Mendelian Inheritance

What genetic principles account for the transmission of traits from parents to offspring?

Theories of Inheritance

- One possible explanation of heredity is "**blending**" **hypothesis**
  - The idea that genetic material contributed by two parents **mixes** in a manner analogous to the way **blue** and **yellow** paints blend to make **green**.
- An alternative to the blending model is the "**particulate**" **hypothesis** of inheritance: the gene idea
  - -Parents pass on discrete **heritable units**, **genes**
  - -This hypothesis can explain the **reappearance** of traits after several generations
- Mendel documented a **particulate** mechanism through his experiments with garden peas.

Others: **Homunculus**, **Pangenesis**

Mendelian Genetics

- Gregor Johann Mendel (1822-1884)
  - Augustinian **monk**, Czech Republic
  - Foundation of modern genetics
  - Studied segregation of traits in the garden pea (*Pisum sativum*) beginning in 1854
  - Published his theory of inheritance in 1865. “Experiments in Plant Hybridization”
  - Mendel was “rediscovered” in 1902
  - Ideas of inheritance in Mendel’s time were vague.

Mendel used the **scientific approach** to identify/ formulate the laws of inheritance

Mendel discovered the **basic principles** of heredity by breeding garden peas in carefully planned experiments

Mendel's Experimental, Quantitative Approach

- Advantages of pea plants for genetic study:
  - There are many varieties with distinct heritable features, or **characters** (such as flower color); **character variants (such as purple or white flowers) are called traits**
  - Mating can be controlled
  - Each flower has sperm-producing organs (stamens) and egg-producing organ (carpel)
  - Cross-pollination (fertilization between different plants) involves dusting one plant with pollen from another

Themes of Mendel's work

- Variation is widespread in nature.
- Observable*** variation is essential for following hereditary factors (genes).
- Variation is **inherited** according to genetic laws and not solely by chance.
- Mendel's laws apply to all **sexually** reproducing organisms.

Heredity

- Heredity - the **transmission** of traits from one generation to the next

- Genetics – the scientific study of heredity and hereditary variation

- Genes – hereditary units endowed from parents
  - **Segments of DNA**
  - **Divided into Chromosomes**
  - **46 in humans**
  - **A gene's specific location on a chromosome is called its locus**

Asexual Reproduction

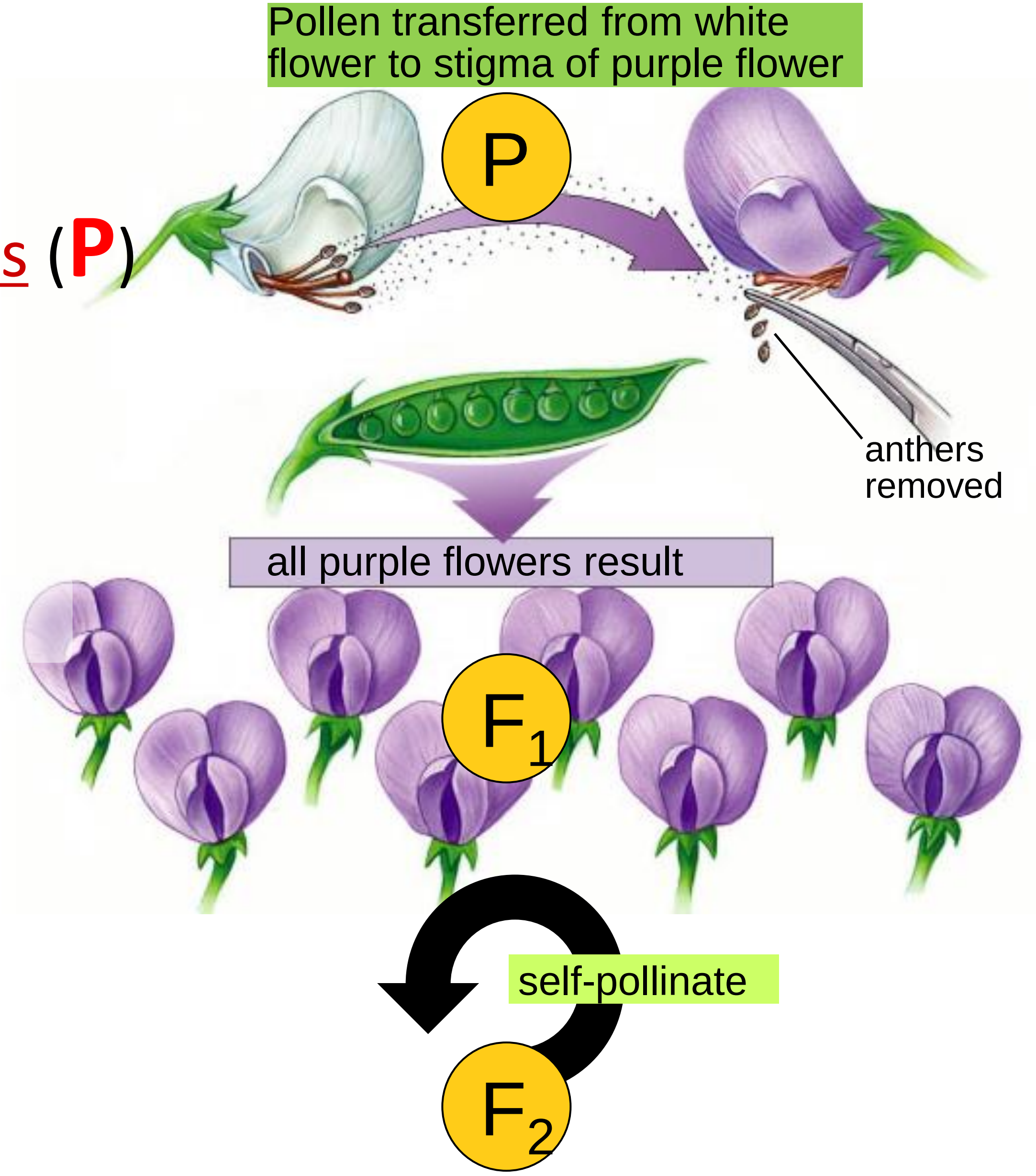
- **One** parent
- Binary fission in bacteria
- Single cell eukaryotes : mitotic cell division
  - DNA is copied and divided equally between daughter cells
- Multicellular organisms – **Budding**
  - Hydra : Buds break off – are **genetically identical** to its parent
- Each offspring in asexual reproduction is called a **clone**

Sexual Reproduction

- **Two** parents
- Results in **greater variation** than asexual reproduction
- Offspring **vary genetically** from siblings and both parents
  - **Behavior of chromosomes during the sexual lifecycle**

Mendel's work

- Bred pea plants
  - cross-pollinate true breeding parents (**P**)
    - P = parental
  - raised seed & then observed traits (**F_1**)
    - F = filial
  - allowed offspring to self-pollinate & observed next generation (**F_2**)



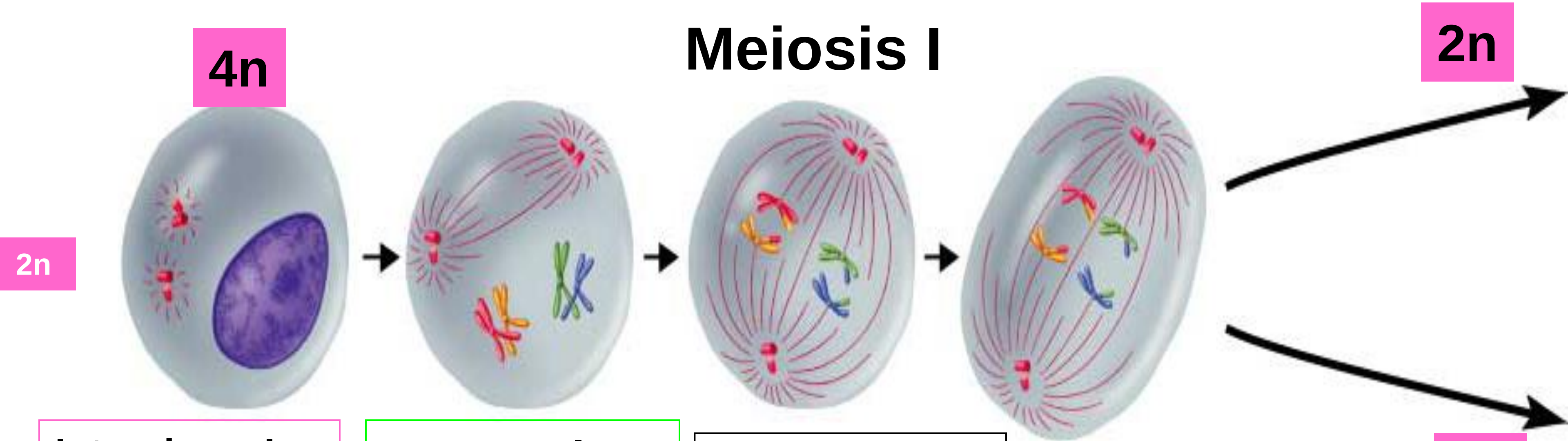
Meiosis

- Cell division to form the **gametes**; sperm (male gamete) and egg (female gamete) - is a characteristic of **eukaryotes** only
- Normal cells are **diploid**: 2 copies (**alleles**) of every gene.
- Gametes are **haploid**: 1 copy of every gene. **What are the advantages of being diploid ?** **(what about triploid, tetraploid etc?)**
- Diploidy is useful because 2 copies of every gene means that there a **backup** copy if one gets **mutated**.

Mutations are very frequent in the cells of large organisms. We wouldn't survive with just one copy of each gene.

- Why have sexual reproduction?
- **Shuffling of alleles** between parents and offspring leads to new combinations.
  - Bad combinations **die** without reproducing; good combinations **survive** and reproduce more offspring.

Meiosis I



Interphase I

Cells undergo a **round of DNA replication**, forming duplicate Chromosomes.

Prophase I

Each chromosome **pairs** with its corresponding **homologous** chromosome to form a tetrad.

Metaphase I

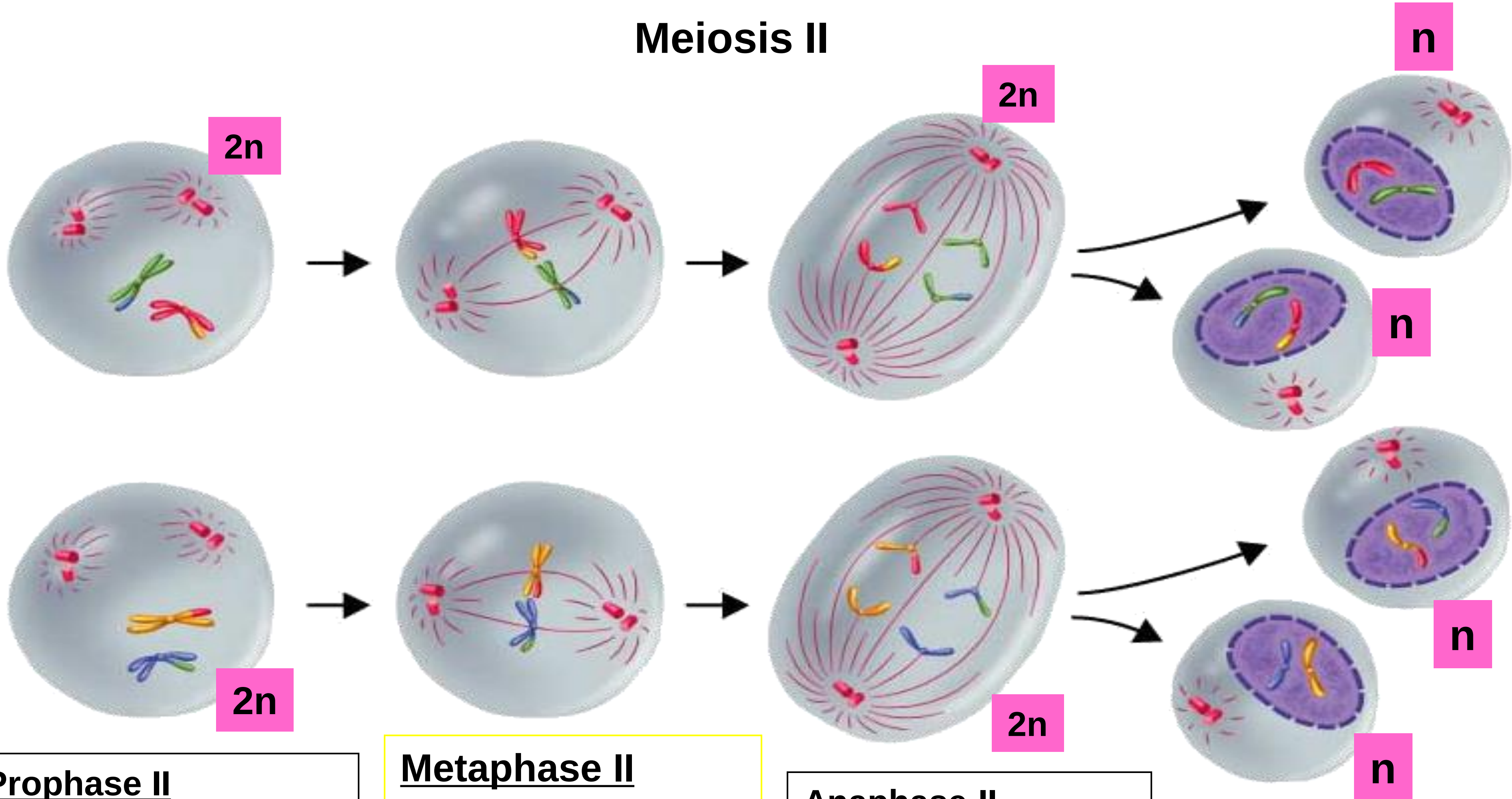
Spindle fibers attach to the chromosomes
.

Anaphase I

The fibers pull the homologous chromosomes toward the opposite ends of the cell.

- The 2 cells produced in Meiosis 1 then go through **another meiotic division**.
- **There is no DNA replication between Meiosis I and Meiosis II.**
- The end result of Meiosis II is **4 cells** with **2 sister chromatids** in them.

Meiosis II



Prophase II

Meiosis I results in two **haploid** (2n) daughter cells, each with half the number of chromosomes as the original.

Metaphase II

The chromosomes **line up** in a similar way to the metaphase stage of mitosis.

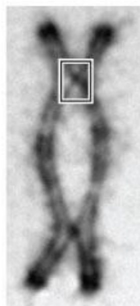
Anaphase II

The sister chromatids **separate** and move toward opposite ends of the cell.

Meiosis II results in four haploid (n) daughter cells.

The results of crossing over during meiosis

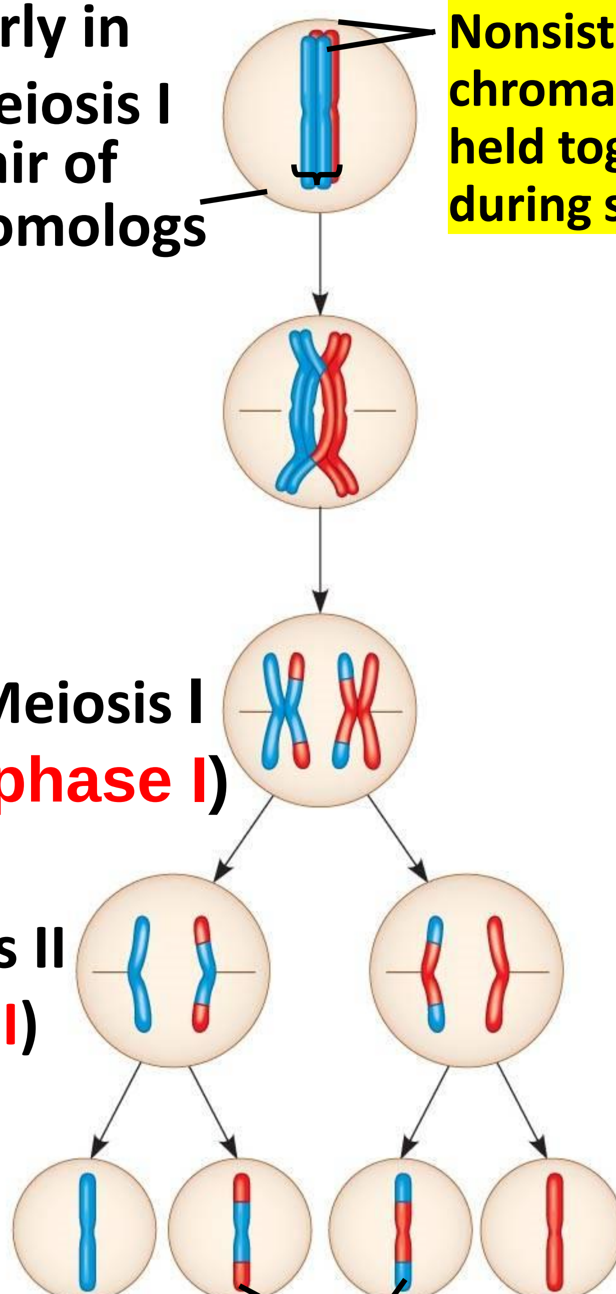
Overview of meiosis: how meiosis reduces chromosome number



Early in Meiosis I

Pair of homologs

Nonsister chromatids held together during synapsis



A single crossing over event leads to 4 genetically unique daughter cells!

during Meiosis I (at anaphase I)

during Meiosis II (at anaphase II)

Parental chromosomes

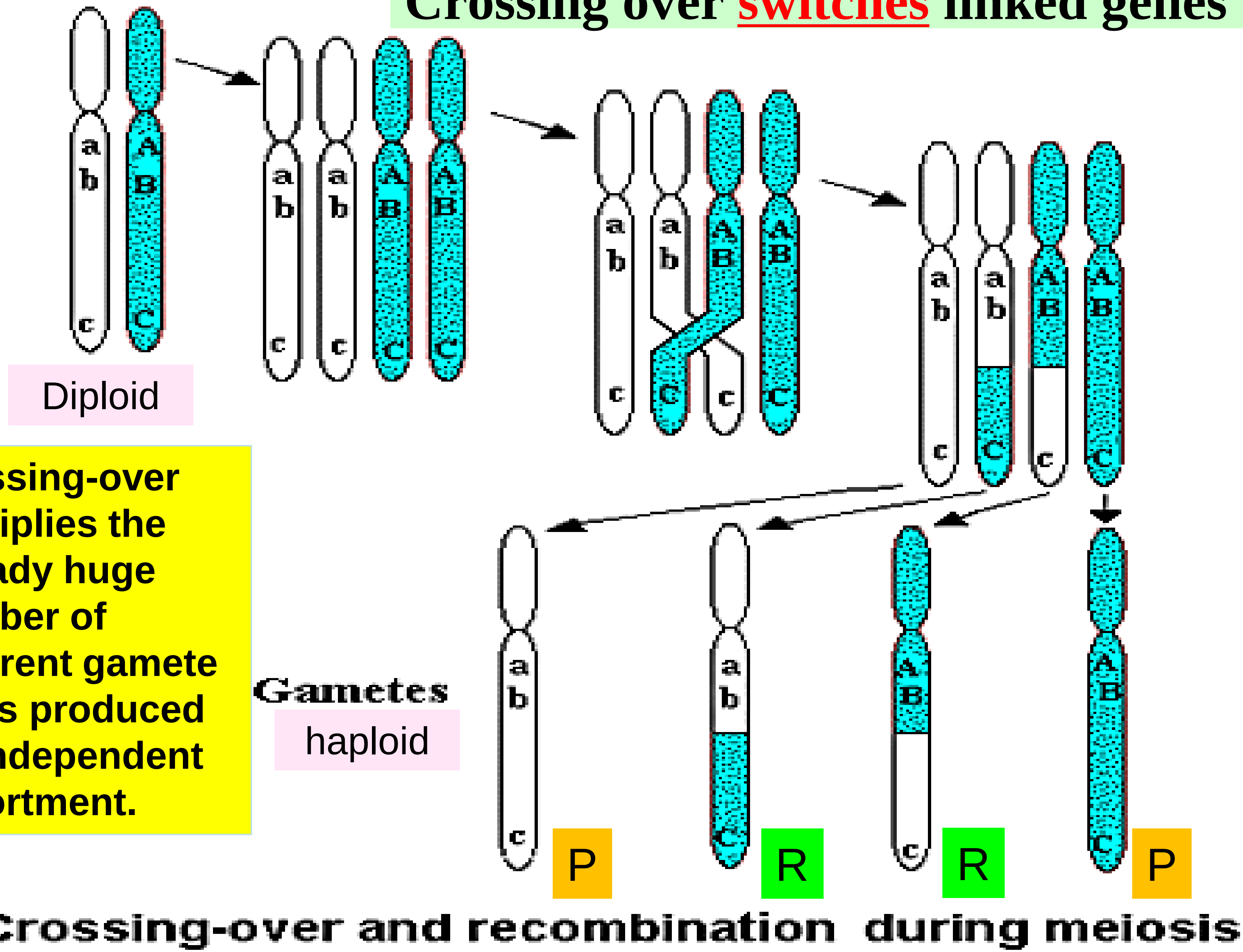
Daughter cells

Gametes - haploid

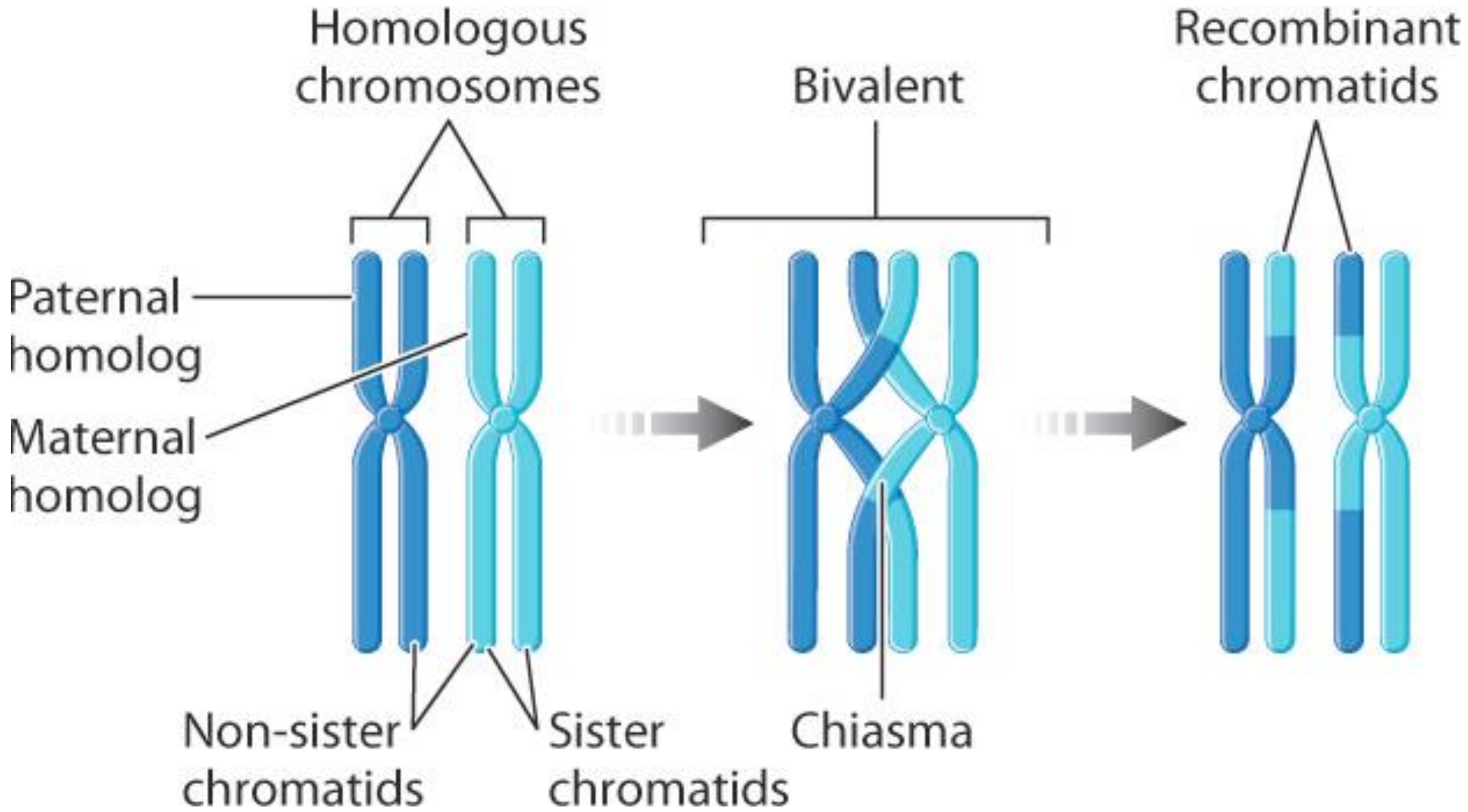
Recombinant chromosomes

Crossing over <u>switches</u> linked genes

Crossing-over multiplies the already huge number of different gamete types produced by independent assortment.



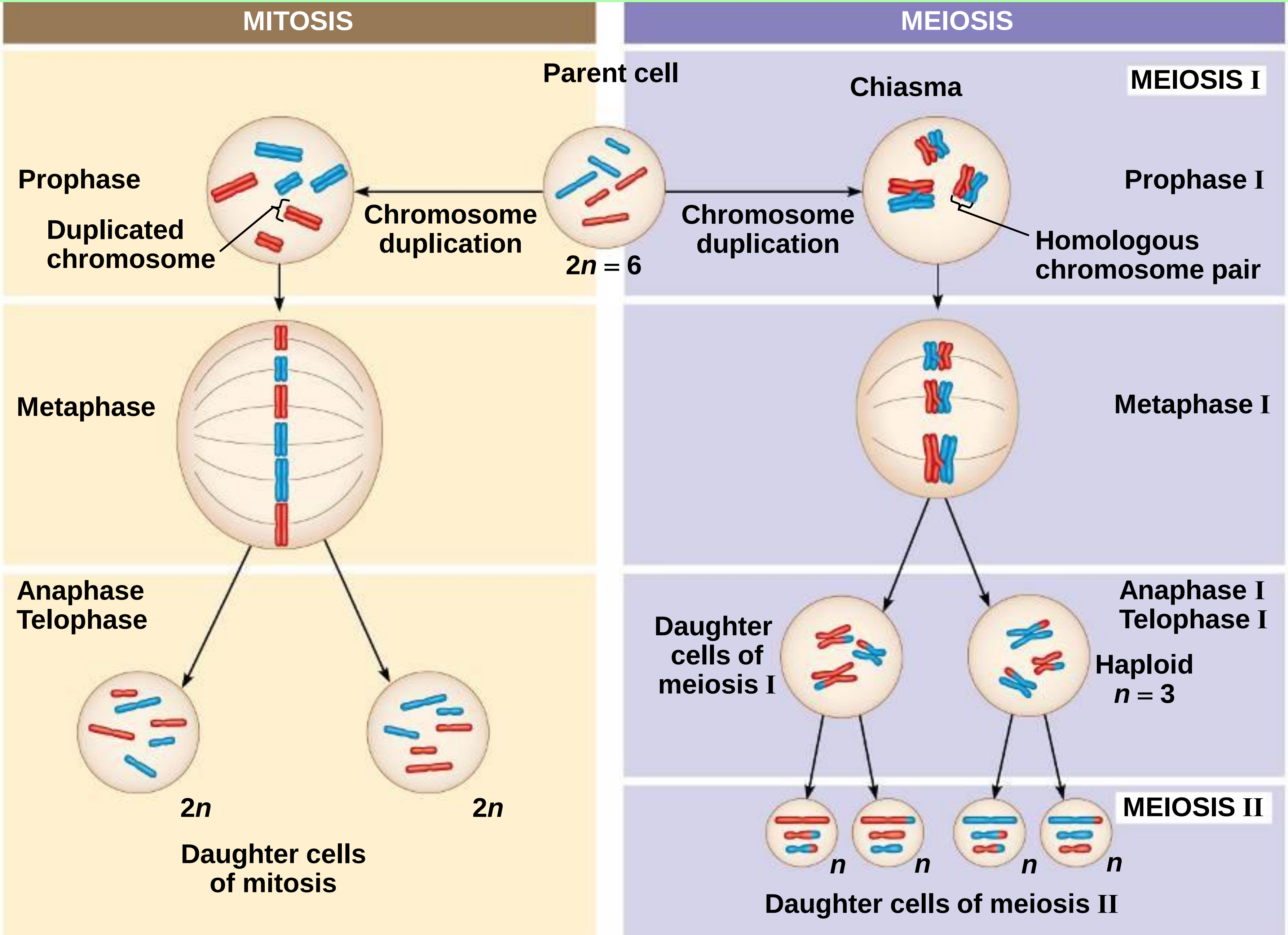
The early stages of meiosis involve pairing of homologous chromosomes and crossing over followed by condensation



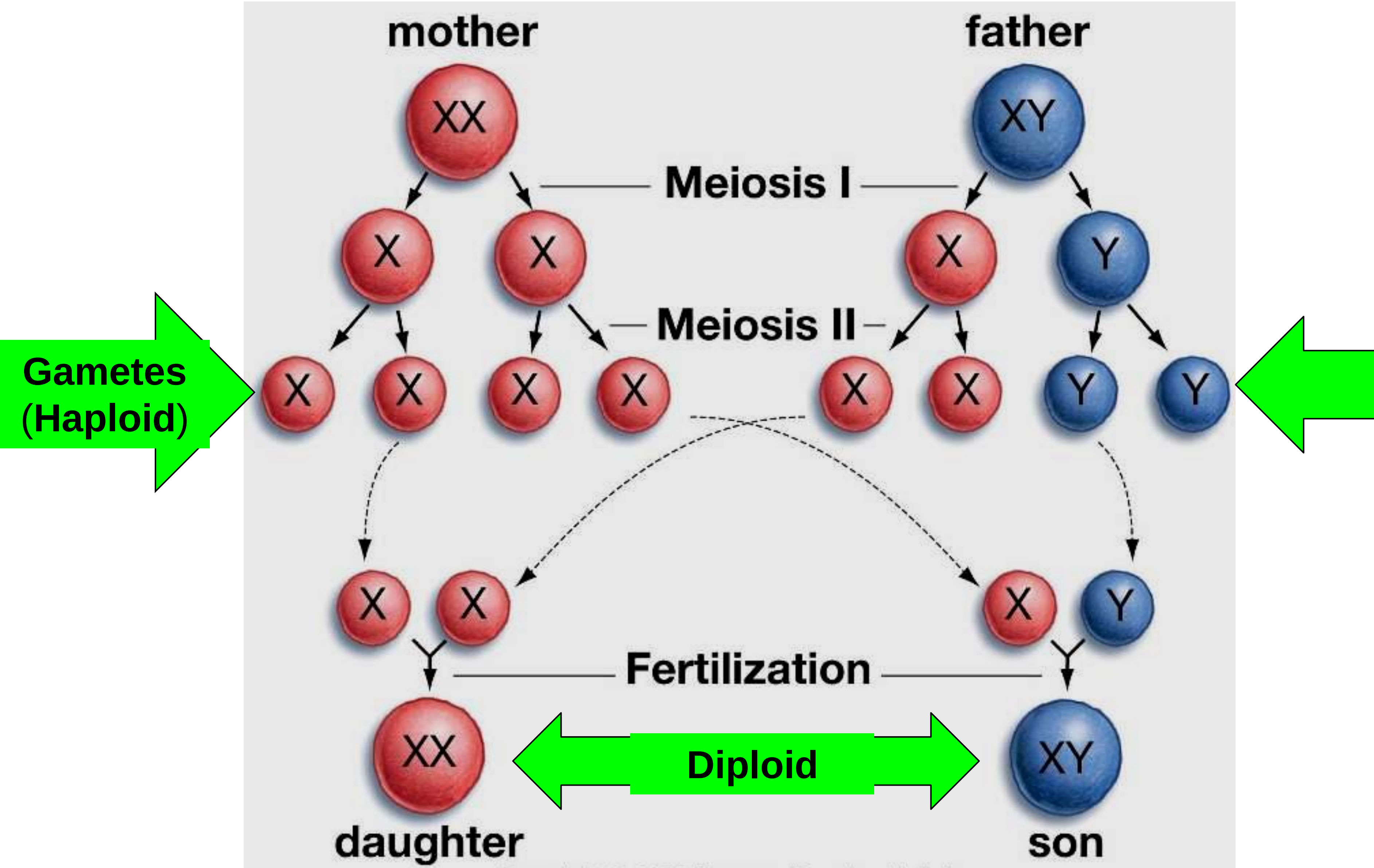
Mitosis and Meiosis differ in several key ways:

		MITOSIS	MEIOSIS
(1)	preceded by replication of chromosomes?	**yes**	**yes**
(2)	# of rounds of cell division	**1**	**2**
(3)	# of daughter cells	**2**	**4**
(4)	# of chromosomes in daughter cells compared to parent cell	**same as parent cell (diploid)**	**half of parent cell (haploid)**
(5)	daughter cells genetically identical to parent cell?	**yes**	**no**
(6)	sister cells thus produced identical to one another?	**yes**	**No** (**gametes**)
(7)	happens in diploid cells, haploid cells, both, or neither?	**both** (***depending on organism***)	**diploid**
(8)	crossing over (synapsis)?	**no**	**yes**

A comparison of mitosis and meiosis in diploid cells.

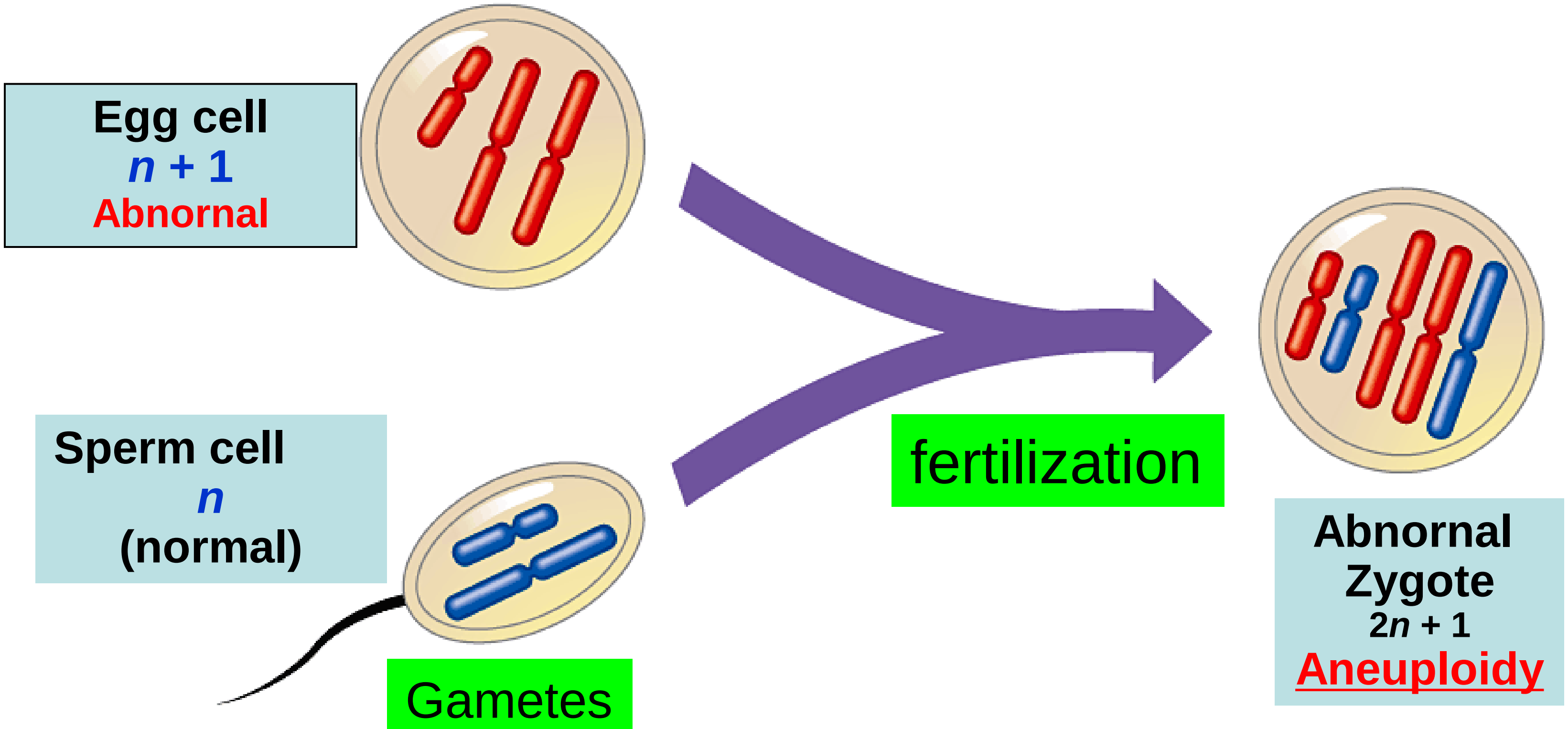


Boy or Girl? The Y Chromosome "Decides"



- Fertilization after **nondisjunction** in the mother results in a zygote with an **extra chromosome**

- Should the gamete with the chromosome pair be fertilized then the offspring will not be 'normal'.
- In humans this often occurs with the 21st pair – producing a child with **Downs Syndrome**



Genetic variation produced in sexual life cycles contributes to evolution

- **Mutations** (changes in an organism's DNA) are the original source of genetic diversity

- Mutations create different versions of genes called **alleles**

- **Reshuffling** of alleles during sexual reproduction produces genetic variation

The Evolutionary Significance of Genetic Variation Within Populations

- Natural selection results in the **accumulation** of genetic variations **favored** by the environment.

- Sexual reproduction **contributes** to the genetic variation in a population, which **originates from mutations.**

- Three mechanisms contribute to genetic variation:
  - Independent assortment of chromosomes (**metaphase I**)
  - Crossing over **(Prophase I)**
  - Random fertilization (**when gamete unite to form Zygote**)

(this is in addition to mutations)

- Each zygote has a **unique** genetic identity.

The Evolutionary Significance of Genetic Variation Within Populations

Example:

- in pea plants, there are 7 pairs of chromosomes; each of the 7 pairs can line up in 2 different ways…this means that there are 128 different sperm or egg cells possible!

  (2^7 = 128)

- And…since there are 128 different possible sperm cells, and 128 different possible egg cells, the # of possible combinations is:

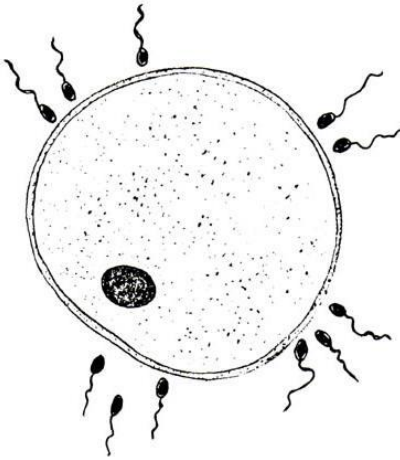
  128 x 128 = 16,384!

- So how many combinations are possible in humans???

  Answer:

- 2^{23} = **8,388,608 different egg** *or* **sperm** cells…and…

  2^{23} X 2^{23} = over 70 trillion different zygotes possible!



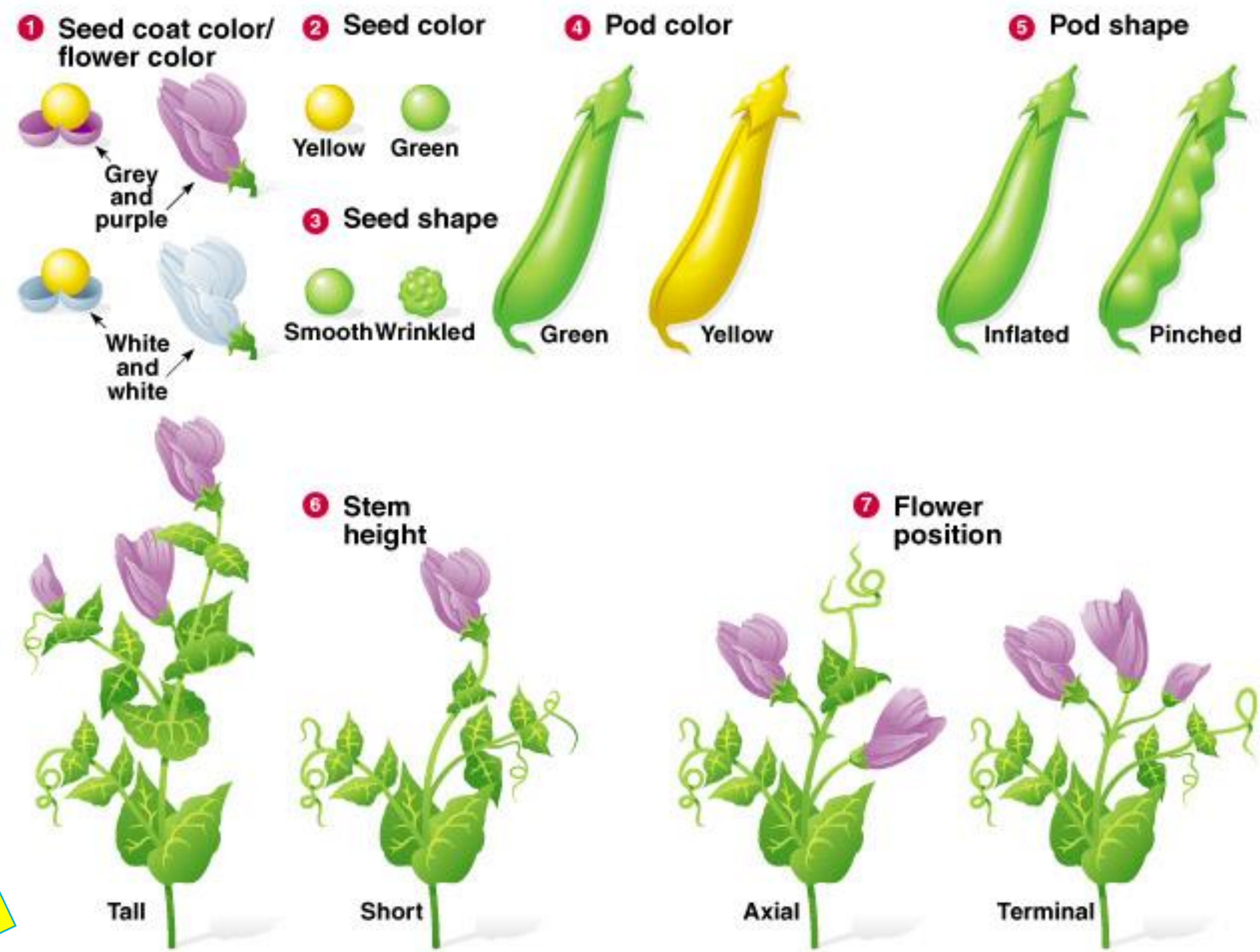
Variation

- Important to population as the raw material for **NATURAL SELECTION.**

- Strongest "most fit" survive to reproduce & pass on traits

Inheritance of Genes

Mendel chose to track only those characters that occurred in two distinct alternative forms



Antagonistic traits

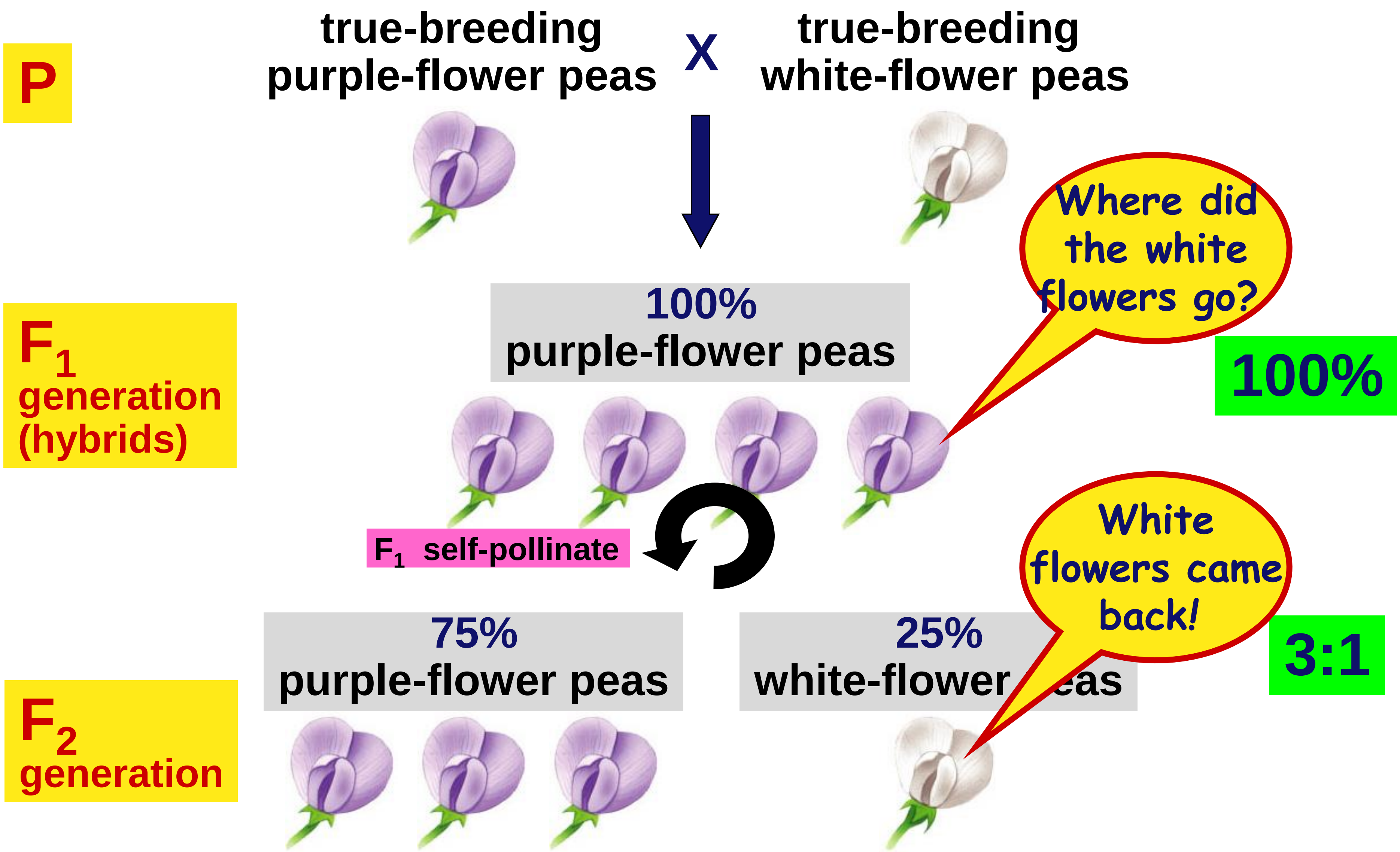
Inheritance of Genes

- He also used varieties that were **true-breeding** (plants that produce offspring of the same variety when they self-pollinate)

Repeated brother-sister mating leads to completely homozygous genome - no variation

- In a typical experiment, Mendel crossed two contrasting, true-breeding varieties, a process called **hybridization**
- The true-breeding parents are the **P generation**
- The hybrid offspring of the P generation are called the **F_1 generation**
- When **F_1** individuals **self-pollinate** or **cross- pollinate** with other F_1 hybrids, the **F_2 generation** is produced

Mendel's Experiments



Law of Dominance

- The **factor** for white flowers **was not diluted** or **destroyed** because it **reappeared** in the F_2 generation(**white was masked –recessive**).

- Traits are controlled by two factors that are either "**dominant**" or "**recessive**."
- In a cross of parents that are pure for contrasting traits, only one form of the trait will appear in the next generation.

- Offspring that are hybrid for a trait will have only the dominant trait in the phenotype.

  - A "dominant" trait shows if the offspring inherits **at least one dominant factor** from one parent.

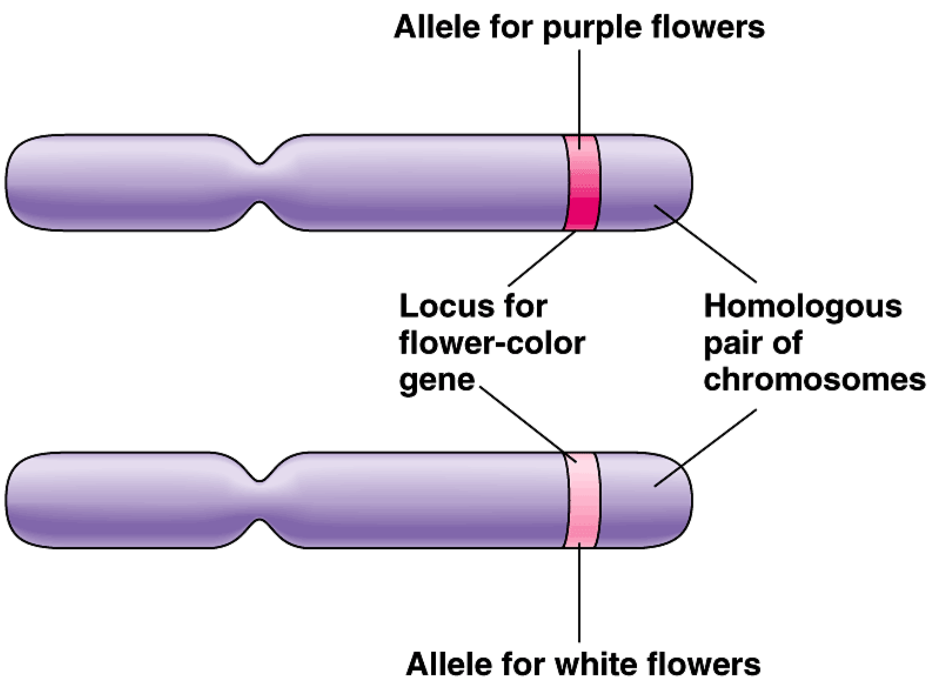
  - A "recessive" trait shows only if the offspring **inherits two recessive factors**, one from each parent.

PP x pp yields all Pp (Purple flowers)

- **Traits come in alternative versions**
  - **Purple** *vs* **white** flower color
  - For each characteristic, an organism inherits 2 alleles, 1 from each parent - **diploid**
  - **Alleles**
    - **different alleles vary in the sequence of nucleotides at the specific locus of a gene**
      - **some difference in sequence of A, T, C, G**

purple-flower allele & **white-flower allele are two DNA variations at flower-color locus**

different versions of gene at same location on homologous chromosomes

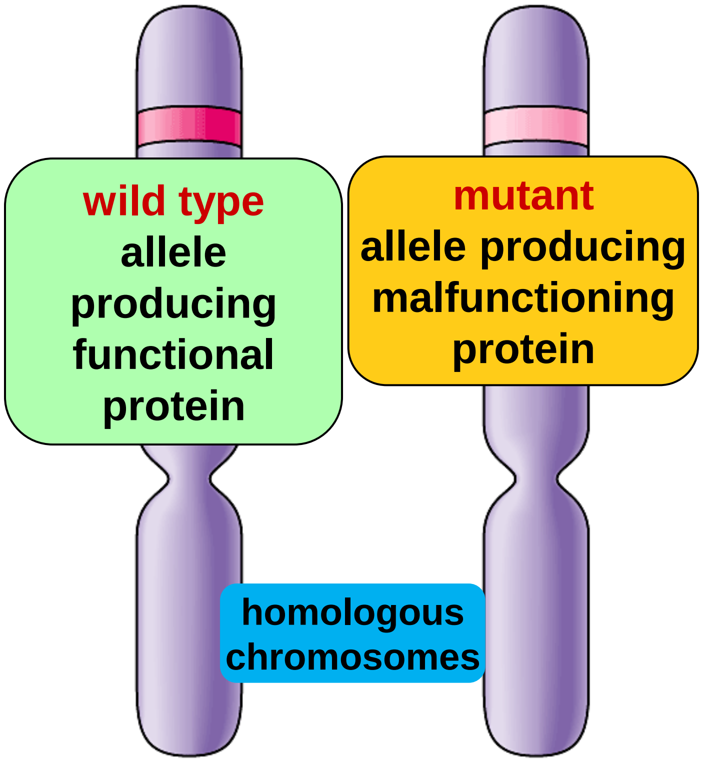


Some traits mask others

- purple & white flower colors are separate traits that do not blend
  - purple x white ≠ light purple
  - purple masked white

In real life situations

Dominant allele producing a *functional* protein masks other alleles (**recessive allele**) producing a *malfunctioning* protein - assuring an organism's survival



Mendel's Model

- Mendel developed a hypothesis to explain the inheritance pattern he observed in $\mathbf{F_1}$ (**100%**) & $\mathbf{F_2}$ (**3:1**) offspring

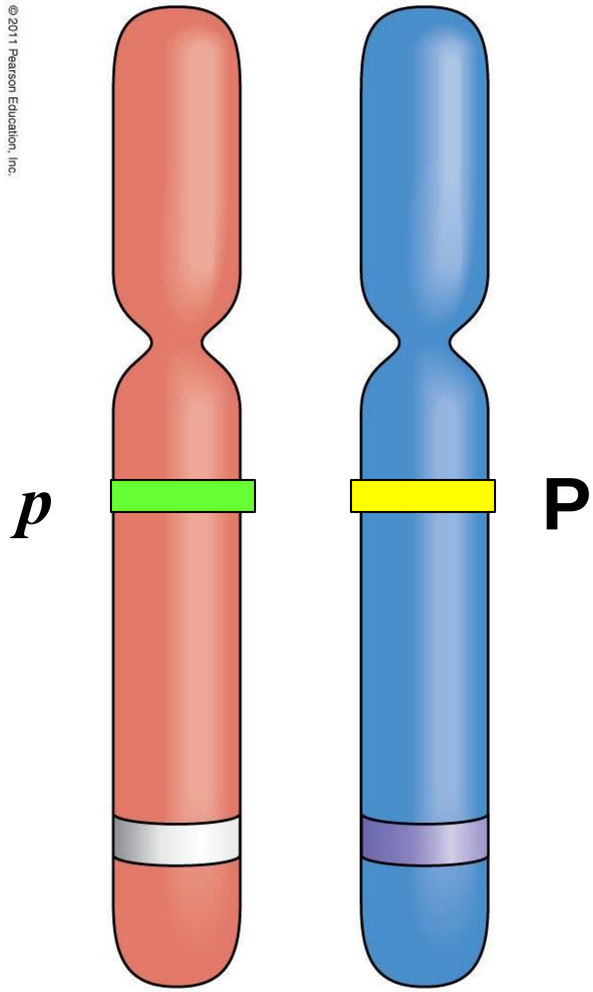
- These concepts can be **related** to what we now know about **genes** and **chromosomes.**

- **The led formulation of the THREE laws of heredity**

Alleles, Locus

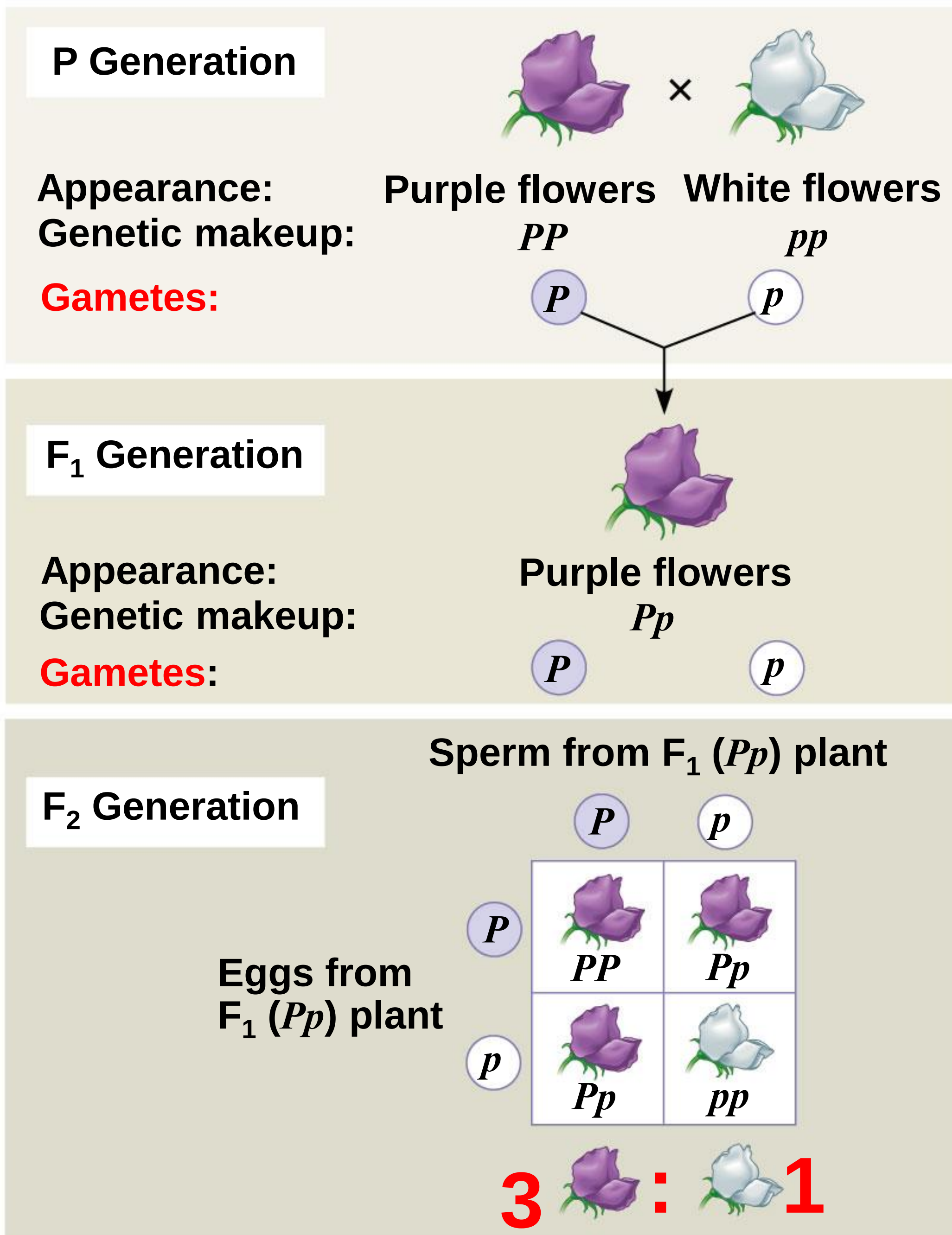
Pair of homologous chromosomes - MONOHYBRID

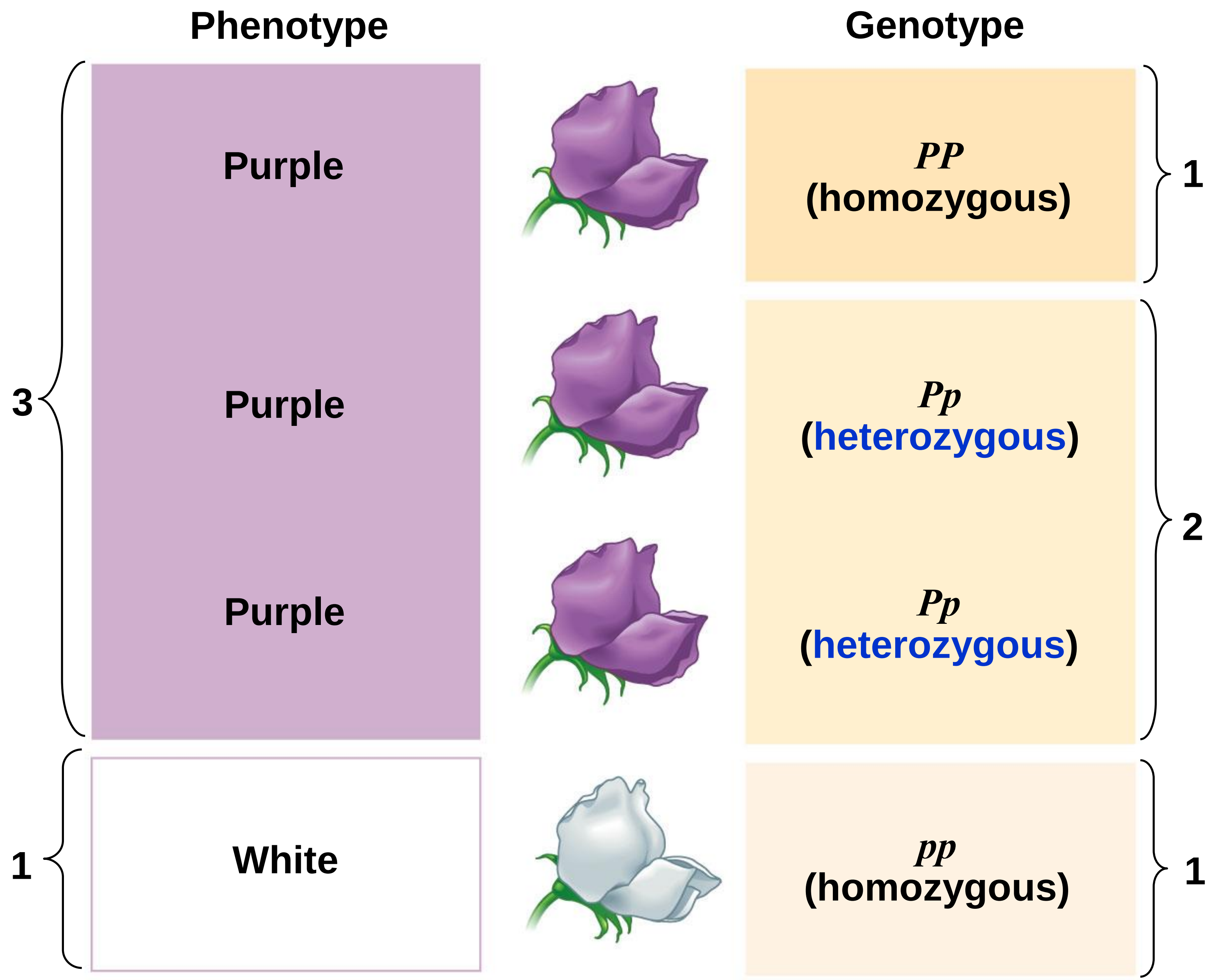
Allele
for white color



Allele
for PURPLE color

Alleles, alternative versions of a gene


P Generation
Appearance:
Genetic makeup:
Gametes:
Purple flowers
PP
White flowers
pp
P
p
F1 Generation
Appearance:
Genetic makeup:
Gametes:
Purple flowers
Pp
P
p
F2 Generation
Sperm from F1 (Pp) plant
P
p
Eggs from
F1 (Pp) plant
P
p
PP
Pp
Pp
pp
3 : 1



Ratio 3:1 | Ratio differ | Ratio 1:2:1

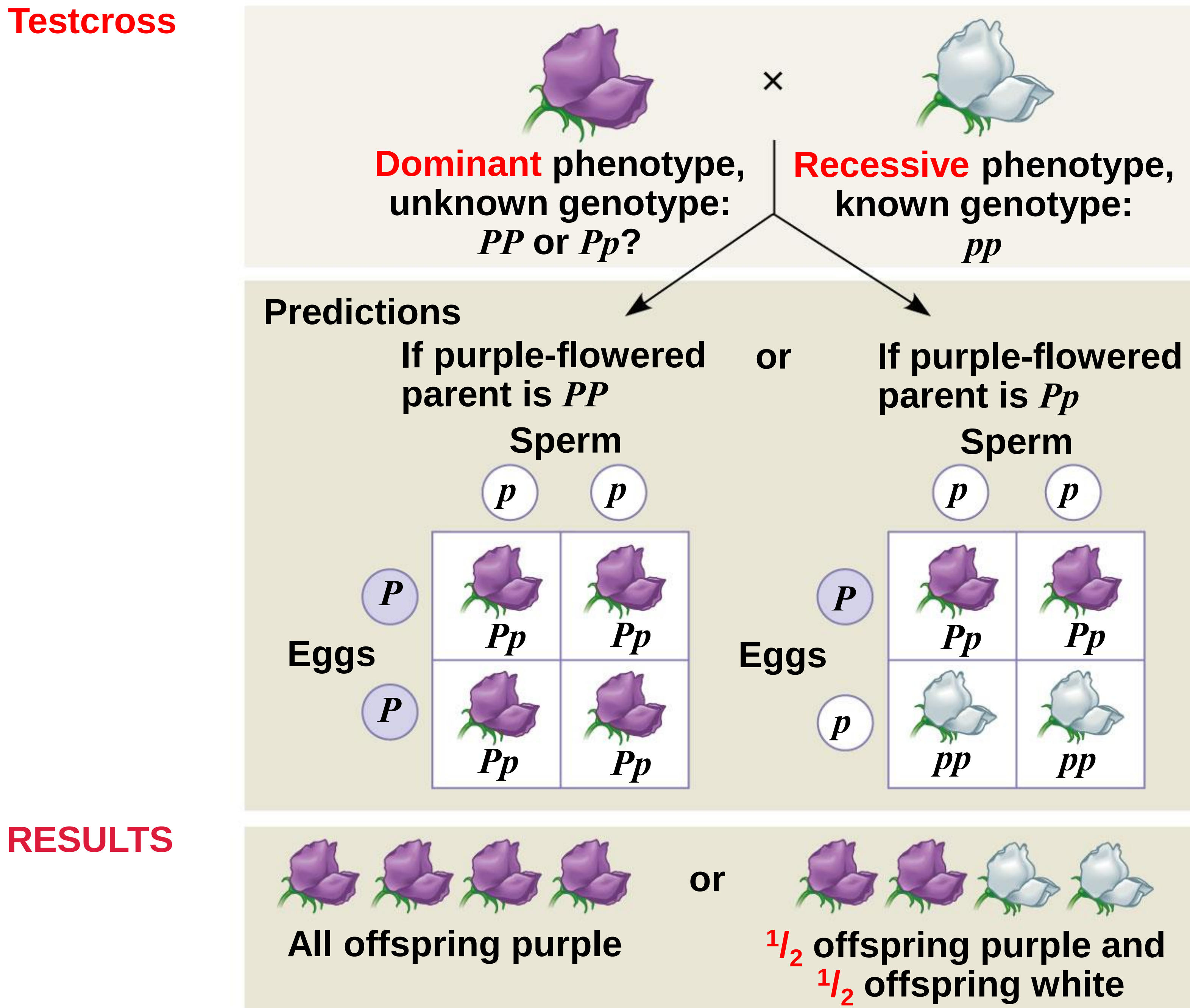
The Testcross

- How can we tell the genotype of an individual with the **dominant** phenotype?

- Such an individual could be either **homozygous** dominant or **heterozygous**

- The answer is to carry out a **testcross**: breeding the mystery individual with a homozygous recessive individual

- If any offspring display the recessive phenotype, the mystery parent must be heterozygous


Testcross
×
Dominant phenotype, unknown genotype: PP or Pp?
Recessive phenotype, known genotype: pp
Predictions
If purple-flowered parent is PP
or
If purple-flowered parent is Pp
Sperm
p
p
Eggs
P
P
Pp
Pp
Pp
Pp
Sperm
p
p
Eggs
P
p
Pp
Pp
pp
pp
RESULTS
All offspring purple
or
1/2 offspring purple and 1/2 offspring white

The Law of Independent Assortment

- Mendel derived the law of segregation by following a **single character**

- The **F_1** offspring produced in this cross were **monohybrids**, individuals that are heterozygous for one character

- A cross between such heterozygotes is called a **monohybrid cross.**

- Mendel identified his second law of inheritance by following **two characters** from parents to **F2** at the same time.

- Crossing two true-breeding parents differing in two characters produces **dihybrids** in the **F_1** generation, heterozygous for both characters

- A **dihybrid cross -** a **cross between F_1 dihybrids**, can determine whether two characters are transmitted to offspring as a package or independently

Dihybrid Cross

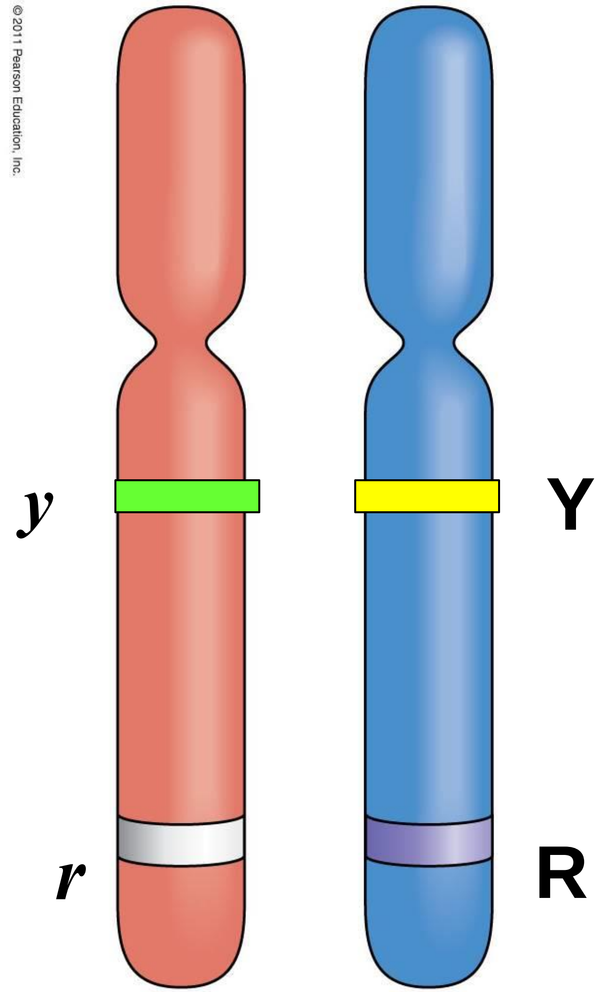
- For example, in pea plants seed **texture** and **colour** are controlled by **two** separate gene loci where:
  - For texture, round (**R**) is dominant to wrinkled (**r**)
    while
  - For **colour** yellow (**Y**) is dominant to green (**y**).

- Mendel started by crossing 2 parental pure-breeding plants: one that is **round** & **yellow** (**RRYY**) seeds and the other with **green** & **wrinkled** (**rryy**) seeds to get F_1. Then he selfed F_1 x F_1 to get F_2 generation.

Pair of **homologous** chromosomes - **DIHYBRID**

Allele
for green color

Allele
for wrinkled texture



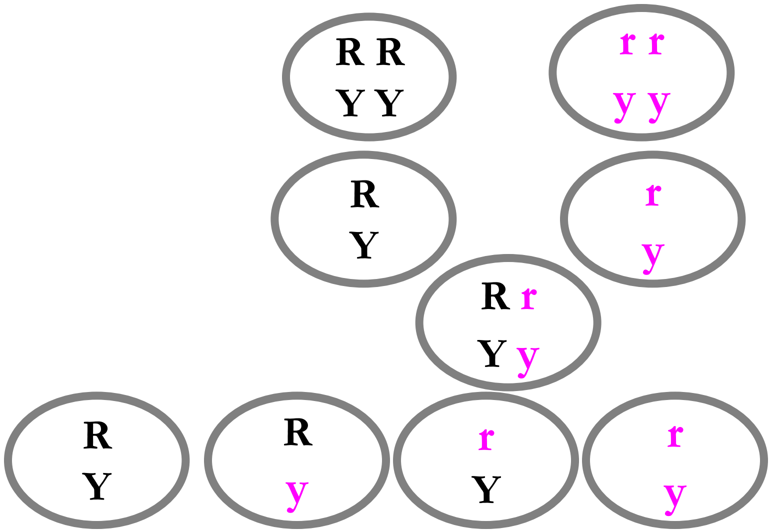
Allele
for Yellow color

Allele
for Smooth texture

Alleles - alternative versions of a gene

Independent assortment

- Alleles at the 2 gene loci segregate (separate) **independently**, and are NOT transmitted as a unit. Therefore, each plant would produce gametes with allele combinations that were not present in the gametes inherited from its parents:



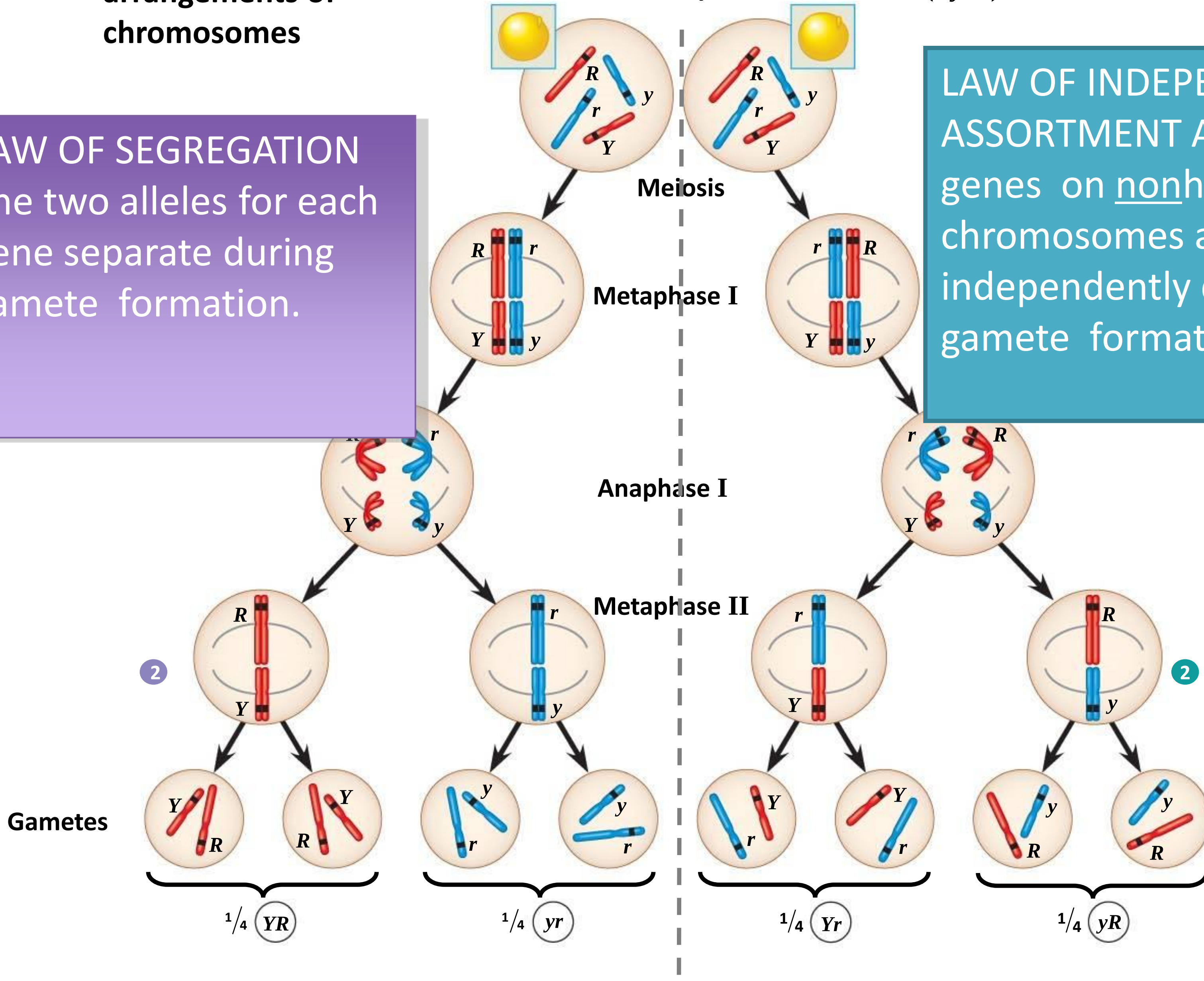
Principle of **Independent Assortment** – Inheritance of one trait has **no effect** on the inheritance of another trait

F_1 Generation: 2 possible arrangements of chromosomes

All F_1 plants produce yellow-round seeds *(YyRr)*

LAW OF SEGREGATION The two alleles for each gene separate during gamete formation.

LAW OF INDEPENDENT ASSORTMENT Alleles of genes on <u>non</u>homologous chromosomes assort independently during gamete formation.



F_2 with **independent** assortment:

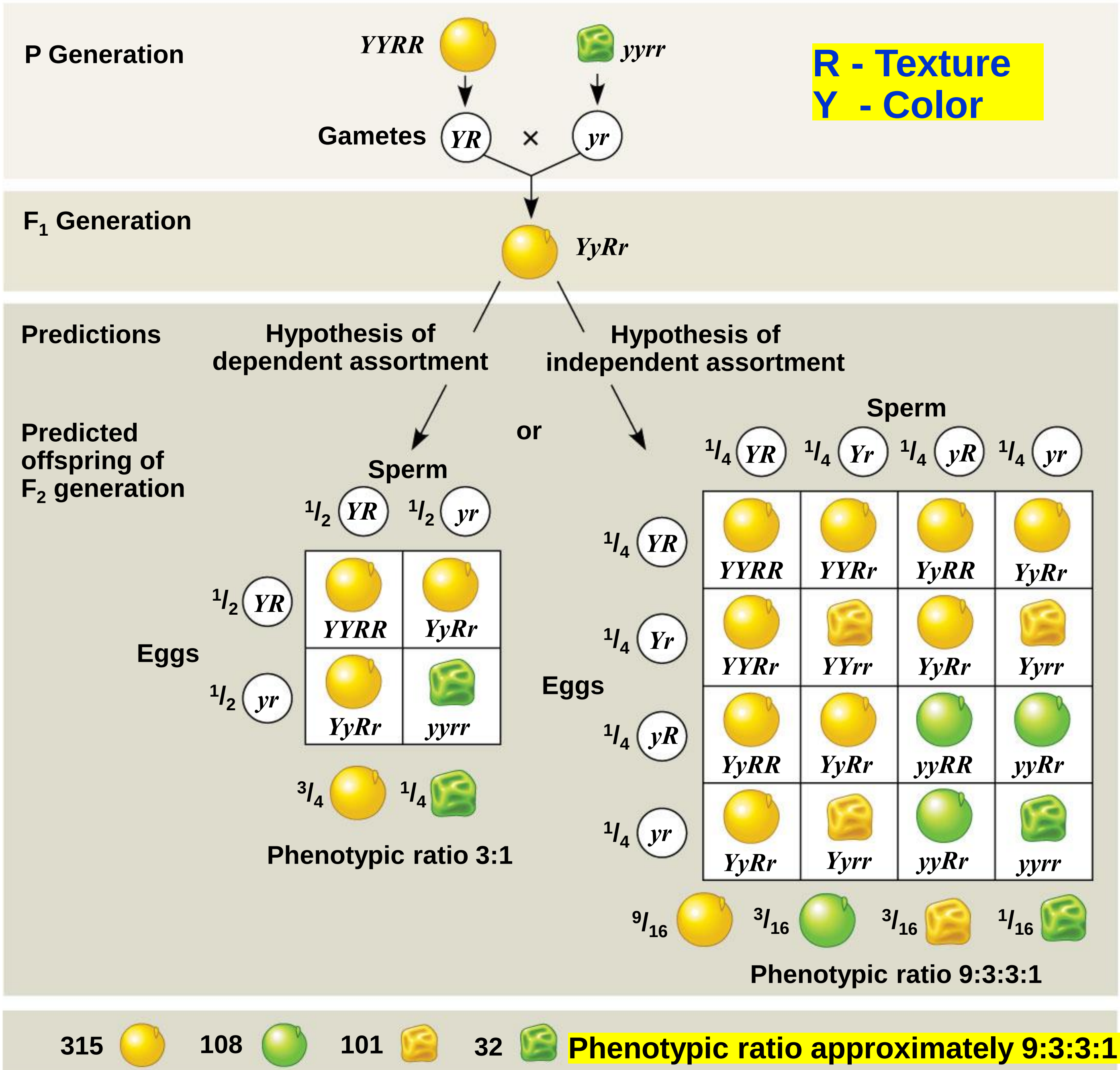
Gametes

	RY	Ry	rY	ry
RY	RR YY	RR Yy	Rr YY	Rr Yy
Ry	RR Yy	RR yy	Rr Yy	Rr yy
rY	Rr YY	Rr Yy	rr YY	rr Yy
ry	Rr Yy	Rr yy	rr Yy	rr yy

F_2 Proegeny

Phenotypic ratio is 9 : 3 : 3 : 1

Dihybrid cross



RESULTS

- Using a dihybrid cross, Mendel developed the **law of independent assortment**

Alleles for two or more *different* traits are distributed to sex cells (& offspring) independently of one another.

in other words,

The allele a gamete receives for one gene does not influence the allele received for another gene

NB:

✓ Strictly speaking, this law applies only to genes on different, nonhomologous chromosomes or those far apart on the same chromosome.

✓ Genes located near each other on the same chromosome **tend to be** inherited together

Exercise:

Use a **Punnet square** to answer the following questions and state the phenotype and genotype **ratios** obtained in the crosses below:

1. Outcome of crossing the F_1 (above) to :
   a) a true breeding round and yellow pea plant?
   b) a true breeding wrinkled and green pea plant?
   c) A round and green pea plant?
   d) A wrinkled and yellow pea plant?

2. What is the outcome of crossing a round and green pea plant with one that is wrinkled and green?