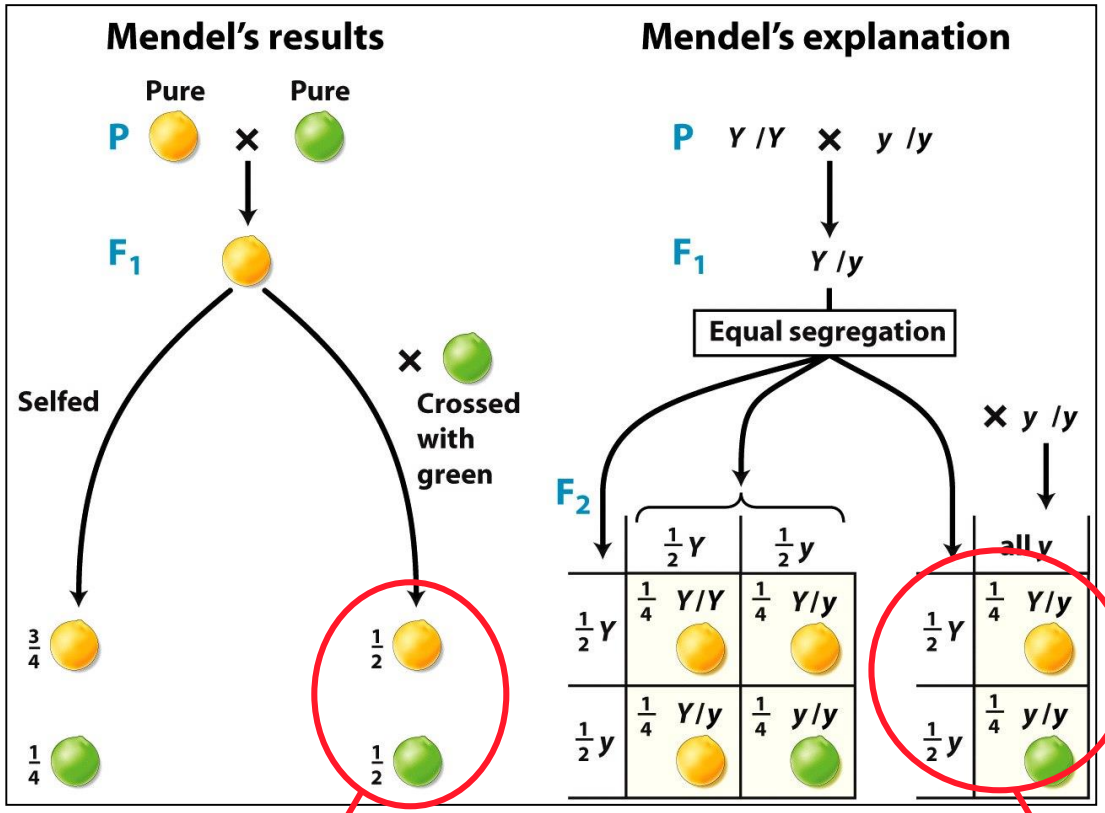
F_2 generation were 1/2 yellow seed, 1/2 green seed $\rightarrow$ 1:1 ratio

Found same ratio with the reciprocal cross

TESTCROSS

Griffiths *et al*. (2012) *Introduction to Genetic Analysis*, p 31

Mendel's Experiments - The Test



TESTCROSS

Tester contributes only recessive alleles - allows gametes produced by individual of unknown genotype to be determined from the progeny resulting from the cross

Phenotypic ratio of 1:1
(1 yellow : 1 green)

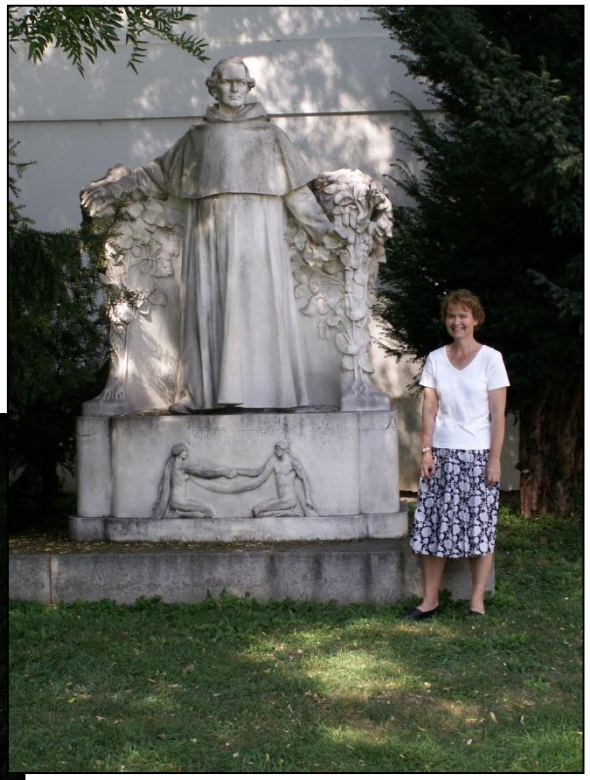
Genotypic ratio of 1:1
(1Y/y : 1y/y)

Griffiths *et al*. (2012) *Introduction to Genetic Analysis*, p 33

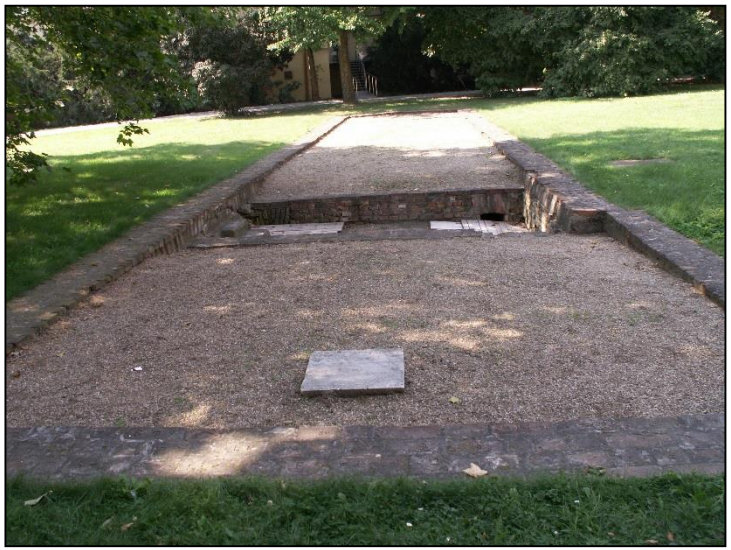
Mendel's First Law - The Law of Equal Segregation

"The two members of a gene pair segregate from each other into the gametes; so half the gametes carry one member of the gene pair and the other half of the gametes carry the other member of the gene pair."

The Augustinian Monastery at Brno 2008



The Augustinian Monastery at Brno 2008



Trivial quiz:

What human phenotype/traits (easily observable) follow Mendelian inheritance?