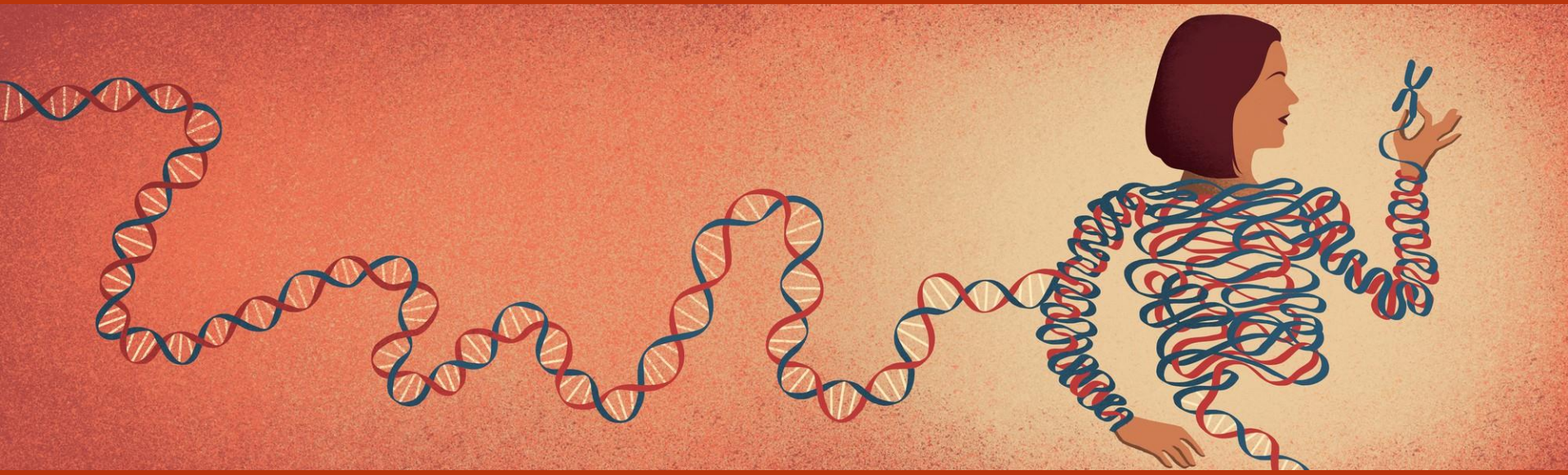
GENETICS
LECTURE 6



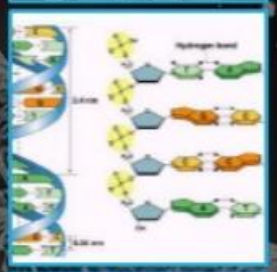
Katarzyna Osmańska-Załuska,PhD

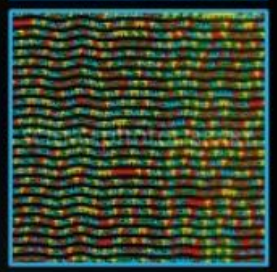
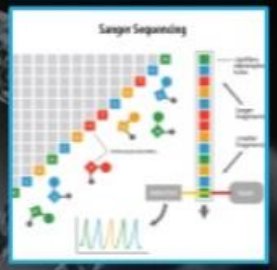
Groundbreaking Discoveries

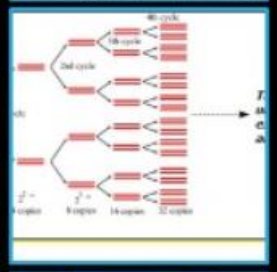
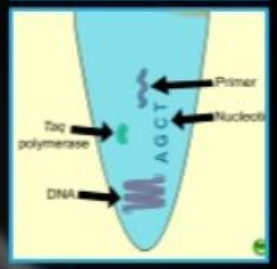






1865 — G. Mendel — Mendel's Laws

1915 — T.H. Morgan — chromosome theory of heredity

1953 — J. Watson, F. Crick, M. Wilkins, R. Franklin — Structure of DNA

1973/ 1977 — F. Sanger, A. Maxam, W.Gilbert — DNA sequencing

1983 — K. Mullis — PCR method

Source: lecture of Agata Daszkowska-Golec, University of Silesia

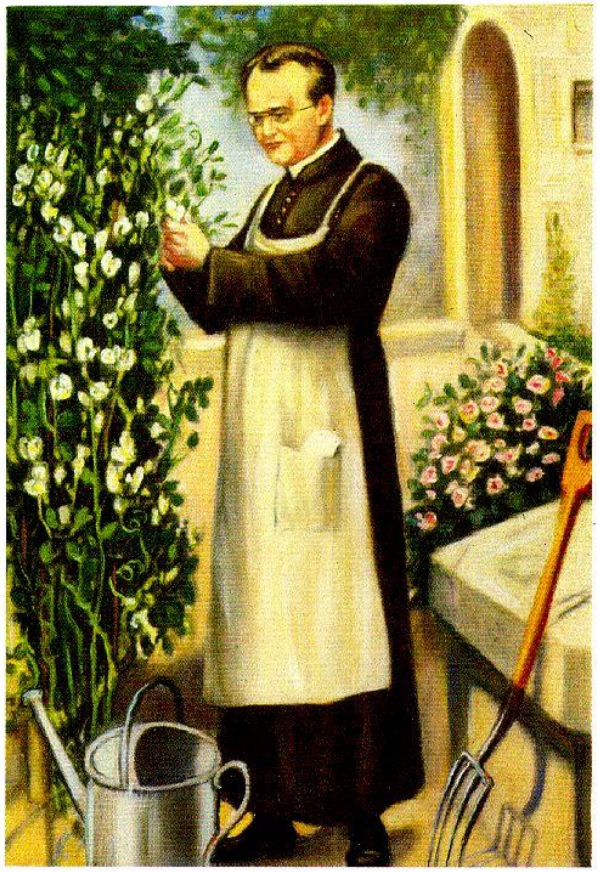
History

- 1803 r. J.C. Otto, 1813. J. Hay, 1815. Buels - description of transmission of haemophilia from mother to son in various unrelated families
- 1814 r. J. Adams - presentation of autosomal recessive inheritance in a family of relatives
- 1850 Maupertuis - description of the inheritance of polydactyly

Gregor Mendel – Father of Genetics

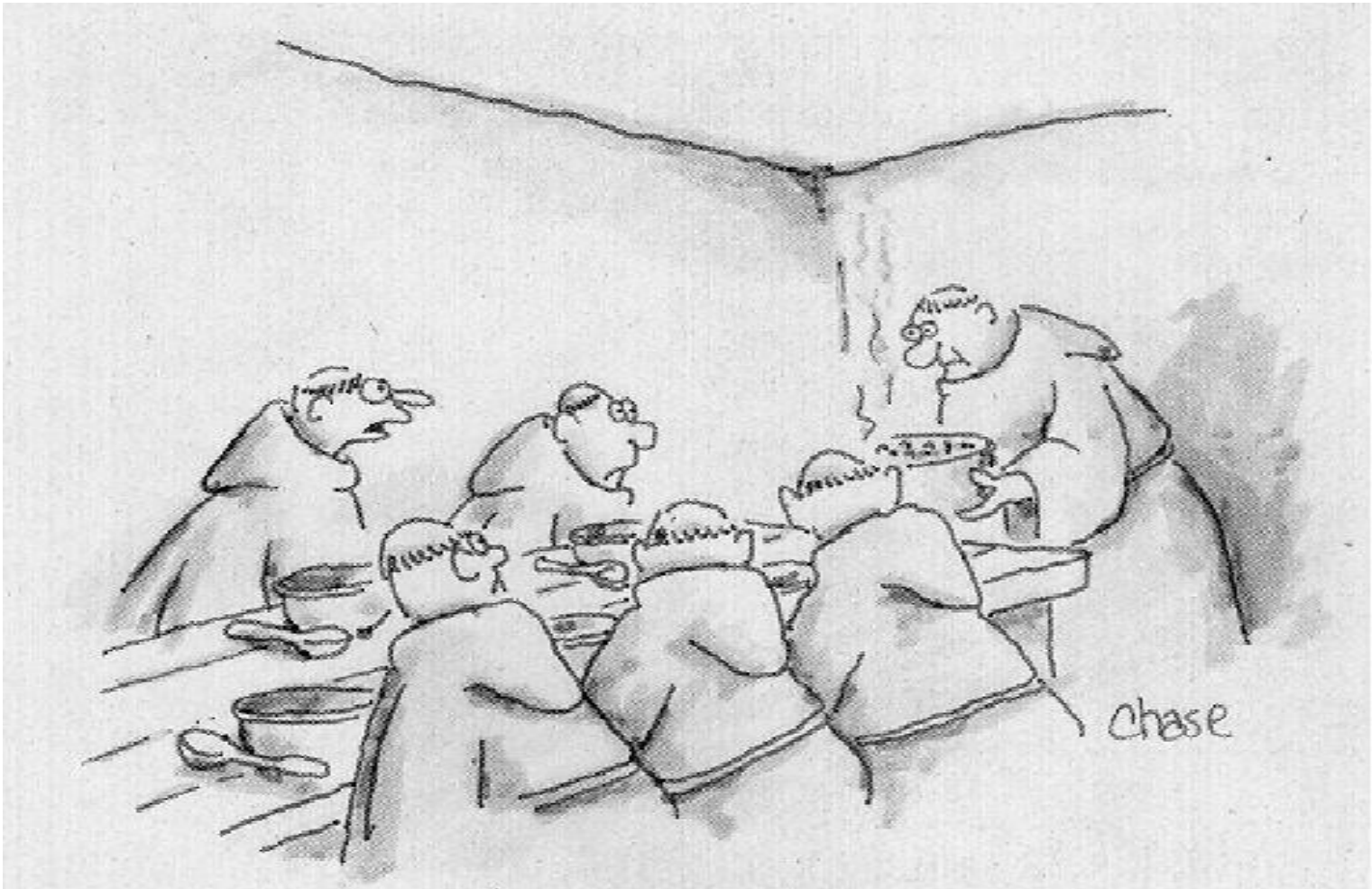
Developer of the theoretical foundations of modern genetics

Mendel's Laws



Gregor Mendel

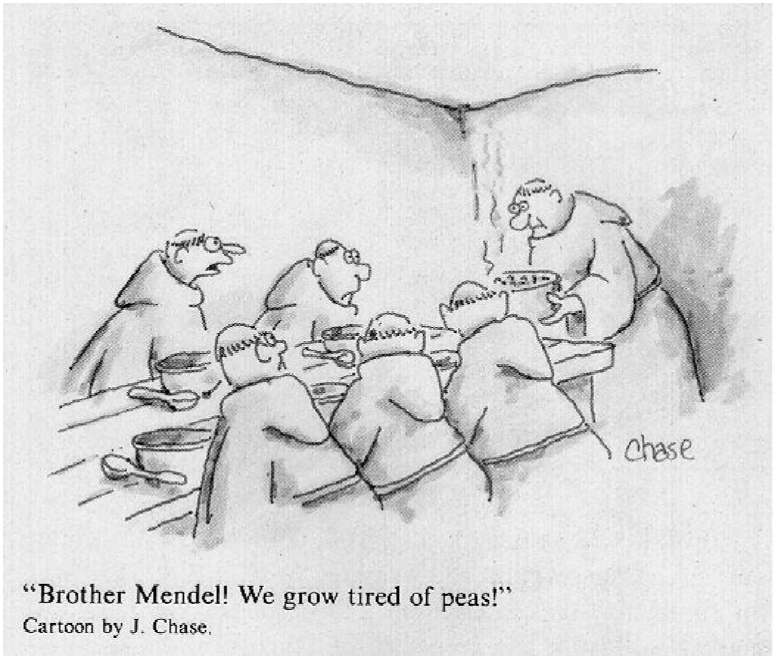
- **Mendel's laws** - rules concerning the way in which traits are passed on to offspring, discovered by the Czech monk, G.J. Mendel, on the basis of studies on the hybridisation of pea plants (Pisum sativum)
- These laws were promulgated by Mendel in 1865 in his work: Studies on Plant Hybrids....



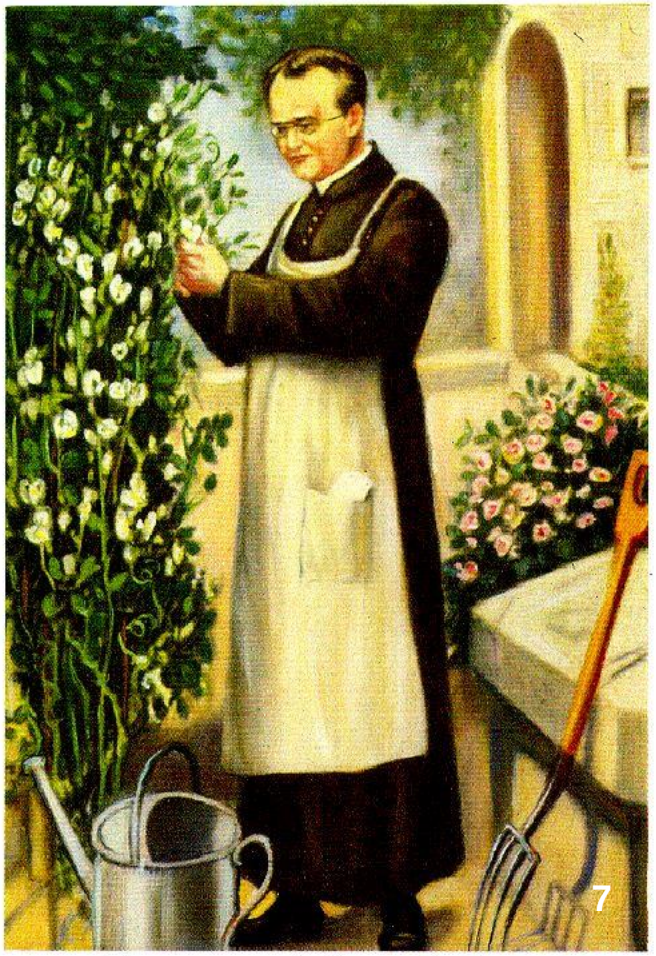
"Brother Mendel! We grow tired of peas!"
Cartoon by J. Chase.

Mendel deduced the underlying principles of genetics from these patterns.

- Segregation
- Dominance
- Independent assortment

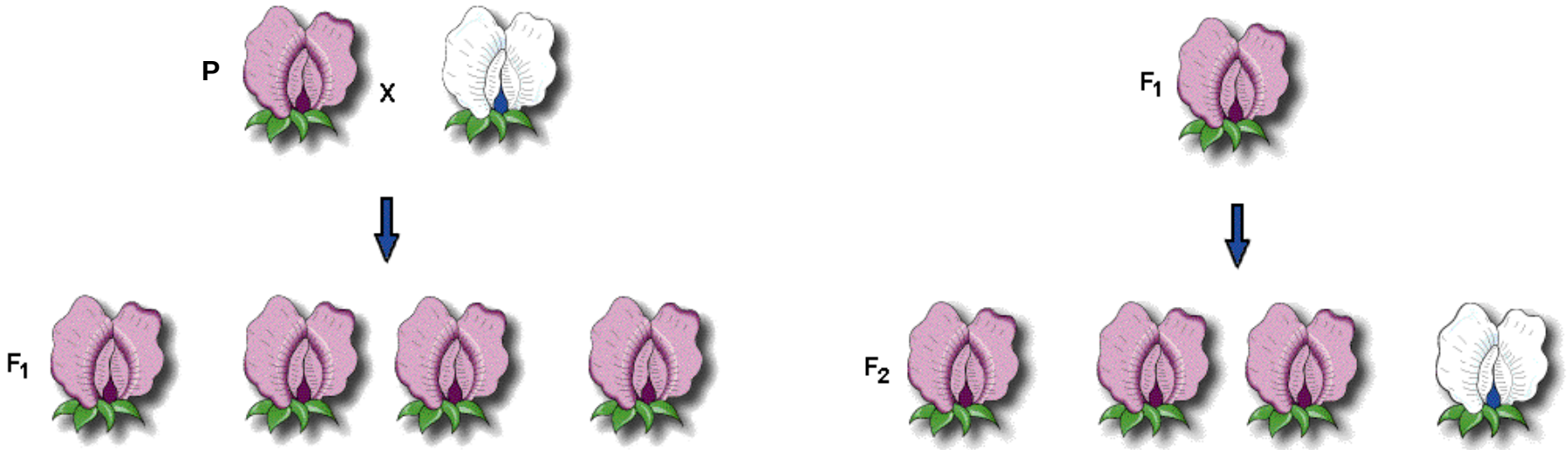


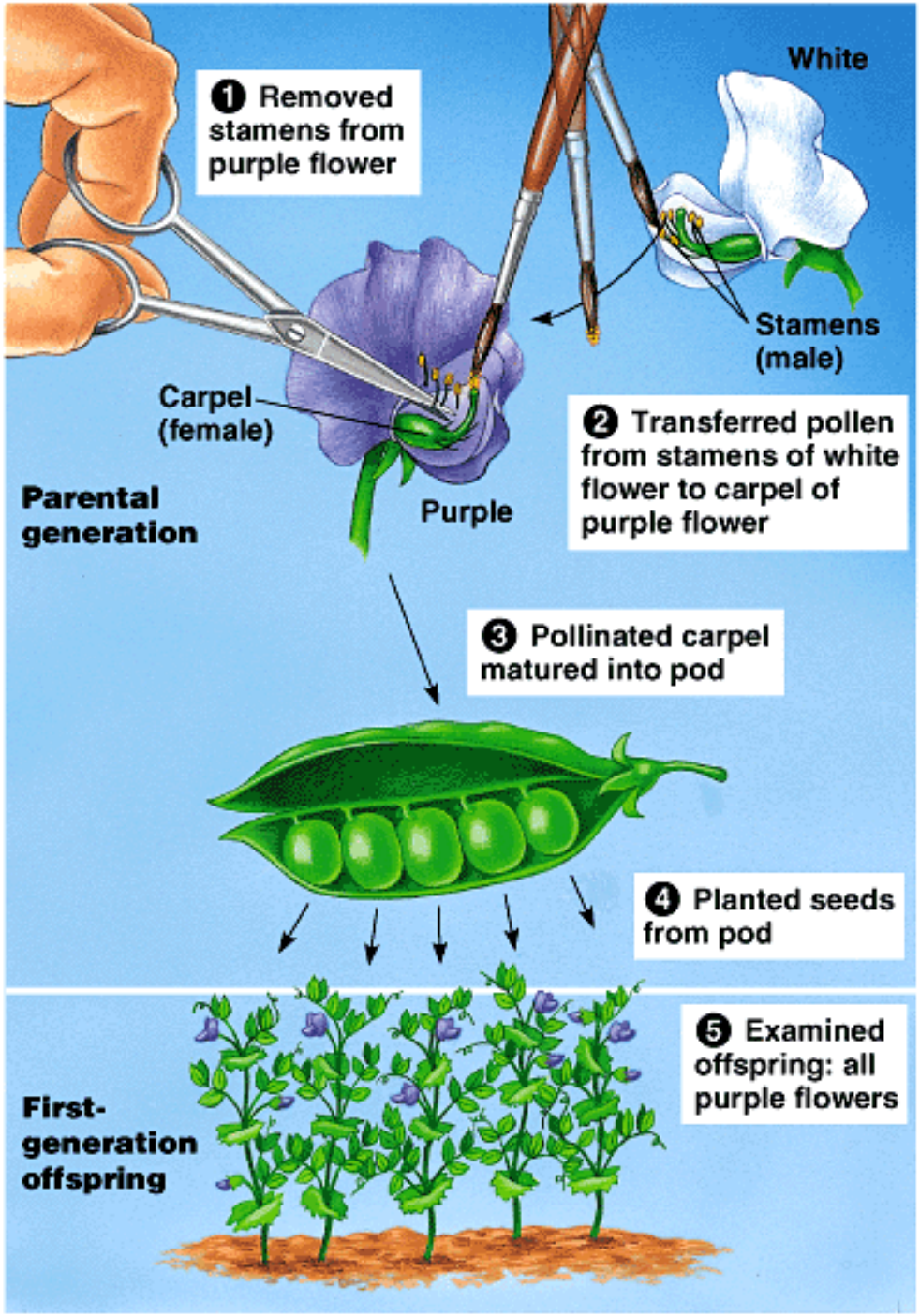
"Brother Mendel! We grow tired of peas!"
Cartoon by J. Chase.

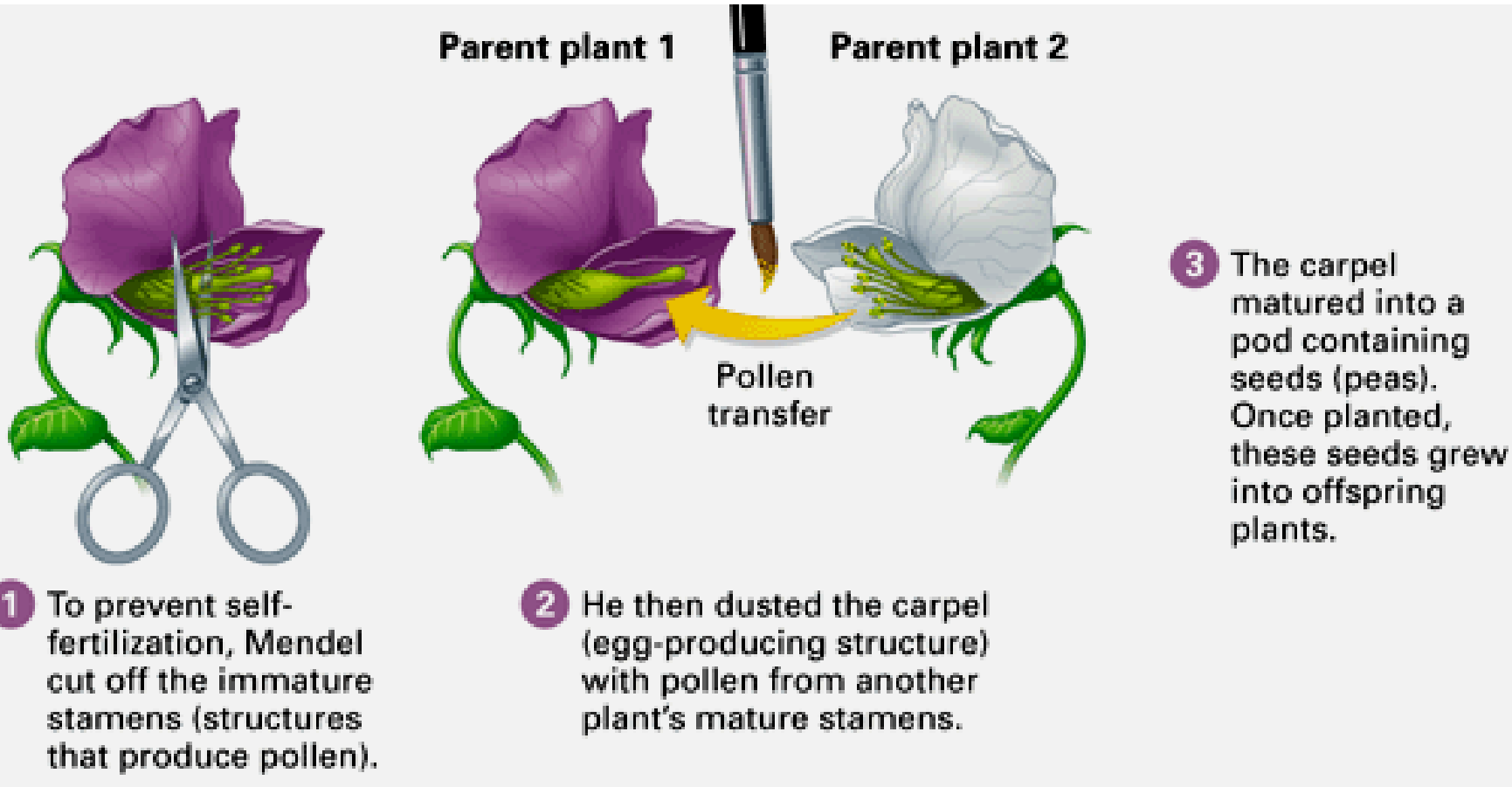


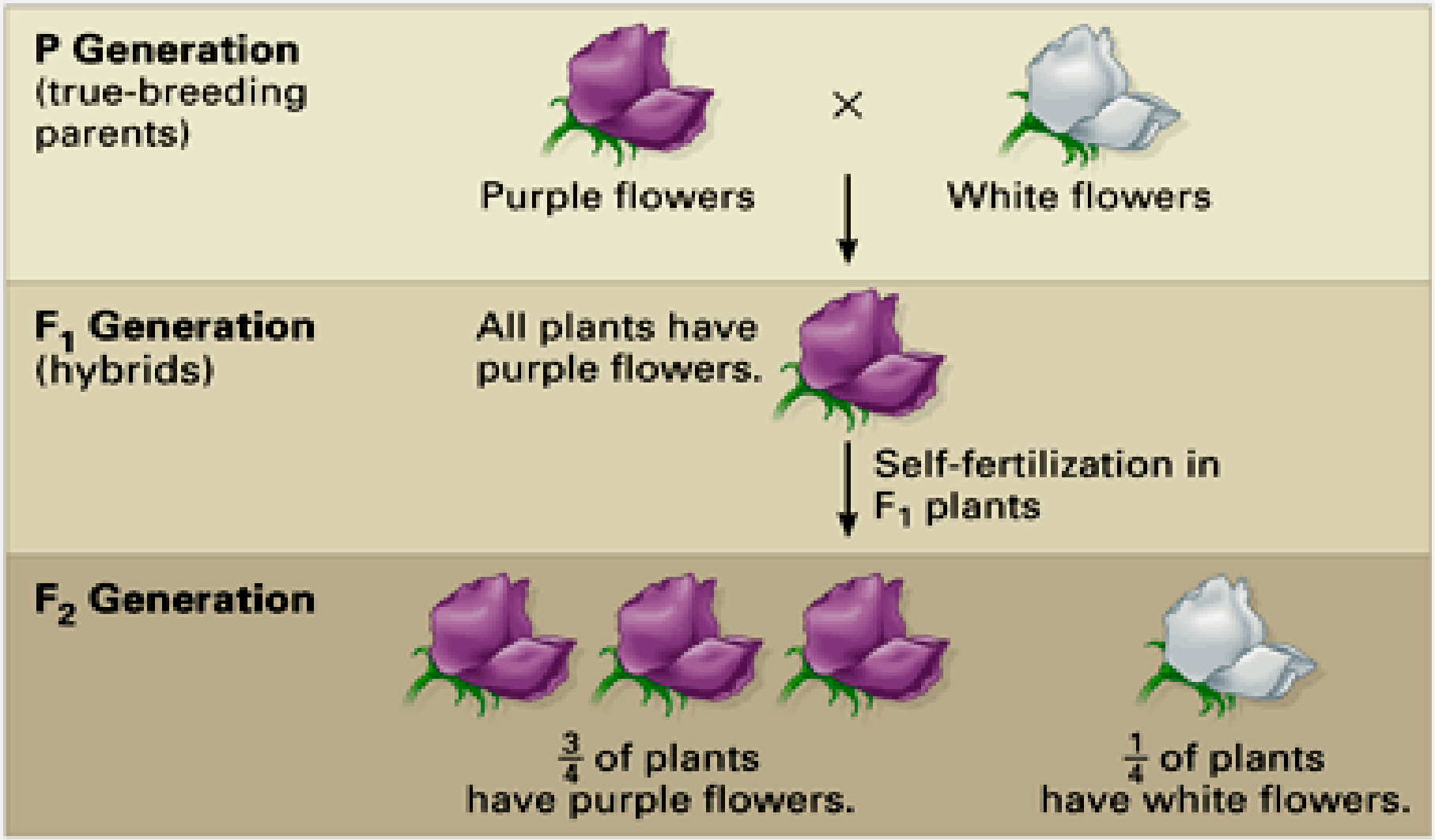
Gregor Mendel

- P – parental generation
- F_1 (first filial) generation – refers to offspring produced by parent generation
- F_2 (second filial) generation refers to offspring from F_1

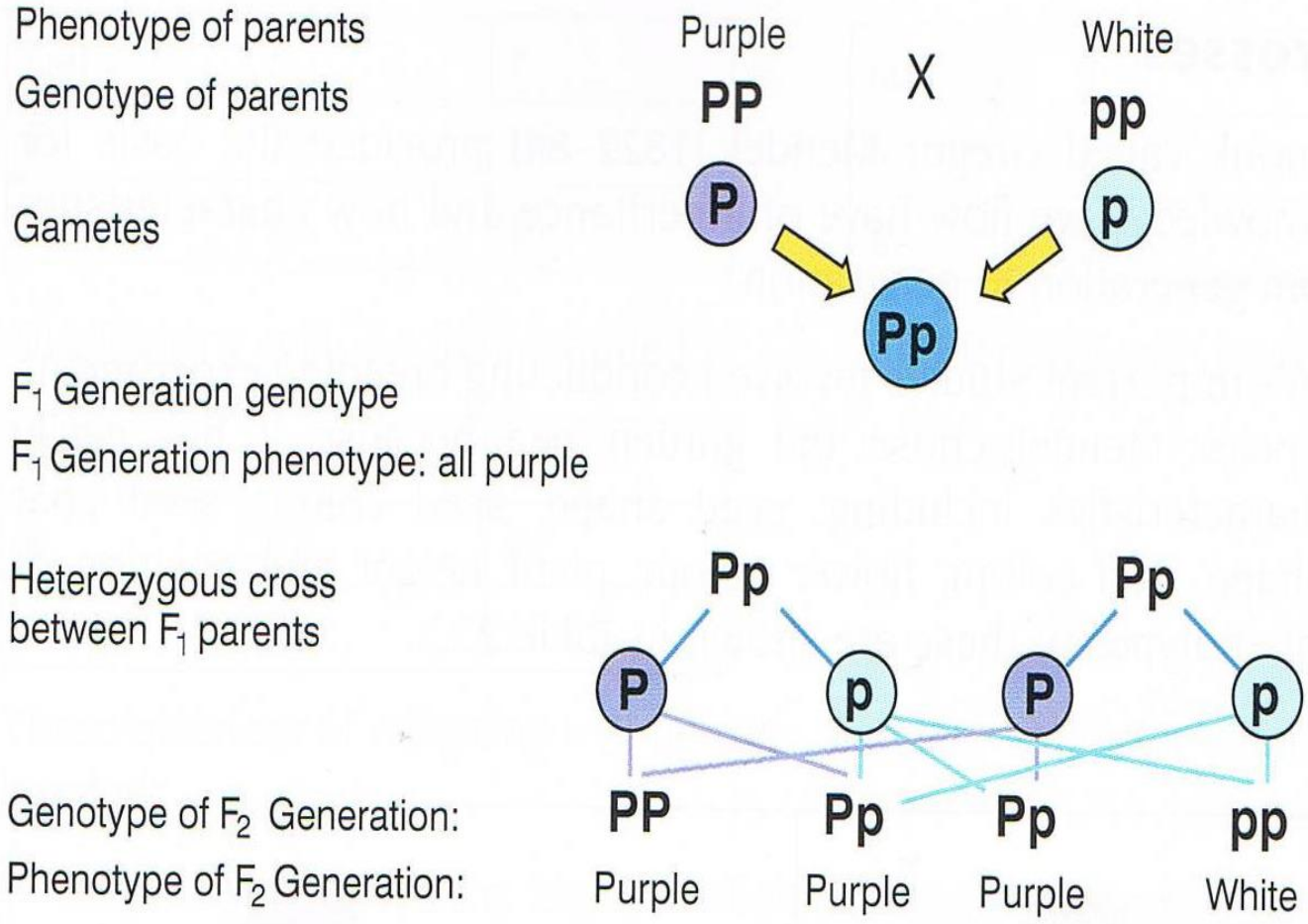


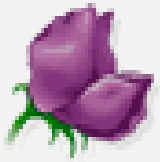
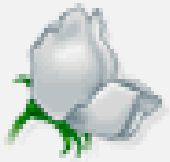
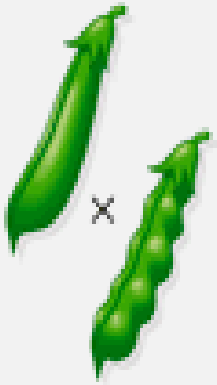
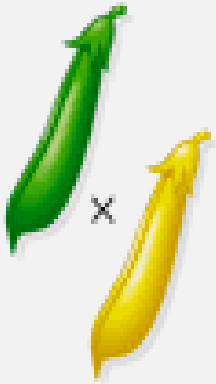
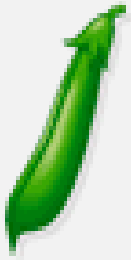
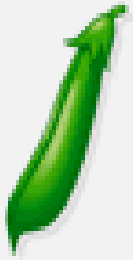

1 Removed stamens from purple flower
White
Stamens (male)
Carpel (female)
2 Transferred pollen from stamens of white flower to carpel of purple flower
Parental generation
Purple
3 Pollinated carpel matured into pod
4 Planted seeds from pod
5 Examined offspring: all purple flowers
First-generation offspring


Parent plant 1
Parent plant 2
Pollen transfer
1 To prevent self-fertilization, Mendel cut off the immature stamens (structures that produce pollen).
2 He then dusted the carpel (egg-producing structure) with pollen from another plant's mature stamens.
3 The carpel matured into a pod containing seeds (peas). Once planted, these seeds grew into offspring plants.


P Generation
(true-breeding parents)
Purple flowers
×
White flowers
F1 Generation
(hybrids)
All plants have purple flowers.
Self-fertilization in F1 plants
F2 Generation
3/4 of plants have purple flowers.
1/4 of plants have white flowers.

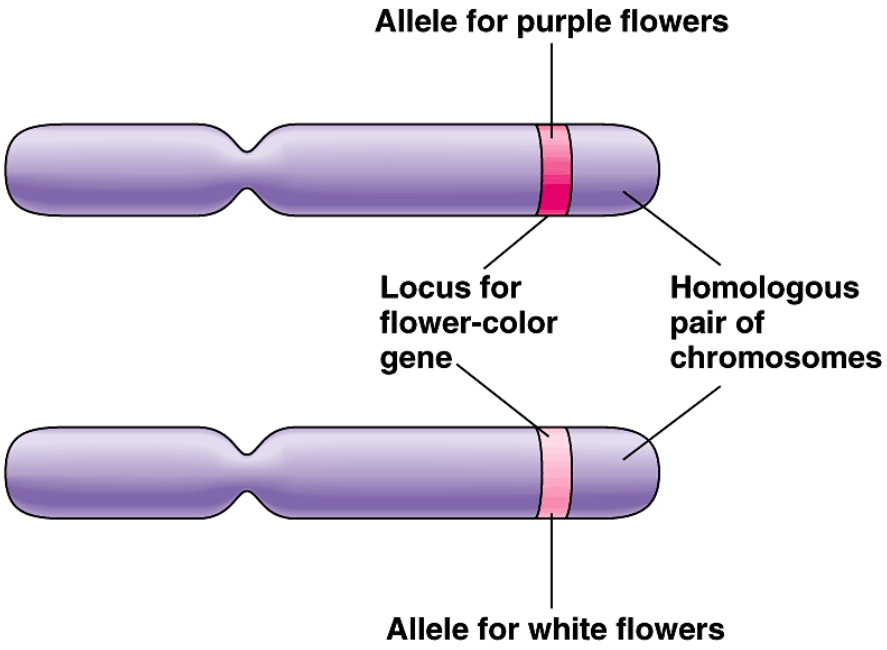
MENDEL'S GENETIC DIAGRAM FOR F1 & F2 GENERATIONS



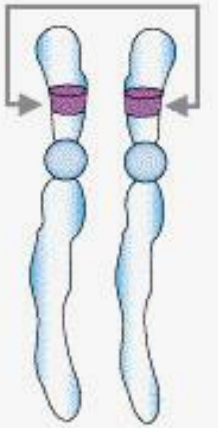
	Flower color	Flower position	Seed color	Seed shape	Pod shape	Pod color	Stem length
P	Purple  ×  White	Axial  ×  Terminal	Yellow  ×  Green	Round  ×  Wrinkled	Inflated  Constricted	Green  Yellow	Tall  Dwarf
F_1	Purple 	Axial 	Yellow 	Round 	Inflated 	Green 	Tall 

Alleles: alternative versions of a gene.

The gene for a particular inherited character resides at a specific locus (position) on homologous chromosome.

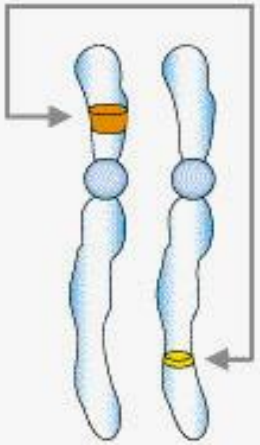


For each character, an organism inherits two alleles, one from each parent



Alleles

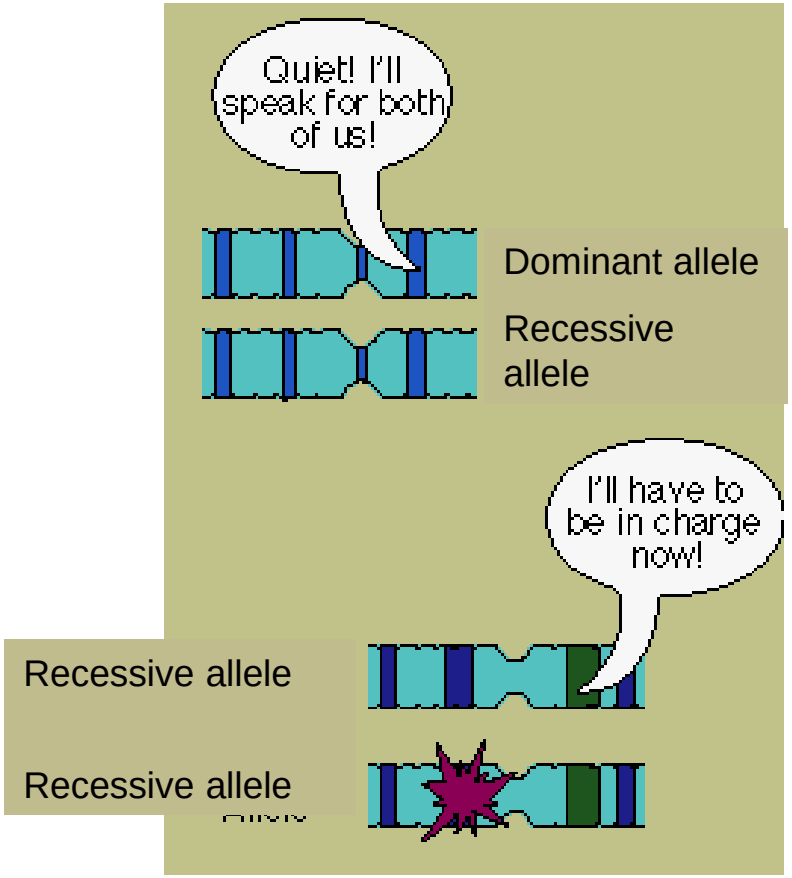
(code for same trait, same location on chromosome)



Genes, but *not* alleles

(code for different trait, different locations on chromosome)

How do alleles differ?



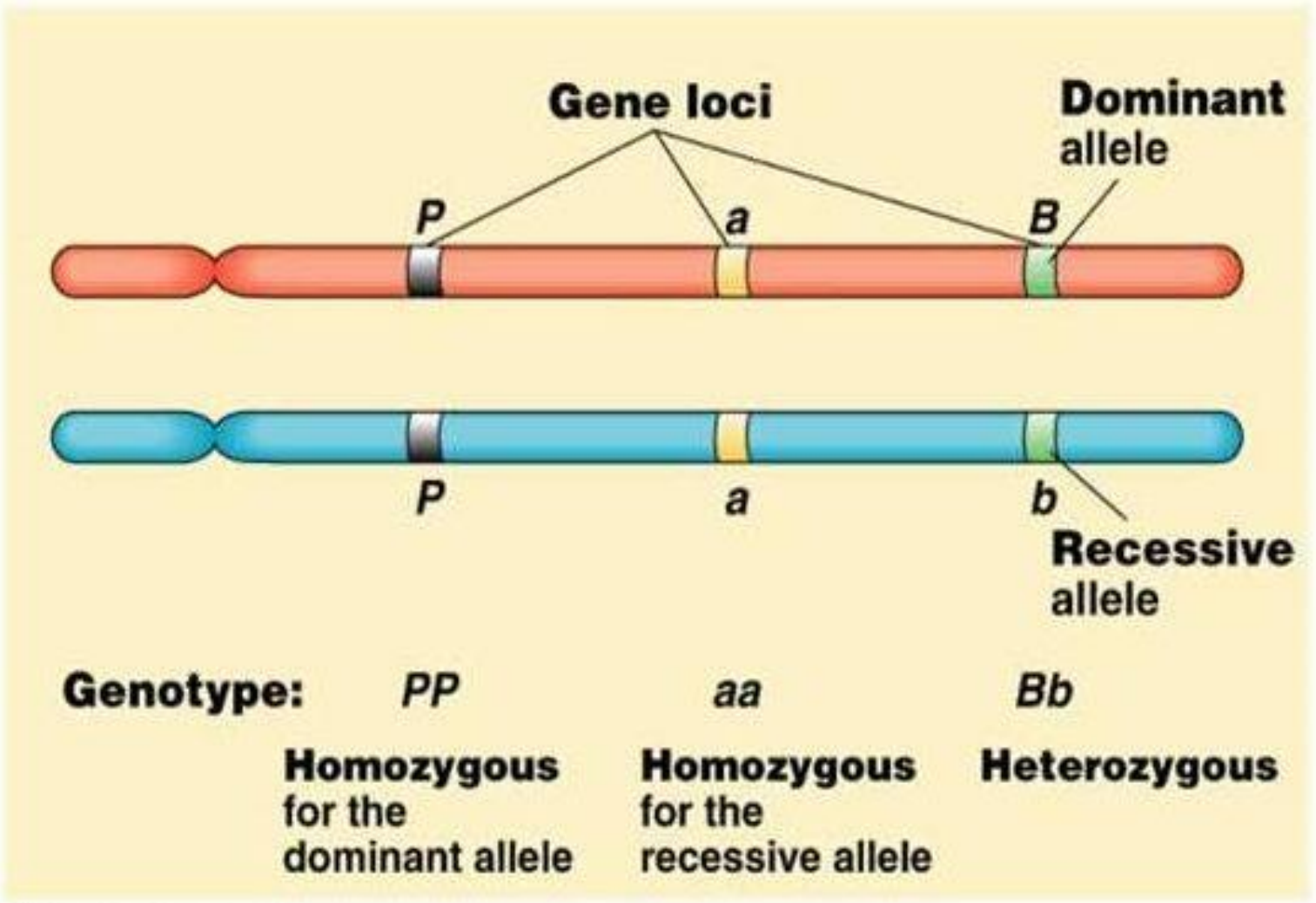
Dominant - a term applied to the trait (allele) that is expressed irregardless of the second allele.

Recessive - a term applied to a trait that is only expressed when the second allele is the same (e.g. short plants are homozygous for the recessive allele).

Dominant and recessive alleles

- Dominant allele
  - a form of the gene that manifests itself in a heterozygote phenotype
  - causes the F1 generation to be completely similar to one of the parents
  - denoted by a capital letter
- Recessive allele
  - a form of the gene not manifesting its phenotypic effect in the F1 generation in a heterozygous system
  - in the F2 generation manifesting at a frequency three times lower than in the offspring with the dominant allele
  - denoted by a small letter

Dominant and recessive alleles



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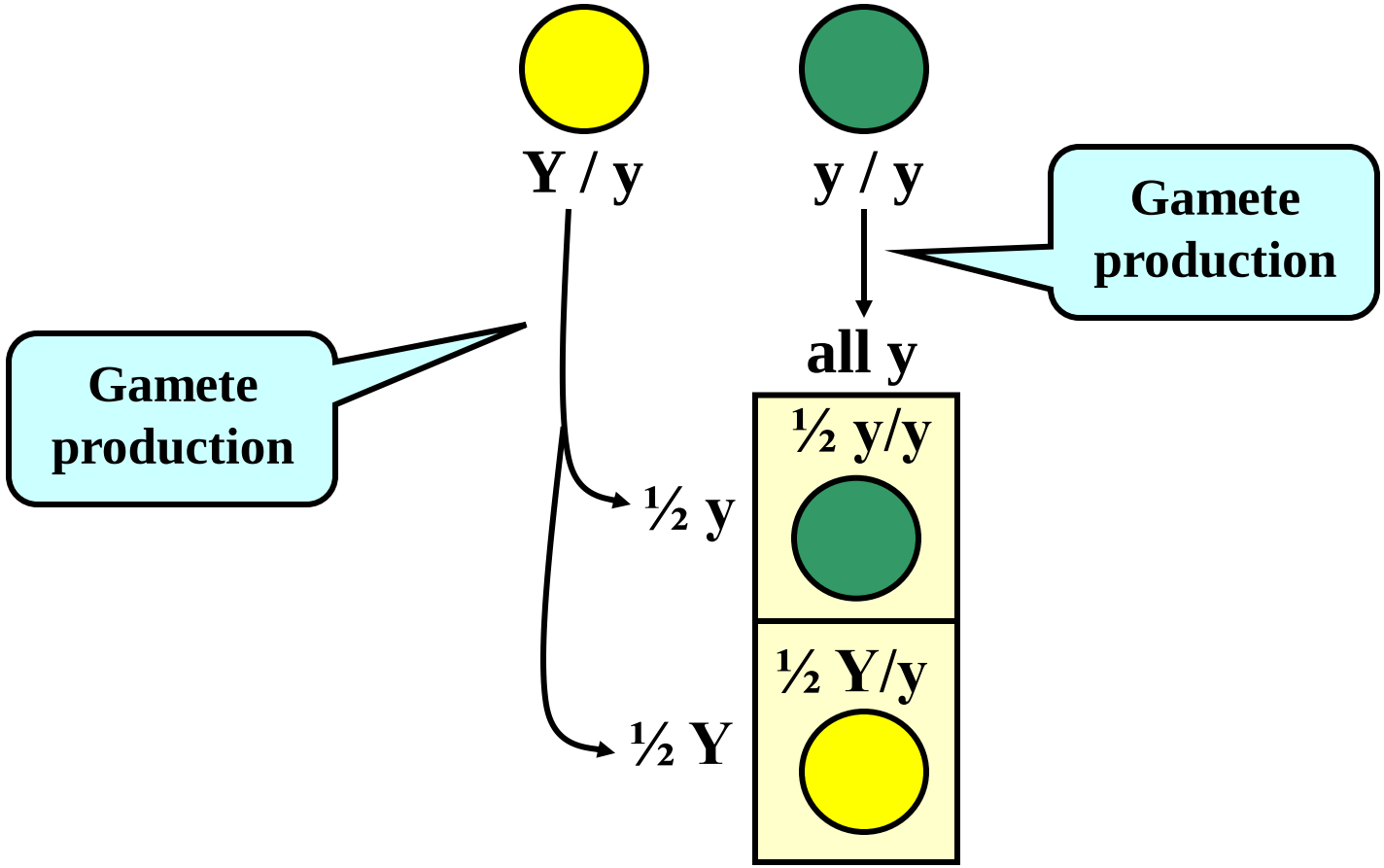
- If both alleles are the same (AA, aa), the organism is HOMOZYGOUS
- If both alleles are different (Aa), the organism is HETEROZYGOUS

MENDEL'S 1ST LAW

Two members of a gene pair segregate from each other into the gametes, so half the gametes carry one member of the pair and the other half carry the other member of the pair.

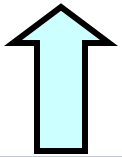


Mendel's 1st Law

- Every organism has two factors - alleles - responsible for the appearance of a given trait
- An organism receives alleles from its parents - one allele from each parent
- There is always only one such factor (allele) in the germ cells (gametes)

MENDEL'S 2ND LAW

- Different gene pairs assort independently in gamete formation.

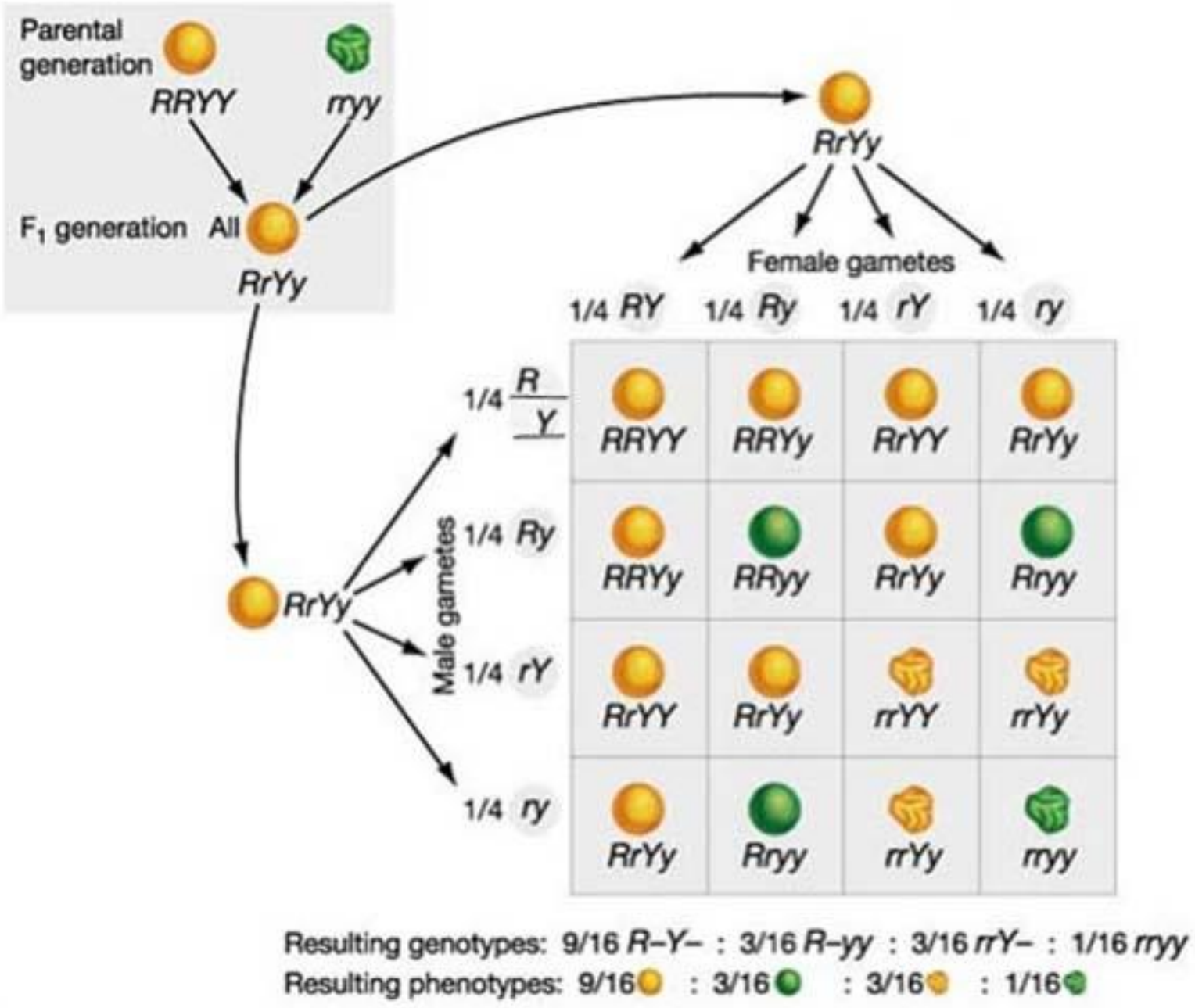


This "law" is true only in some cases.

Gene pairs on **SEPARATE CHROMOSOMES** assort independently at meiosis.

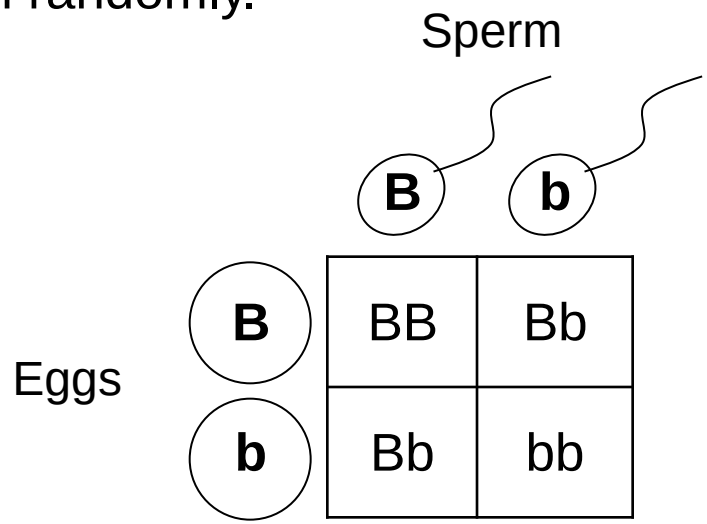
Mendel's 2nd Law

- Genes inherit independently
- Refers to the inheritance of two or more traits
- Traits, e.g. seed colour and seed shape of peas, inherit independently
- It is now known that this is a valid statement as long as the genes determining these traits are not coupled, i.e. they are located in different, non-homologous, chromosomes


Parental generation
RRYY
rryy
F1 generation All
RrYy
RrYy
Female gametes
1/4 RY
1/4 Ry
1/4 rY
1/4 ry
Male gametes
1/4 R/Y
1/4 Ry
1/4 rY
1/4 ry
RrYy
RRYY RRYy RrYY RrYy
RRYy RRyy RrYy Rryy
RrYY RrYy rrYY rrYy
RrYy Rryy rrYy rryy
Resulting genotypes: 9/16 R–Y– : 3/16 R–yy : 3/16 rrY– : 1/16 rryy
Resulting phenotypes: 9/16 : 3/16 : 3/16 : 1/16

Mendel's law of segregation

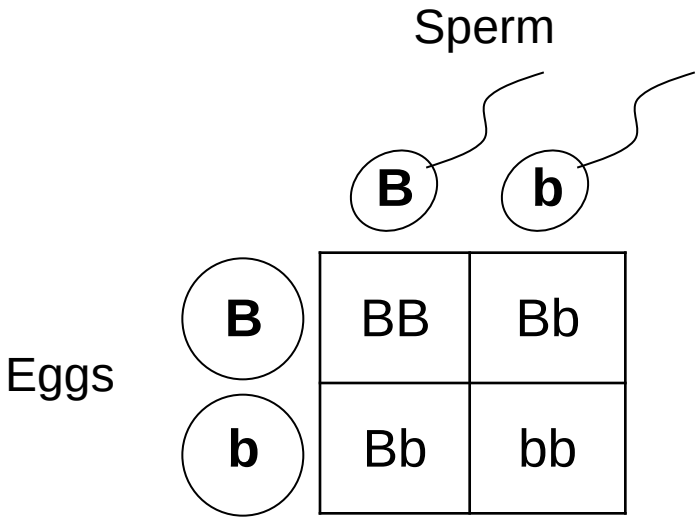
- A normal (somatic) cell has two variants (alleles) for a Mendelian trait.
- A gamete (sperm, egg, pollen, ovule) contains one allele, randomly chosen from the two somatic alleles.
- E.g. if you have one allele for brown eyes (B) and one for blue eyes (b), somatic cells have Bb and each gamete will carry one of B or b chosen randomly.



Mendel's law of dominance

- If your two alleles are different (*heterozygous*, e.g. Bb), the trait associated with only one of these will be visible (dominant) while the other will be hidden (recessive).

  E.g. B is dominant, b is recessive.



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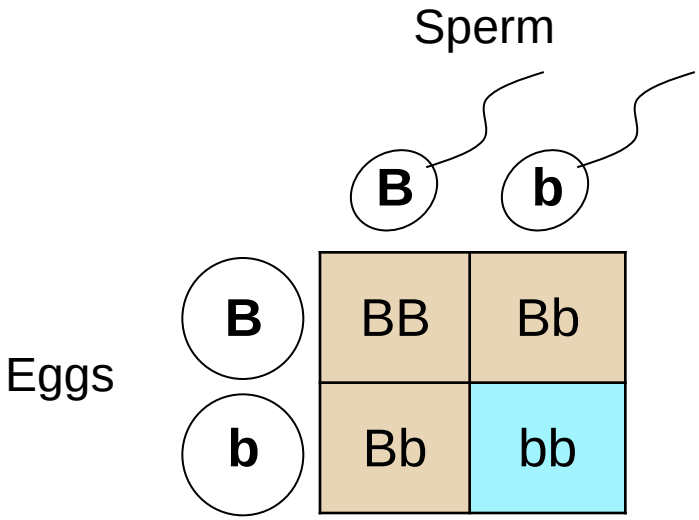

Punnett squares - probability diagram illustrating the possible offspring of a mating.

Testcross

A testcross is designed to reveal whether an organism that displays the dominant phenotype is homozygous or heterozygous.

MENDEL TEST CROSS

- Phenotype of parents
- Genotype of parents
- Gametes
- F_1
- Probability: 100 % purple

Or 50% purple: 50% white

In 2nd case 1 purple: 1 white

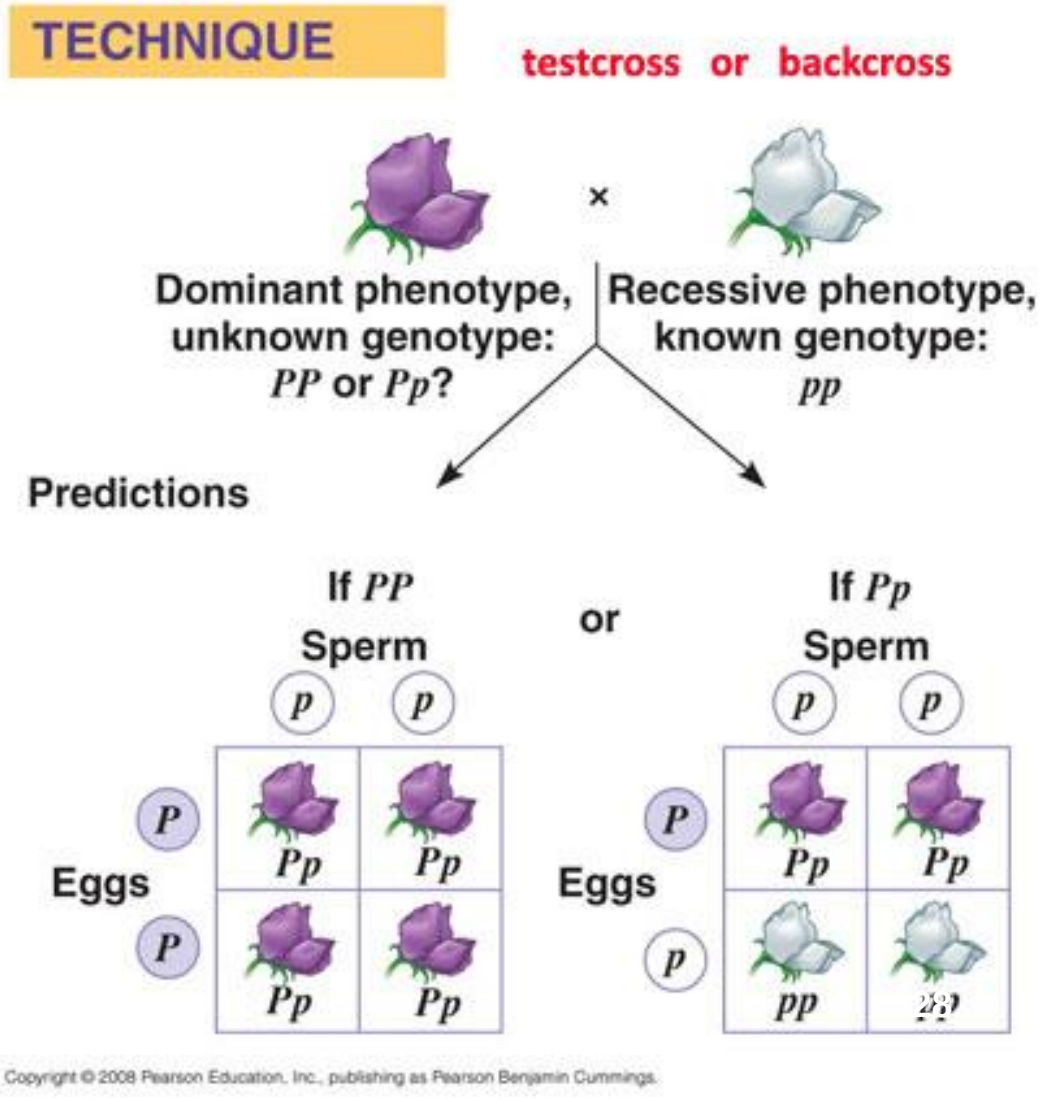

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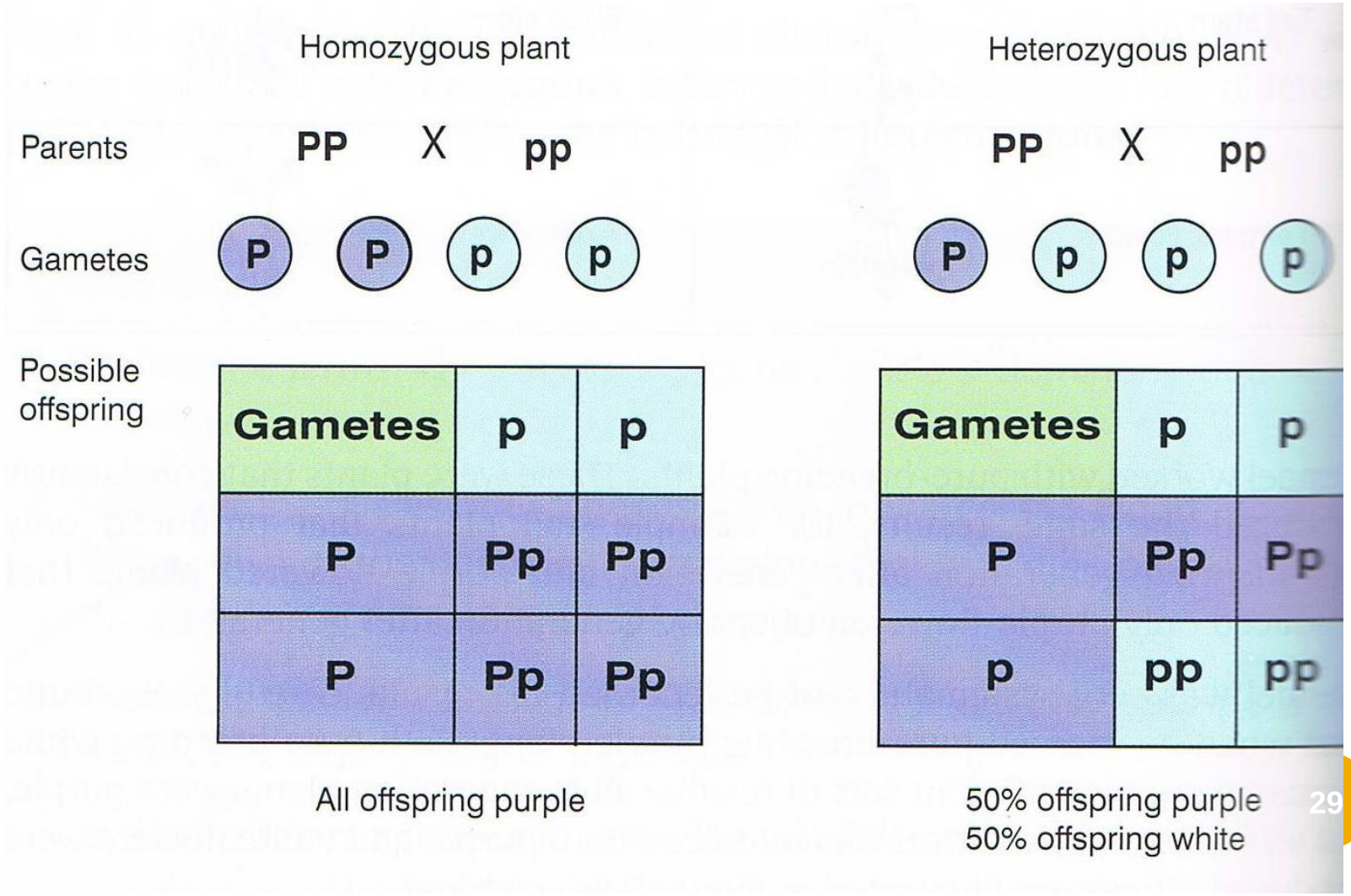
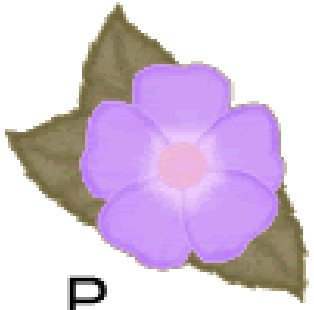
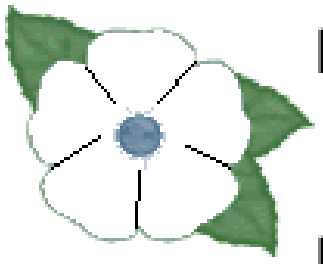
Figure 23.5 Two possible results from the test cross of purple- and white-flowered plants



TEST CROSS DIAGRAM



P ?



p	Pp	Pp or pp
p	Pp	Pp or pp

If all offspring are purple, then parent must be PP
If half are white and half are purple, parent must be Pp

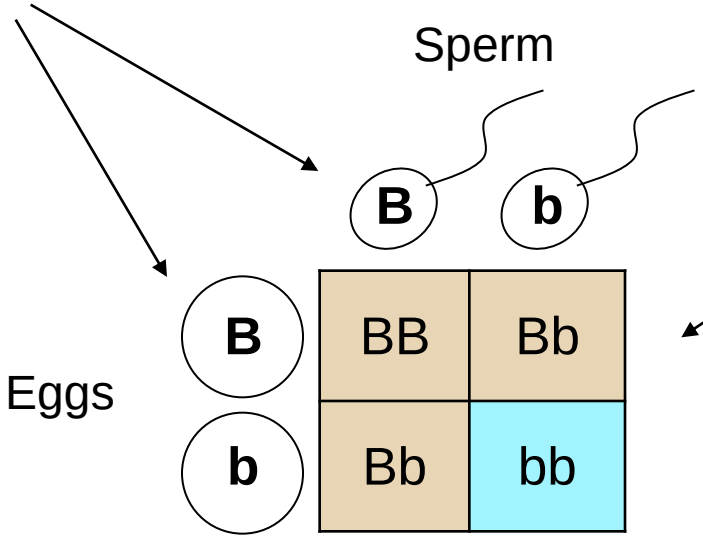
<u>Genotype possibilities:</u>
Pp, pp

<u>Phenotype possibilities:</u>
white, purple

Terminology

Haploid: containing one copy of each chromosome (n=23)

Diploid: containing two copies of each chromosome (2n=46)



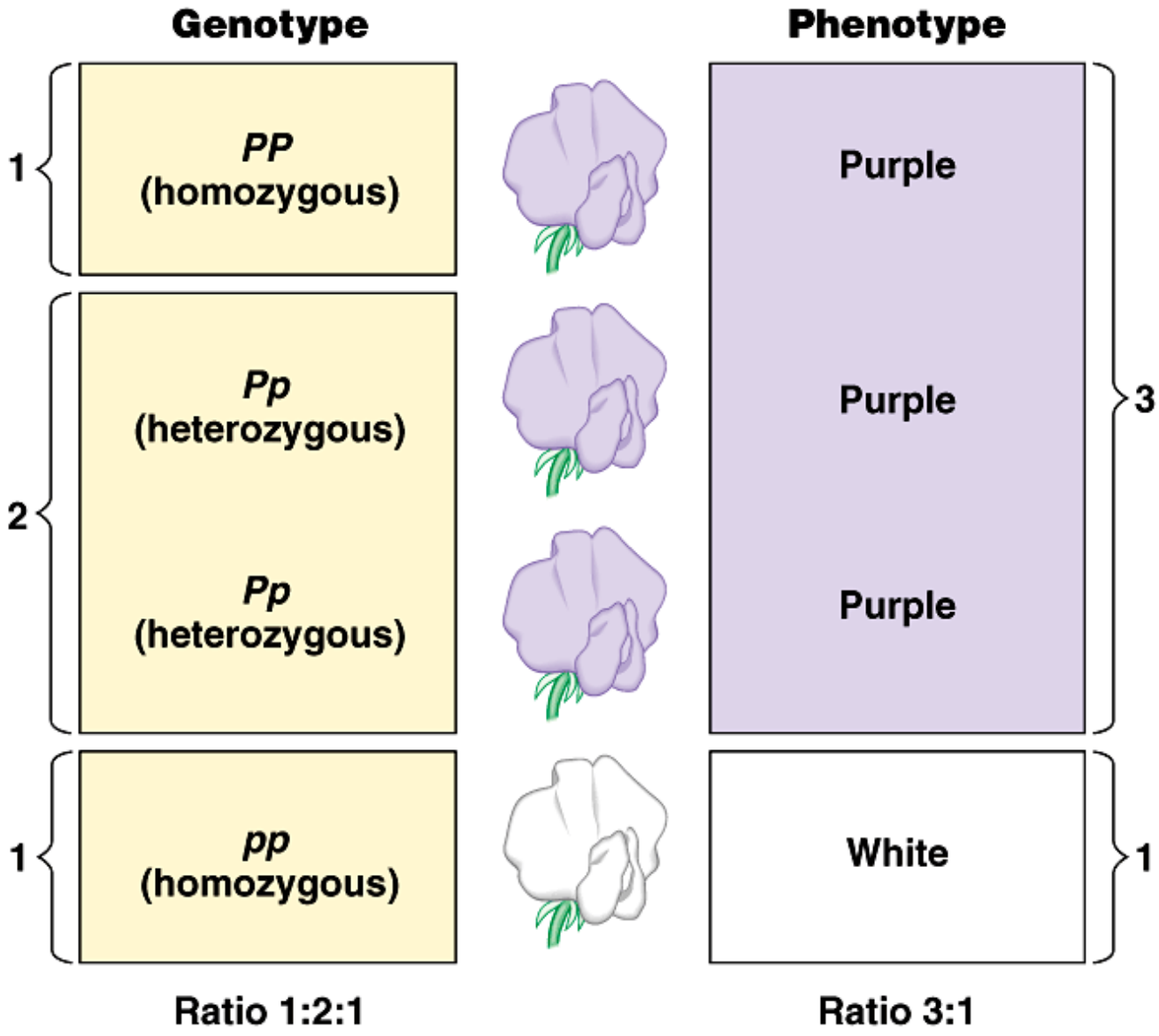
Terminology

- **Genotype**: the states of the two alleles at one or more locus associated with a trait

- **Phenotype**: the state of the observable trait

Genotype	Phenotype
BB (homozygous)	Brown eyes
Bb (heterozygous)	Brown eyes
bb (homozygous)	Blue eyes

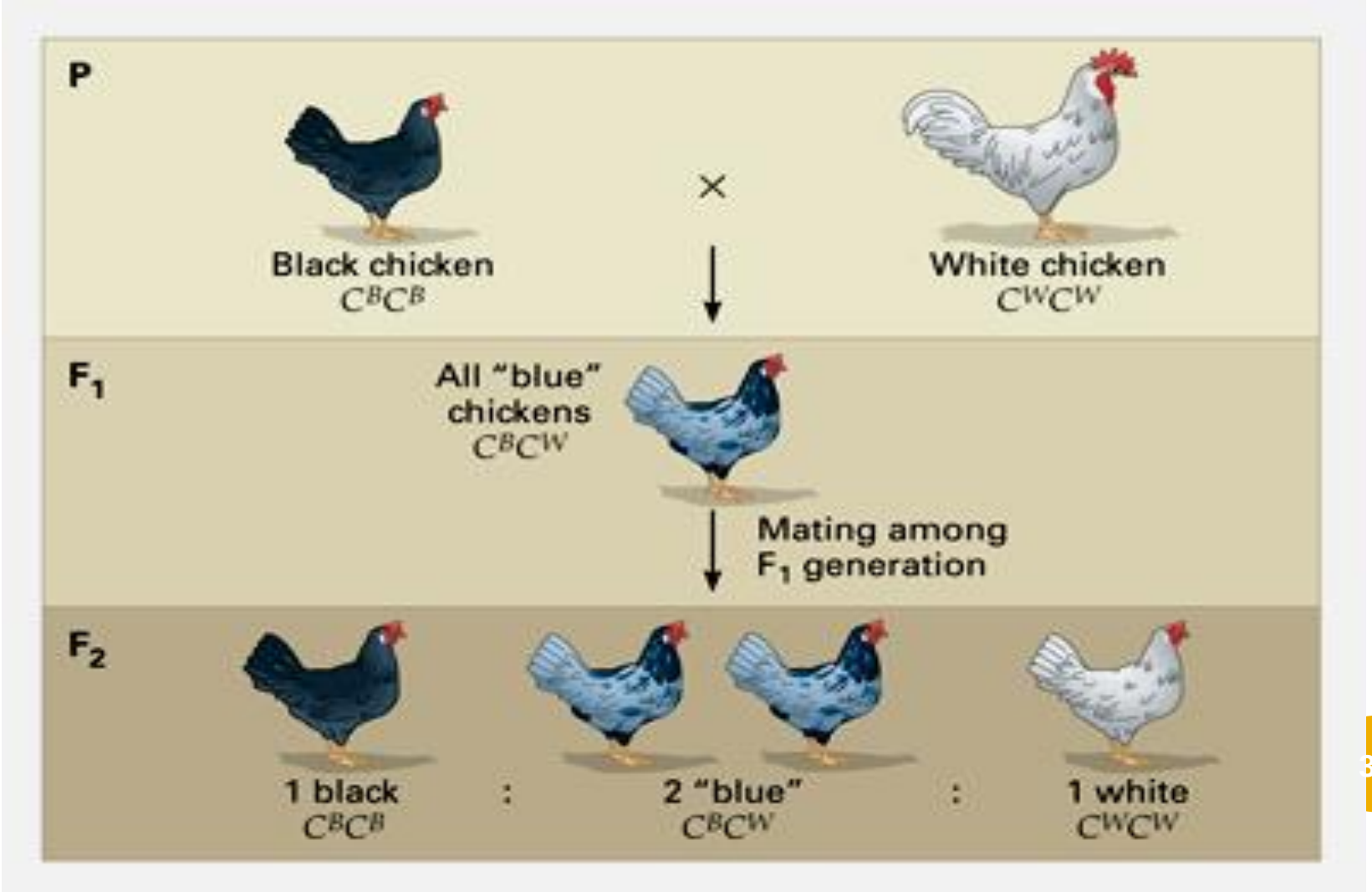
Genotype versus phenotype.



How does a genotype ratio differ from the phenotype ratio?

Variation in Patterns of Inheritance

Intermediate Inheritance (blending): inheritance in which heterozygotes have a phenotype intermediate between the phenotypes of the two homozygotes



	DOMINANT TRAITS	RECESSIVE TRAITS
eye coloring	brown eyes	grey, green, hazel, blue eyes
vision	farsightedness normal vision normal vision normal vision	normal vision nearsightedness night blindness color blindness*
hair	dark hair non-red hair curly hair full head of hair widow's peak	blonde, light, red hair red hair straight hair baldness* normal hairline
facial features	dimples unattached earlobes freckles broad lips	no dimples attached earlobes no freckles thin lips

* sex-linked character

Morgan's chromosome theory of heredity

- Thomas Morgan, American scientist, early 20th century
- Work based on experimental studies and mathematical calculations on fruit flies (Drosophila melanogaster)
- The fruit fly as a model organism has many advantages:
  - small size
  - clear differences between females and males (sexual dimorphism)
  - well expressed traits dependent on single genes
  - ease of breeding
  - low chromosome number (3 autosomes + 1 sex chromosome)
  - significant fertility
  - short life cycle

Morgan's chromosome theory of heredity

Main assumptions:

- Genes are located in the chromosomes of cell nuclei (chromosome theory of heredity)
- Genes are duplicated (replicated)
- Each gene occupies a well-defined location on a chromosome (locus)
- Alleles of the same gene are located in homologous chromosomes (from father and mother) exactly opposite each other, they have the same locus
- Alleles of different genes occupy different positions
- Offspring cell genes are identical to the parental genes

Morgan's chromosome theory of heredity

- Genes are arranged linearly in the chromosomes in a well-defined order and sequence
- Genes located on the same chromosome are genes that are linked, and genes located on separate chromosomes are unlinked
- Only genes that lie on different chromosomes and are uncoupled are inherited independently
- Genes located in the same chromosomes form a so-called coupling group
- This means that they are inherited together, which is incompatible with Mendel's second law
- Coupled genes are inherited together, but not always completely
- During meiosis, each of the homologous chromosomes undergoes random segregation into one of the cells

Morgan's chromosome theory of heredity

- Gene uncoupling occurs as a result of the crossing-over process
- Crossing-over is a phenomenon that takes place in gametes at the beginning of meiosis
- Involves the exchange of chromatid segments between homologous chromosomes
- Connections called chiasms are formed between the chromatids
- These then break down and sections of chromosomes are exchanged
- The frequency of crossing-over is determined by the position of the genes in the chromosome
- If genes are close to each other, the crossing-over rate is low
- The c.o. results in 4 classes of individuals: 2 have the same trait arrangement as the parents (these are the most numerous classes), and 2 are recombinants - they have a different arrangement of genes on the chromosome than the parents

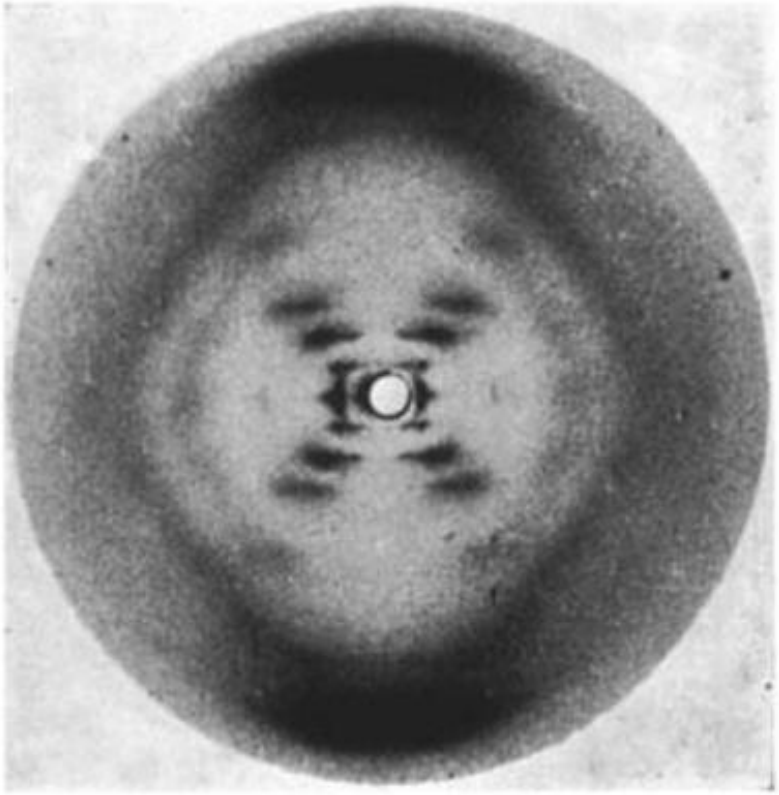
Morgan's chromosome theory of heredity

- The greater the distance between two genes on a chromosome, the greater the frequency of crossing-over.
- Based on the frequency of crossing-over, the position of genes on the chromosomes can be determined and chromosome maps can be drawn up (determine the position of genes on a chromosome)
- The results of genetic cross-overs depend on whether genes are located on one chromosome or on other chromosomes
- 1933 - Morgan received the Nobel Prize for the chromosome theory of inheritance

Discovering the structure of DNA

- 28 February 1953 - discovering the structure of DNA: James Watson and Francis Crick announced in The Eagle Pub that they had solved the secret of life (as immortalised by a plaque in that pub)
- James Watson, Francis Crick, Maurice Wilkins, Rosalind Franklin

Discovering the structure of DNA

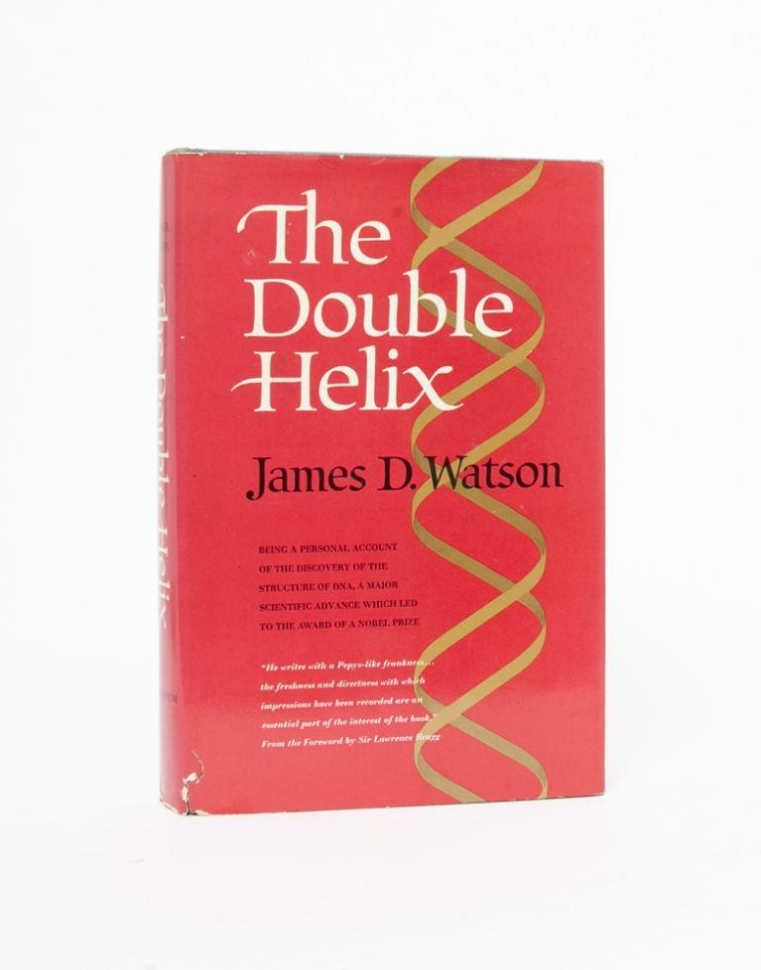


Photograph 51' showing a DNA sodium crystal made by Rosalind Franklin

Discovering the structure of DNA

- 1962 r. - award of the Nobel Prize: Francis Crick, James Watson and Maurice Wilkins
- Nobel Prize - Rosalind Franklin (died 4 years earlier) and Erwin Chargaff were omitted
- Chargaff's rule: guanine content equals cytosine content, while thymine content equals adenine content

Discovering the structure of DNA



"The Double Helix"

An autobiographical book by American geneticist James Watson describing the story of the discovery of the structure of DNA