Inheritance pattern of single trait (gene) governed by Mendel’s 1st law or Principle of Segregation established by monohybrid cross

Inheritance pattern of single trait (gene) governed by Mendel's 1st law or Principle of Segregation established by monohybrid cross

Gregor Johann Mendel, an Austrian monk performed a series of experiments with garden pea (*Pisum sativum*) from 1856-1863 in a Monastery in Brno (now in Czech Republic) and published the results of his research work in 1865.

He sent his publications to several libraries and scientists worldwide, including the famous Charles Darwin (who wrote 'On the Origin of Species'), who even did NOT read the articles by Mendel (Refer the book 'Genetics: Genes to Genome').

Moreover, it was not recognized during his time and its importance was not realized until the early of 1900, when three botanists- Carl Correns, Hugo de Vries and Erich von Tschermark independently reached the same conclusion as Mendel reported before.

Mendel's experiment

Mendel did not make any mutant or variant, but collected 34 plant lines of garden pea having different traits. He selected **seven (7) traits** (**each** having contrasting **pair of phenotypes**) where there were true-breeding or pure line strains. These are:

1. Seed shape: Round vs. wrinkle
2. Seed color: Yellow vs. green
3. Flower color: Purple vs. white
4. Pod shape: Inflated vs. pinched/constricted
5. Pod color: Green vs. yellow
6. Flower position: Axial vs. terminal
7. Stem height: Tall/Long vs. short

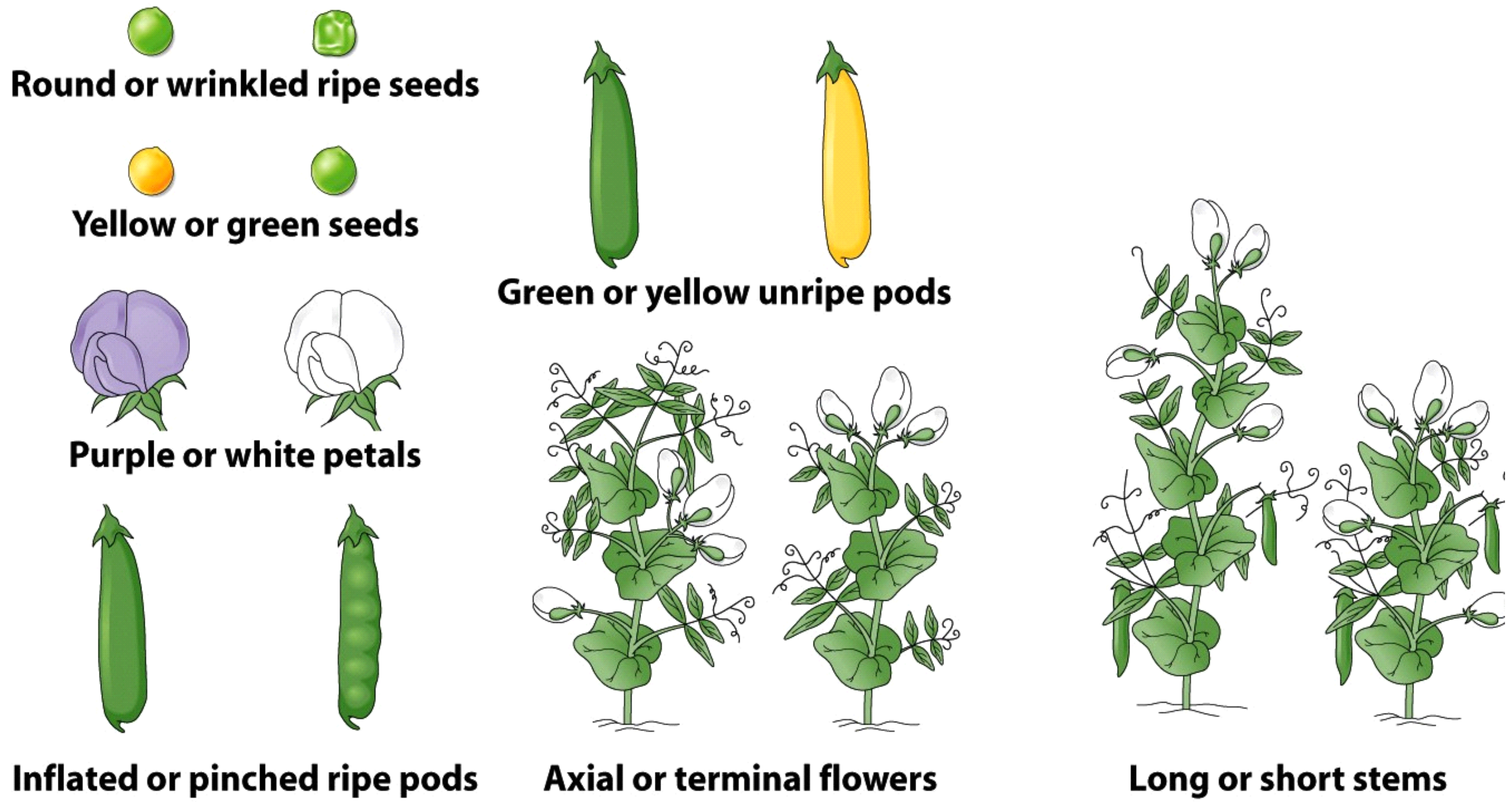

Figure 2-9
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Mendel’s experiment (contd..)

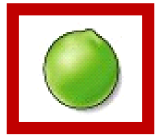
His selection of pea plant and traits were very good choice.

Many scientists during that time studying on inheritance worked with other systems and failed to give any conclusion. Charles Darwin (who proposed Evolution by Natural Selection) was in favor of “Blending Theory”.

Why?:

- Garden pea is a self-pollinated plant, cross-pollination/ hybridization can be performed manually and easily due to relatively larger flower
- There are many varieties (traits & phenotypes)
- It produces many seeds within shot time period (large number of offspring & short generation time)
- Later it was discovered that the 7 genes (for selected 7 traits) were either located in different chromosomes or far apart in the same chromosome (which has crucial implication in operating Mendelian Principles, which we will learn afterwards)

Results of 1st monohybrid cross and its reciprocal cross

 X 

Parental generation (P) | **First filial (F_1) generation**

Female from **round-seeded** line **X** **male** from **wrinkle-seeded** line — all peas **round**

Male from **round-seeded** line **X** **female** from **wrinkle-seeded** line — all peas **round**

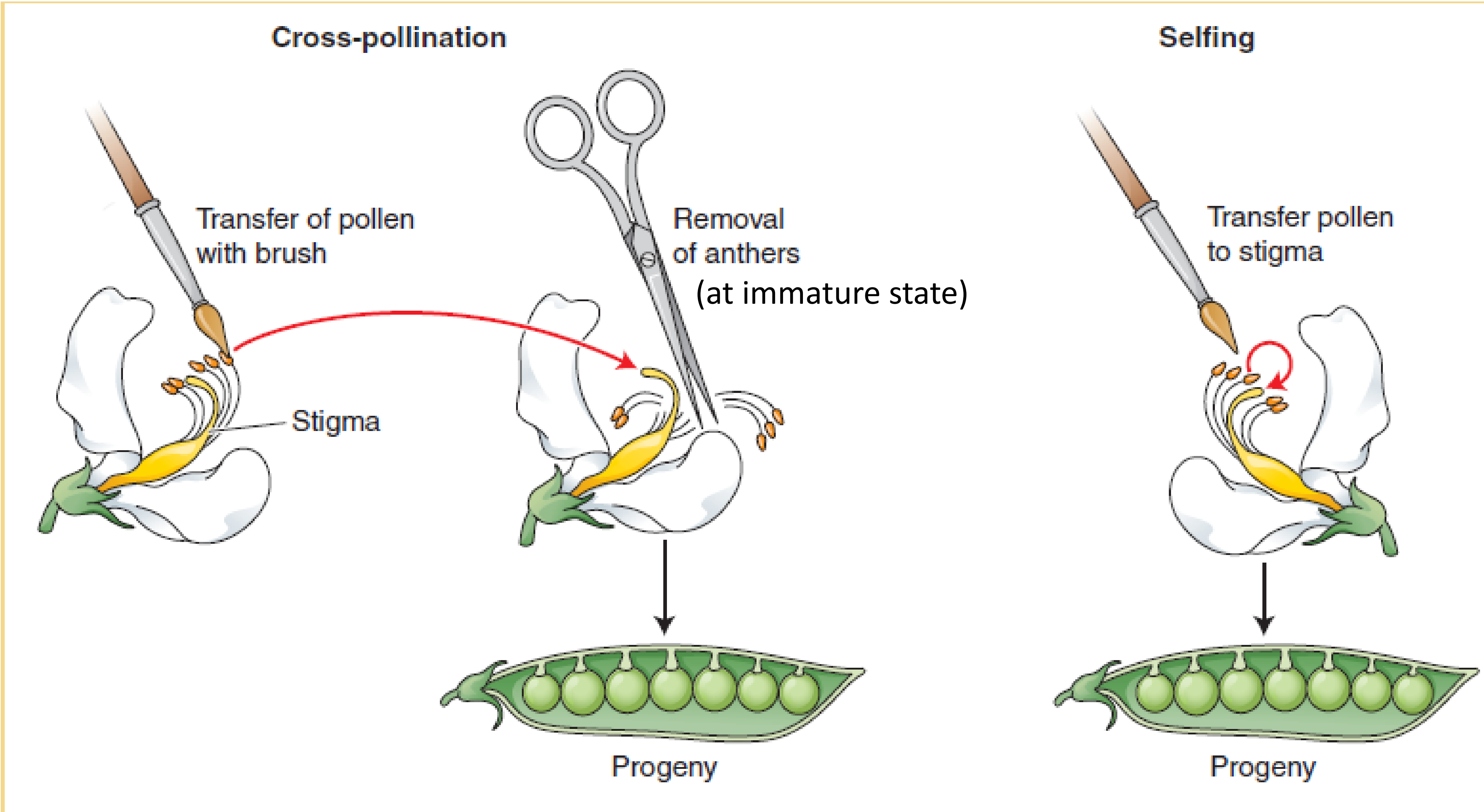
♂ ♀

[Note: Feline = cat-related; Filial = related to children or progeny]

A **monohybrid cross**: deliberate matting between true-breeding or pure lines (e.g., ***RR*** x ***rr***) that are different with respect to the **contrasting phenotype of the single traits** or alternate alleles of a gene.

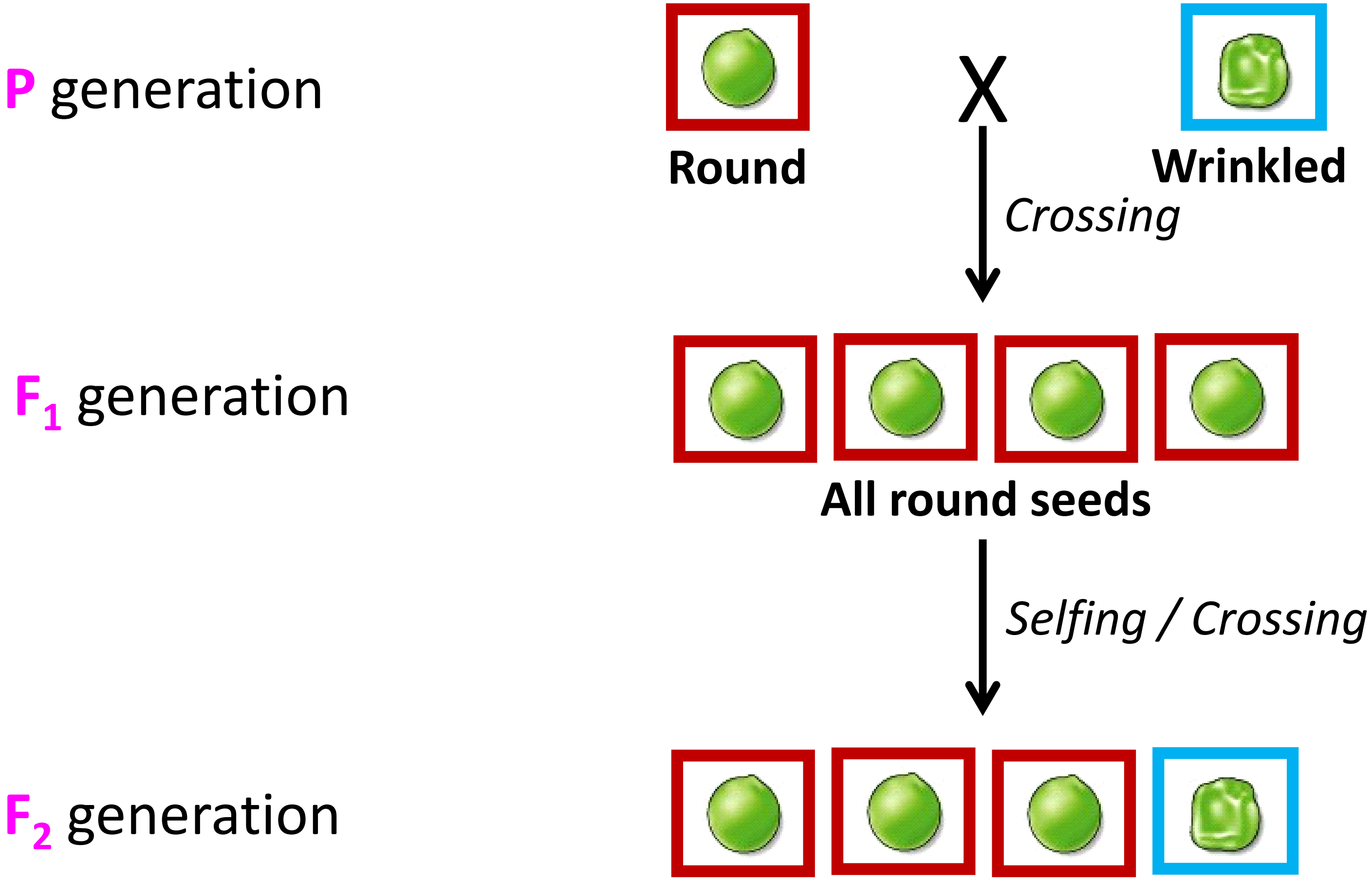
It also refers to a selfing or crossing between two individuals which are **heterozygous for a single gene** (or having two different alleles in the same locus of the homologous chromosome), e.g., ***Rr*** x ***Rr***)

Explain **emasculation**: Pea is a self-pollinated plant. To achieve cross-pollination, Mendel removed immature stamens



Introduction to Genetic Analysis by Griffiths et al. 11th Ed.

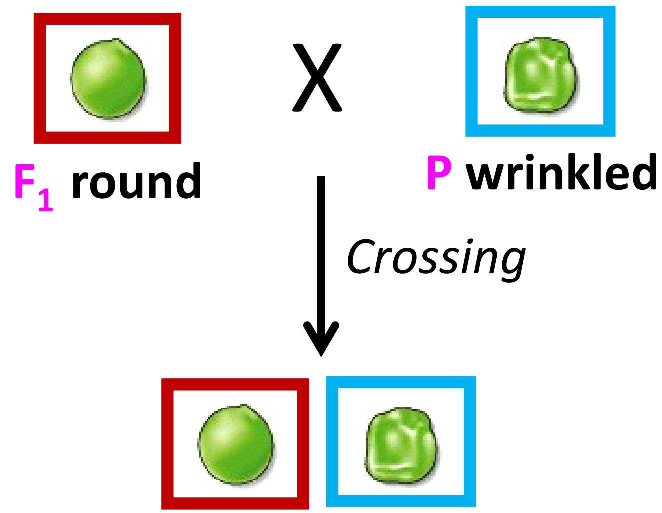
Results/ Phenotypes of Mendel's 1st monohybrid cross



Approx. ¾th round seeds and ¼th wrinkle seeds

3:1 ratio of round: wrinkle seeded plants

Phenotypes of Mendel's 1st test cross

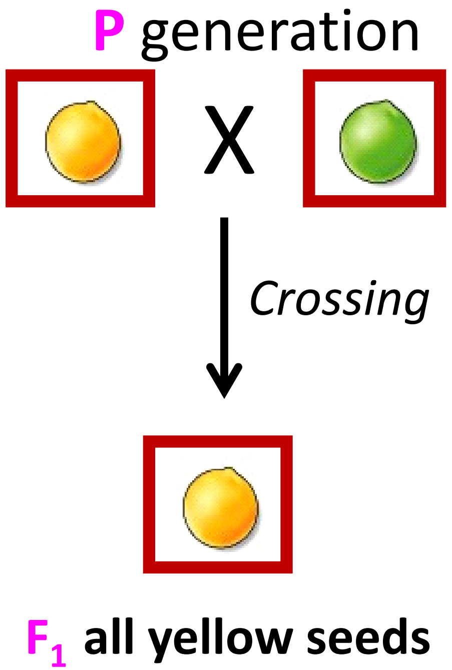


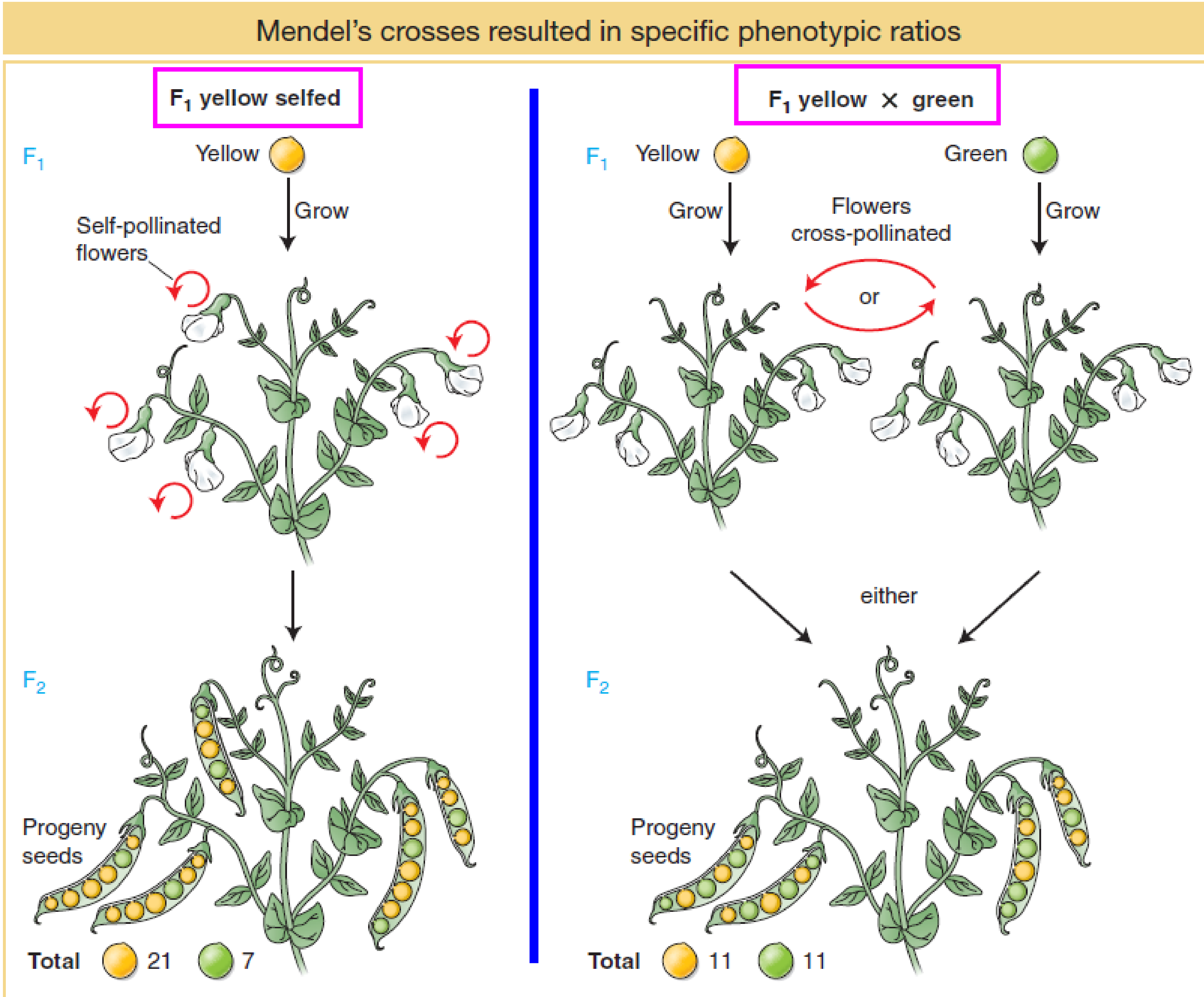
½ round seeds and ½ wrinkle seeds

1:1 ratio of round: wrinkle seeded plants

- Interestingly, the wrinkle-seeded phenotype disappeared in F_1, but re-appeared in F_2
- Moreover, all the round-seeded plants of F_2, when selfed produced offspring of different types, but wrinkle-seeded F_2 plants when selfed produced only wrinkle seeds just like pure line.
- **These phenotypic ratios, i.e. 3:1 in F_2 progeny of monohybrid cross and 1:1 in progeny of monohybrid test cross are characteristic of the single gene inheritance and these are found in all the seven traits that Mendel studied in pea plant.**

Results/ Phenotypes of 2nd monohybrid cross and test cross





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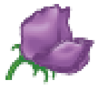
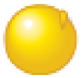
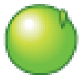
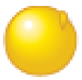
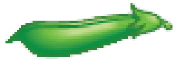
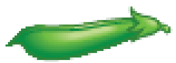
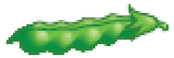
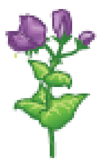
Results of Mendel's all crosses in which parents differed in contrasting phenotypes of one trait

Table 14.1 The Results of Mendel's F_1 Crosses for Seven Characters in Pea Plants

Character	Dominant Trait	×	Recessive Trait	F_2 Generation Dominant: Recessive	Ratio
Flower color	Purple 	×	White 	705:224	3.15:1
Seed color	Yellow 	×	Green 	6,022:2,001	3.01:1
Seed shape	Round 	×	Wrinkled 	5,474:1,850	2.96:1
Pod color	Green 	×	Yellow 	428:152	2.82:1
Pod shape	Inflated 	×	Constricted 	882:299	2.95:1
Flower position	Axial 	×	Terminal 	651:207	3.14:1
Stem length	Tall 	×	Dwarf 	787:277	2.84:1

Biology: A Global Approach by Campbell et al. (11th Ed.)

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

Table 2-1
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Mendel made three important conclusions from the observation data and the analysis of monohybrid cross

1) The contrasting phenotypes of a trait are determined by **particulate factors** (now known as the alleles of the gene) which are transmitted from parents to progeny.

2) The F_1 individual inherits one factor for each trait (i.e., one allele of the allelic pair for a gene) from each parent through gamete, and thus F_1 individual has two factors for each trait. [Recall that diploid parents have two particulate factors (for each trait) located in homologous chromosomes]

3) One of the pair of factors (one allele of the pair) may not show up its phenotype (i.e., remains masked) in F_1, but it does not get blended or mixed with the other member of the factor pair. On the other hand, **the members of the factor pair segregate from each other during gamete formation, and the phenotype reappears in the next generation (F_2)**. [Here comes dominant & recessive phenotypes/factors or alleles].

This is known as **Mendel's law or principle of segregation** or first law of Mendelian inheritance.

Mendel's Explanation

Monohybrid cross between pure Yellow-seeded X pure Green-seeded & Test cross between F_1 yellow X pure green

In the context of modern terms and concept of chromosomal location of genes, the **phenotypic ratio** and **genotypic ratio** of a **monohybrid cross can be explained using Punnett square** as shown in the diagram.

[Note: R.C. Punnett is British mathematician & geneticist, who discovered also genetic linkage using chicken and sweet pea (*Lathyrus odoratus*) systems along with William Bateson]

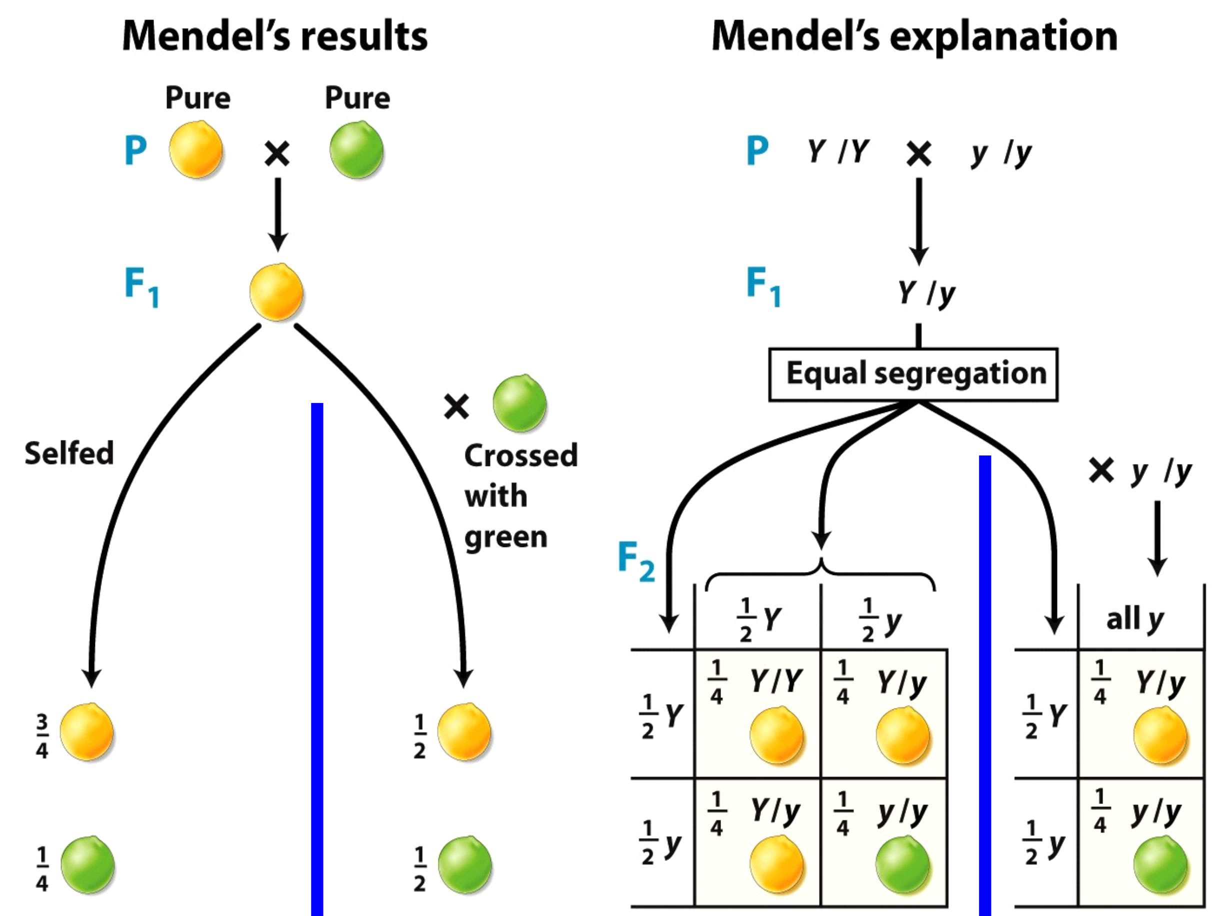

Figure 2-12
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Phenotypic **3:1** | **1:1**

Genotypic: **1:2:1** | **1:1**

Show gametes in circle

In Test cross, show as ½ **y** & ½ **y;** Then, 2/4 **Yy** & 2/4 **yy**

Mendel's first law of inheritance or Principle of Segregation

In heredity, two members of the particulate factors (= two alleles of the gene) controlling the particular/specific trait segregate from each other during gamete formation (in meiosis), half of the gametes carry one factor (=one allele) and the other half carry the second factor (=another allele).

Up to this slide, Lecture # 5 delivered on 29.08.2023

Inheritance pattern of two (or more) traits (genes) governed by Mendel's 2nd law or Principle of Independent Assortment established by dihybrid (or multiple-hybrid) cross

Before starting the 2nd law:

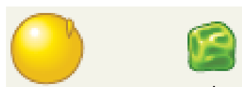
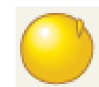
(1) Recapitulate the concept of 1st law,

(2) Ask students how to write Mendel's observations/results on monohybrid cross of Tall **X** Dwarf plants & how to explain the findings through Punnett square

Mendel’s dihybrid cross experiment

Once he established the inheritance pattern of the single trait or character (controlled by single gene having two alleles) using the pure lines differing with respect to two contrasting phenotypes; **Mendel started working with the pure lines that were different with respect to the pair of traits each with two contrasting phenotypes** from the combination of seven different traits initially he selected in pea plants.

Results of the dihybrid cross and its reciprocal cross:

Parental generation (P)  **First filial (F_1) generation** 

Round, yellow-seeded **female** X wrinkled, green-seeded **male** — **all peas round, yellow**

Round, yellow-seeded **male** X wrinkled, green-seeded **female** — **all peas round, yellow**

♂ ♀

Dihybrid cross

Deliberate matting between true-breeding or pure lines (e.g., ***RRYY*** X ***rryy***) that are different with respect to the contrasting phenotypes of the two traits or alternate alleles of the two genes.

It also refers to the selfing or a cross between two individuals (e.g., ***RrYy*** X ***RrYy***) which are heterozygous for two genes (or having two sets of different alleles in the two loci that are located in two different sets of homologous chromosomes or situated far apart in the same set of the homologous chromosomes)

Discuss: Now, we know where the Mendel's 7 particulate factors/genes are located on different chromosomes in pea plant. Chromosome carries the **particulate factor/gene/allele** corresponding to the **trait/phenotype**.

In chromosome #1 : 2 particulate factors/genes/alleles

In chromosome #4 : 3 particulate factors/genes/alleles

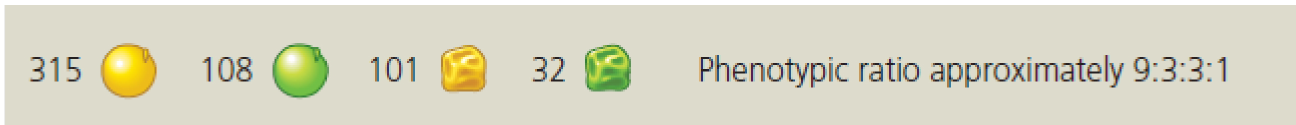
In chromosome #5 : 1 particulate factor/gene/allele

In chromosome #7 : 1 particulate factor/gene/allele

Results of selfing of the plants from the first filial (F_1) generation

Progeny of the second filial (F_2) generation

Phenotype of the seeds	total number	approx. proportion
round, **yellow** seeds	315	9/16
round green seeds	108	3/16
wrinkled **yellow** seeds	101	3/16
wrinkled green seeds	32	1/16



Interestingly, just like the results of monohybrid cross, the wrinkled or the green-seeded phenotype that **disappeared** in F_1 **re-appeared** in F_2 .

However, the F_2 phenotypic ratio 9:3:3:1 of dihybrid cross is **more complex** compared to the 3:1 ratio of the monohybrid cross.

Although, initially it seems complicated, but **careful analysis revealed** that **with respect to <u>one trait</u>** (or corresponding two contrasting phenotypes) **at a time, <u>the F_2 phenotypic ratio is close to 3:1</u>**; i.e. **for seed shape-** round (315+108): wrinkled (101+32) is **~ 3:1**; and for **seed color-** yellow (315+101): green (108+31) is also **~ 3:1**.

Phenotype of the seeds	total number	approx proportion
round, yellow seeds	315	9/16
round green seeds	108	3/16
wrinkled yellow seeds	101	3/16
wrinkled green seeds	32	1/16

Mendel realized that **the ratio 9:3:3:1 is actually random combination of two different 3:1 ratios**, i.e. multiplication of two separate 3:1 ratios, one for seed shape and another one for seed color.

(3:1) **x** (3:1) = 9:3:3:1

With respect to proportion, calculation looks like:

(¾ : ¼) **x** (¾ : ¼) = 9/16 : 3/16: 3/16: 1/16

Mendel reasoned or explained that it was **possible only when the two particulate factors for each trait** (e.g., seed shape) **not only separate from each other following Principle of Segregation but they do so independently of the factor-pair for the other trait** (i.e., seed color) and **combine or assort randomly** to produce new combination, in addition to the parental combination of phenotypes.

Using modern terms of genes and alleles controlling the traits and phenotypes, respectively and the concept of chromosomal location of genes;

a dihybrid cross between round, yellow-seeded and wrinkled, green-seeded pure lines of pea plant can be explained and the progeny genotypes can be assigned using **Punnett square**.

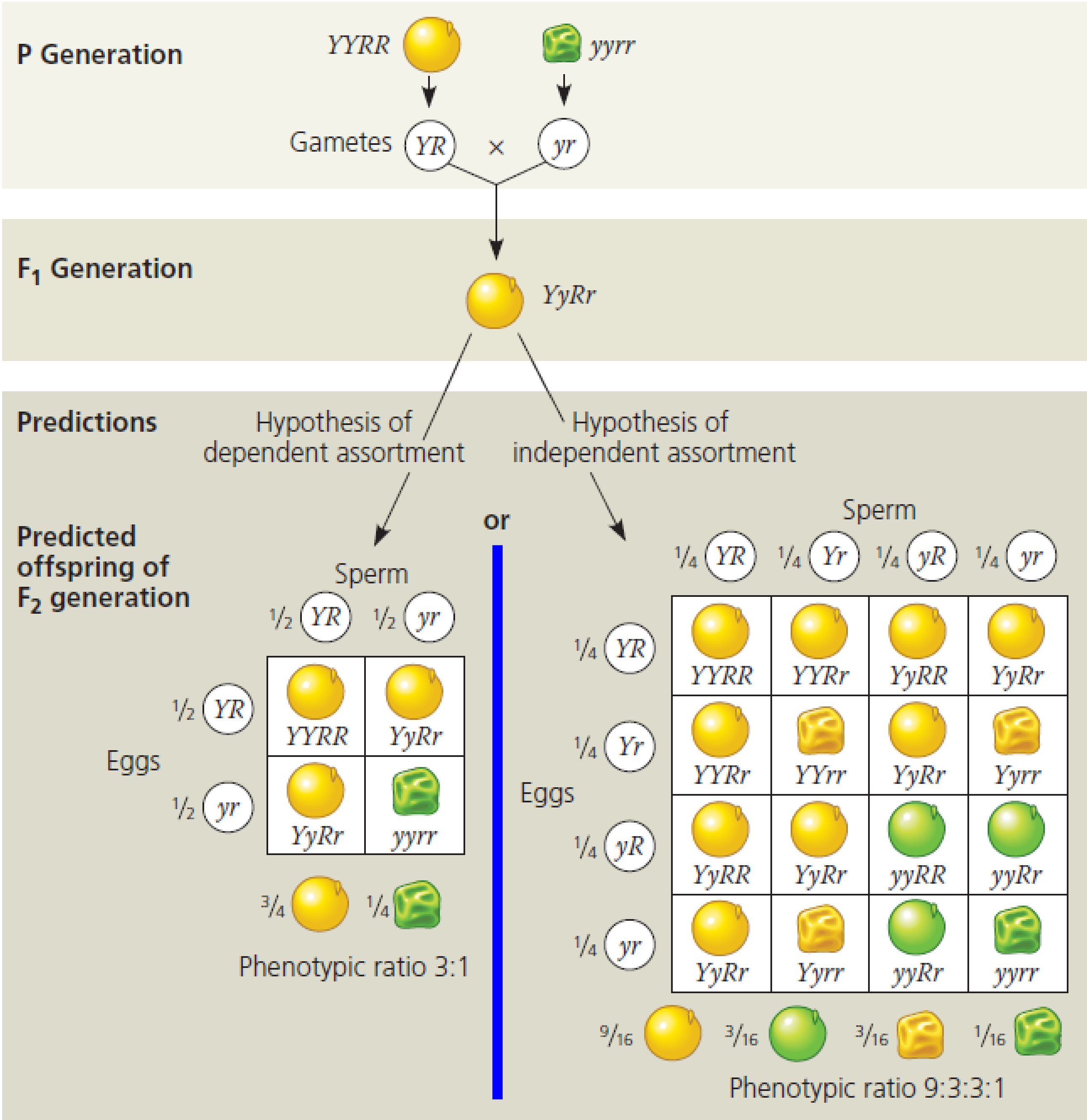
[Location of different genes in different chromosomes or in loci far apart in the same chromosome are indicated by semicolon **;**]

Symbol of chromosome here: **/**

Symbol of homologous chromosomes here: **//**

However, most of the times these symbols are not used at all.]

Mendel's Explanation



YYRR X ***yyrr***
meaning precisely
Y//Y; R//R X ***y//y; r//r***

Hypothesis of <u>dependent</u> assortment

Hypothesis of <u>Independent</u> assortment

Biology: A Global Approach by Campbell et al. (11th Ed.)

Dihybrid Test cross

To further verify his hypothesis of independent assortment, Mendel performed **crosses of F_1 dihybrid individuals (from round, yellow seeds) with the original parental pure line of recessive traits i.e., wrinkled, green-seeded plants**; just what he did for monohybrid test cross. This time he obtained the progeny as follows:

Phenotype	approx. proportion
round, **yellow** seeds	¼ th
round green seeds	¼ th
wrinkled **yellow** seeds	¼ th
wrinkled green seeds	¼ th

This progeny phenotypic ratio 1:1:1:1 (combination of four phenotypes) from dihybrid test cross is similar (i.e., 1:1 of two phenotypes) to that of monohybrid test cross.

Mendel also explained this on the basis of multiplication of two separate ratios (1:1) or proportions (½ : ½) of monohybrid test cross.

(1:1) x (1:1) = 1:1:1:1

(½ : ½) x (½ : ½) = ¼ : ¼ : ¼ : ¼

Discuss **dihybrid test cross** using Punnett square

Thus, the phenotypic ratios- 9:3:3:1 in F_2 progeny of **dihybrid cross** and 1:1:1:1 in progeny of **dihybrid test cross** are **characteristics of the inheritance pattern of two genes**, and these were found in dihybrid with any combination of two traits out of the **seven traits that Mendel studied in pea plant**.

Do you see any differences (phenotypes & genotypes) in the parental lines, F_1 and F_2 progenies between the left & right panels ?

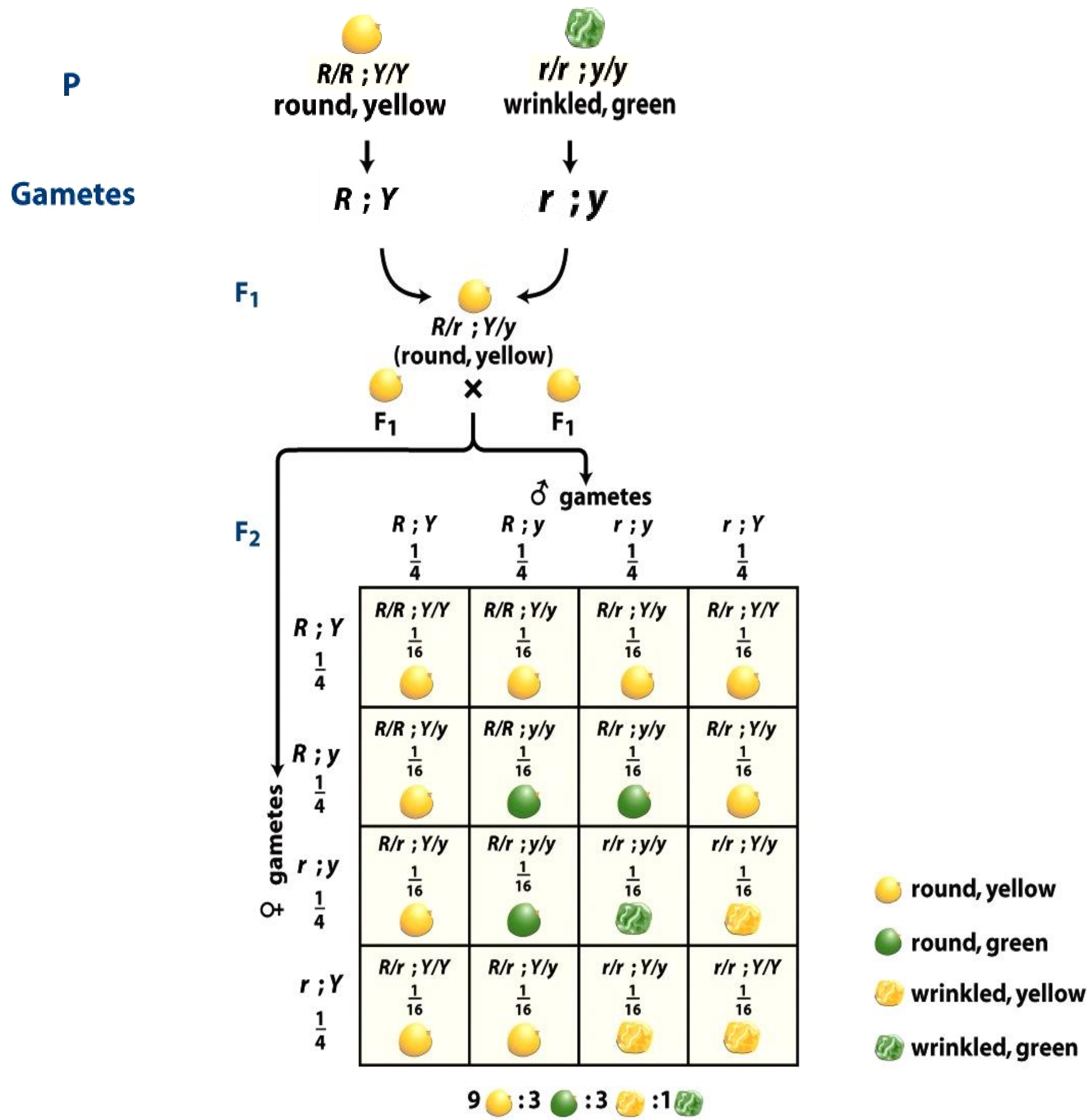


Figure 3-4
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Modified by MKM

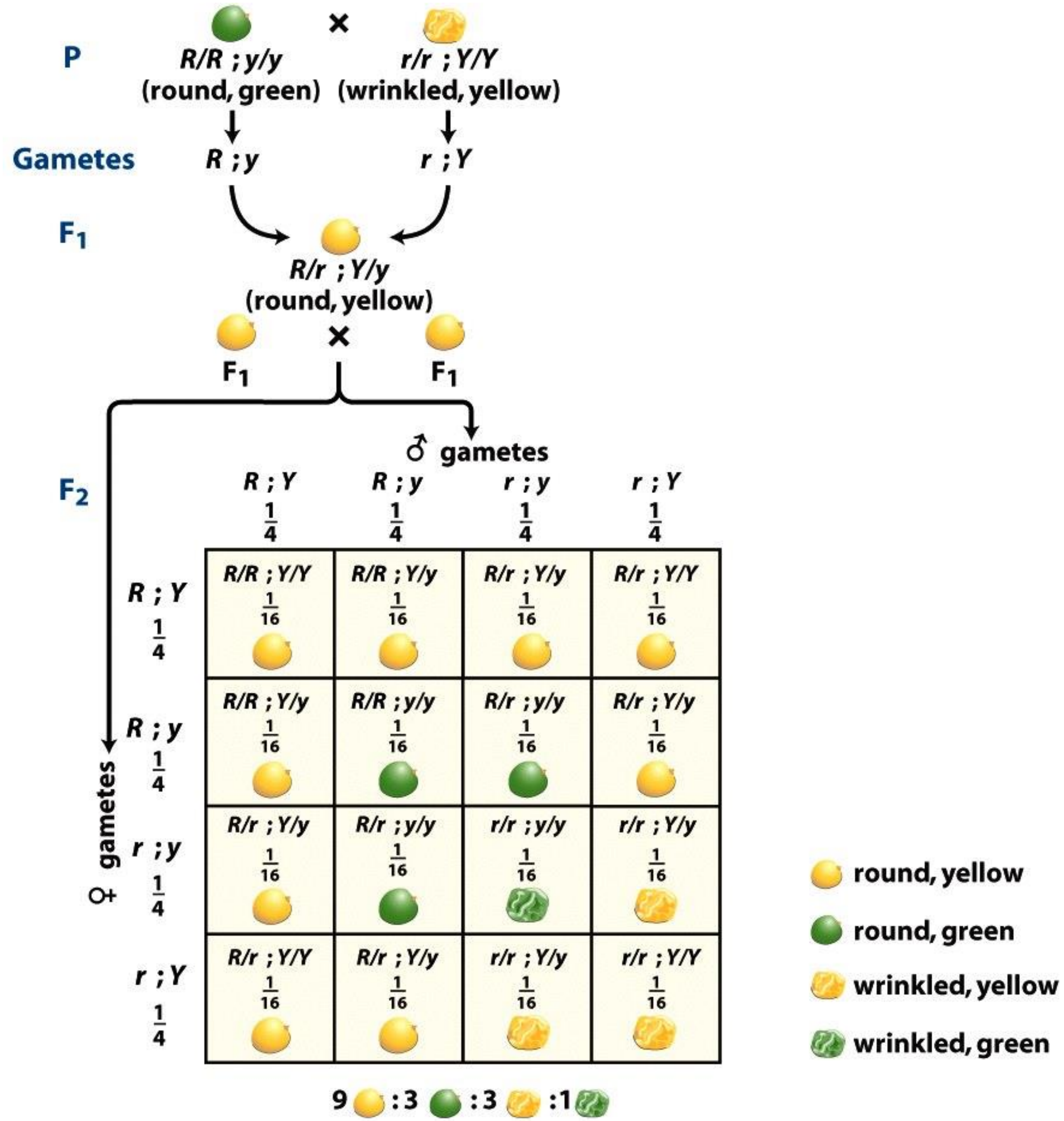

Figure 3-4
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Mendel's 2nd Law of Inheritance or Principle of Independent Assortment

In heredity, the individual members of the **different particulate factor pairs** (= each allele of the different genes) controlling the different traits **not only segregate but also assort independently** of each other during gamete formation, **thus bringing new combination of genotypes and phenotypes of the progeny** in addition to the parental combinations.

Thus, the 2nd Law encompasses inherently the concept of 1st Law.

Ask the student: notice this point very carefully. Fundamentally, the Law of Segregation and Law of Independent Assortment are pointing out the segregation of allelic pair, and their independent assortments when more than one allelic pairs.

Trihybrid cross

Mendel also conducted genetic analysis using parental lines that are different with respect to three traits or three pairs of contrasting phenotypes to confirm his laws of inheritance.

He used two parental lines:

one giving **round**, **yellow seeds** and **purple flowers**; and

the other producing **wrinkled, green seeds** and **white flowers**.

As usual the F_1 trihybrid progeny showed only one set of phenotype, i.e. round, yellow seeds and purple flowering plants,

while in the F_2 progeny he found **different combination of total 8 phenotypic classes in a ratio of 27:9:9:9:3:3:3:1**.

Similar results were also obtained with other combination of phenotypes in pea trihybrid crosses.

Trihybrid cross

Parental type: ***R//R*; *Y//Y*; *P//P*** [***RRYYPP***] **X** ***r//r*; *y//y*; *p//p*** [***rryypp***]

(round, yellow seeds, purple flowers) (wrinkled, green seeds, white flowers)

F_1: ***R//r*; *Y//y*; *P//p*** **[*RrYyPp*]**

(round, yellow seeds, purple flowers)

Now, for F_2 progeny development:

Ask the students to draw **Punnett square** with 8 gametes in row and same 8 gametes in column to find the genotypes and phenotypes of F_2 progeny

Note that symbol '/' is not written in gametes; but gamete should be in circle:

1/8(***RYP***) 1/8(***RYp***) 1/8(***RyP***) 1/8(***Ryp***) 1/8(***rYP***) 1/8(***rYp***) 1/8(***ryP***) 1/8(***ryp***)

Up to this slide, Lecture # 6 delivered on 29.08.2023