Chapter 2

Mendel's Principles of Heredity

Sections to study

2.1 Background: The historical puzzle of inheritance

2.2 Mendel's approach to genetic analysis

2.3 Mendelian inheritance in humans.

2.1 Background: The historical puzzle of inheritance

- ***Artificial selection*: Purposeful control over mating by choice of parents for the next generation.**
- **An important practice since before recorded history.**
  - **Selective breeding of plants**
  - **Domestication of animals**

The 2800-yr old Assyrian relief

Fig. 2.4

- By 19th century, plant and animal breeders had created many strains in which offspring often carried a prized trait.
- Breeders could not explain why a valued trait would sometimes disappear and then reappear in only some offspring.

Three basic questions of genetics proposed by Abbot Napp in 1837

1. What is inherited?
2. How is it inherited?
3. What is the role of chance in heredity?

Historical theories of inheritance

1. One parent contributes most features (Nicolaas Hartsoeker in 1694).
2. **Blended inheritance** – parental traits become mixed and forever changed in offspring.

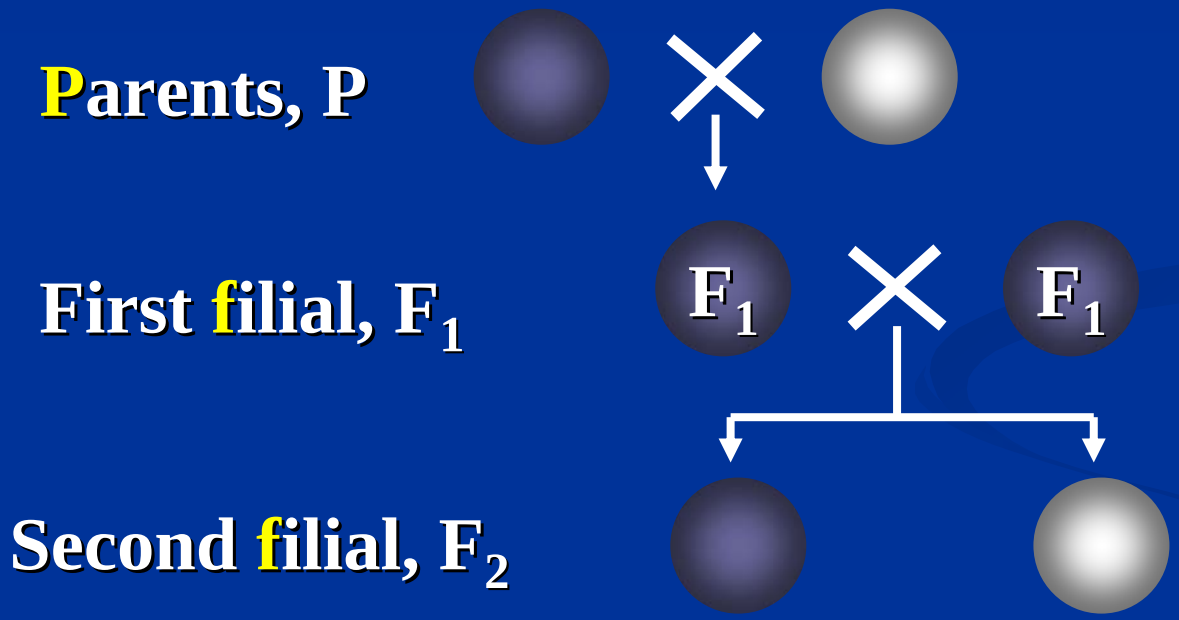
The homunculus

Fig.2.6

Plant hybrid experiments before Mendel

1. Thomas Andrew Knight (1797, Britain)

Crossbreed gray garden pea with white garden pea



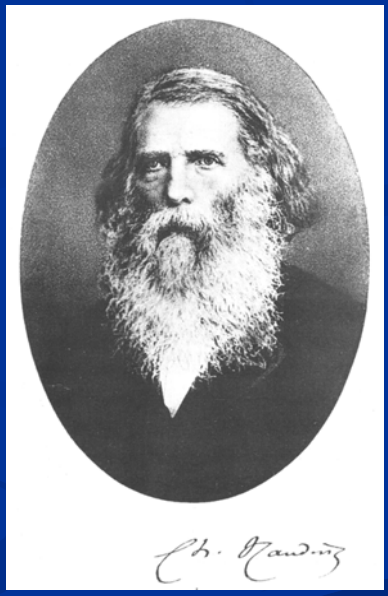

(a) *Pisum sativum*

But he did not calculate the number!

2. Charles Victor Naudin (France, his work btn 1854-1865)

His main publication is *Mémoire sur les hybrides du règne végétal* which appeared in *Recueil des savants étrangers* and won him the Grand Prize of the Institute of Botany in 1862.

- **Reciprocal crosses** with the same results
- The **'specific essence'** controls the traits.
- Factors **must go to different gametes** during gamete formation.



2.2 Gregor Mendel's approach to genetic analysis

- **Discoverer of general principles of heredity.**
- **Performed plant hybrid experiments to see whether there is a "generally applicable law governing the formation and development of hybrids".**
- **In 1866, he published a paper "*Experiments on Plant Hybrids*".**

Gregor Mendel (1822-1884)

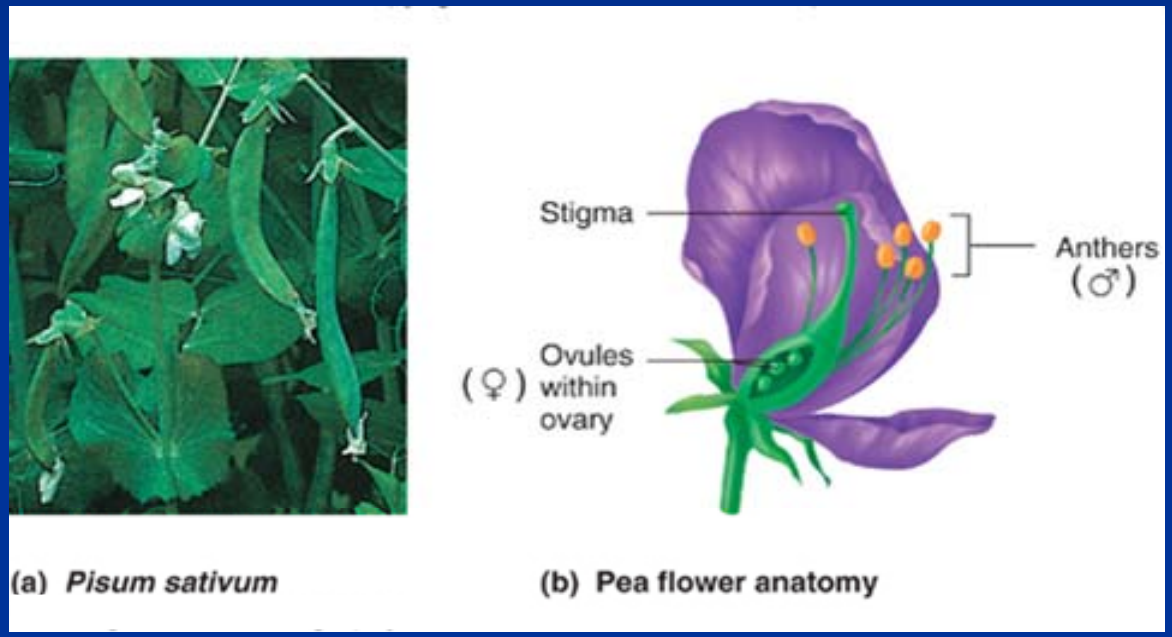
Fig. 2.2

Mendel's workplace

Fig. 2.5

Keys to Mendel's success

1. He chose an ideal experimental organism, the garden pea.



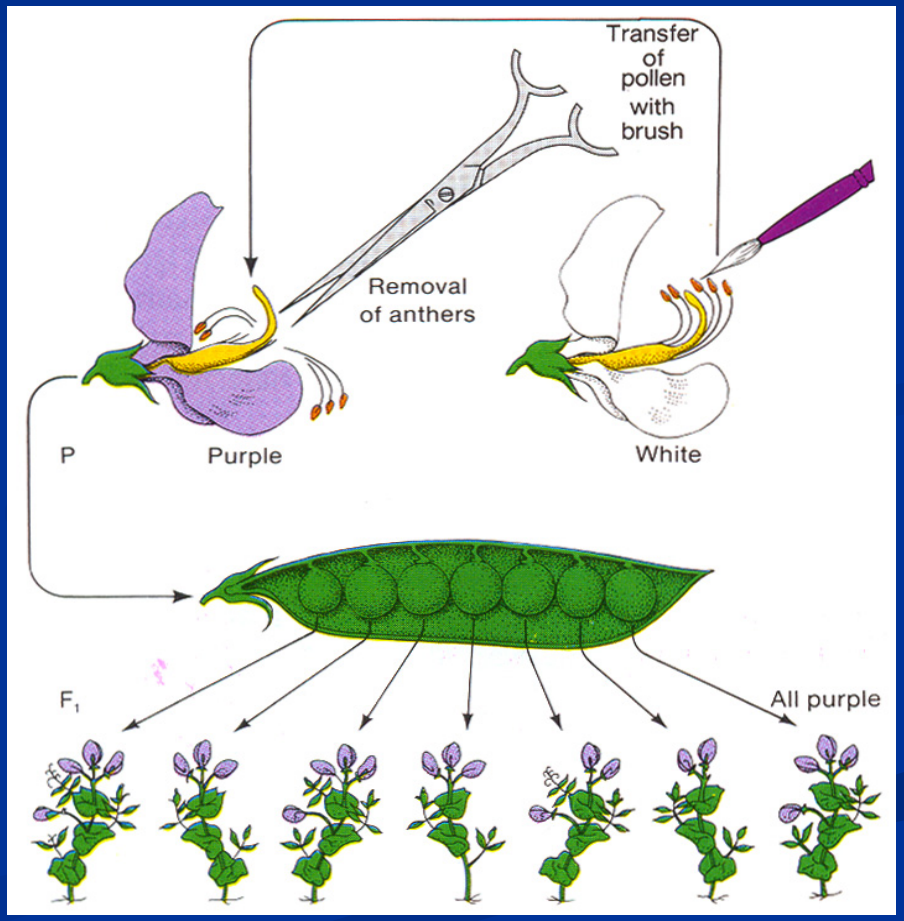


Fig. 2.7

2. He examined the inheritance of traits with discrete alternative forms.

Discrete trait: Trait that exhibits a clear either/or status (*e.g.* purple versus white flowers).

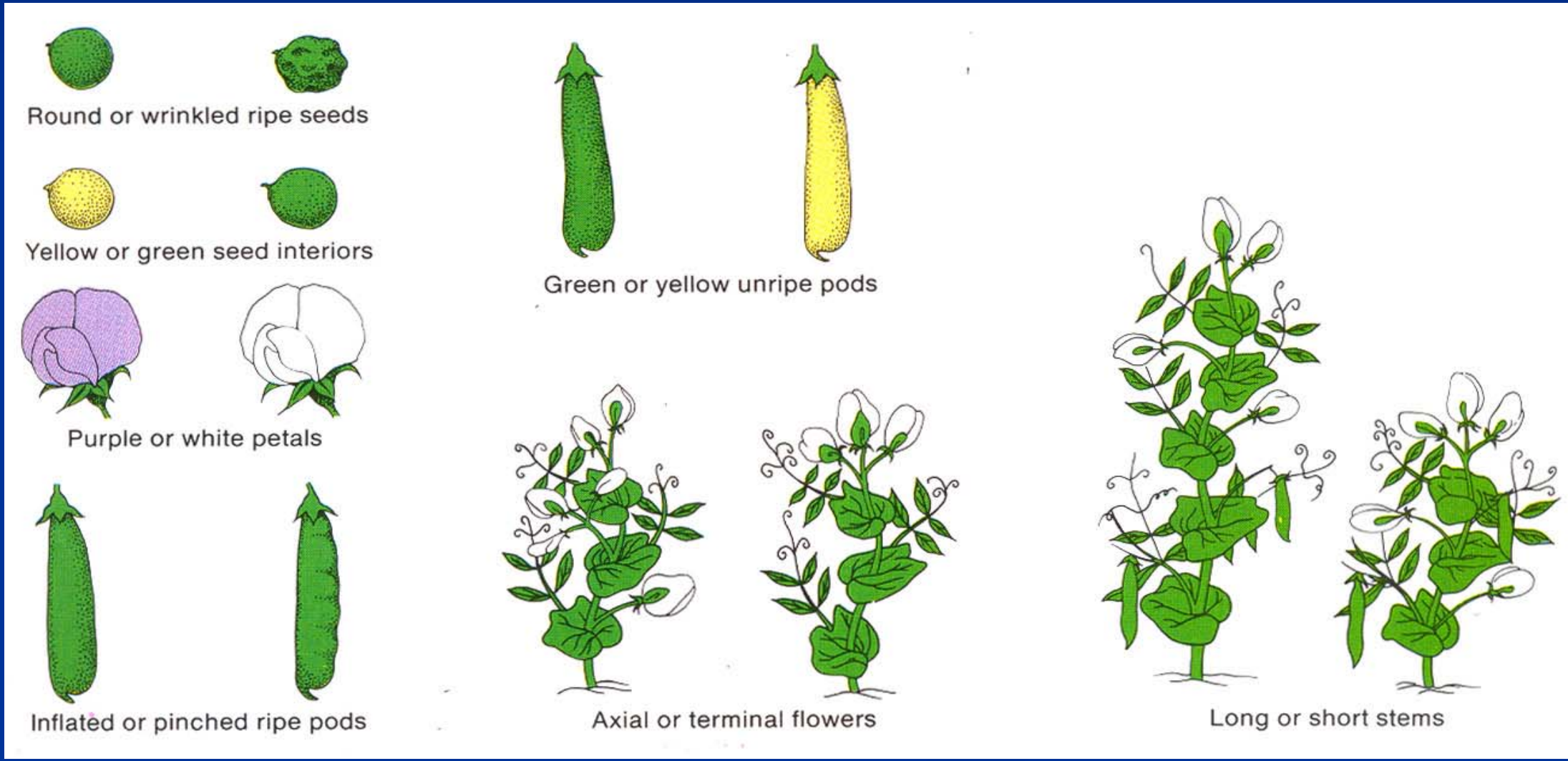


Fig. 2.8

3. He established **pure-breeding lines** to conduct his experiments.

Pure-breeding lines: Families of organisms that produce offspring with specific parental traits that remain constant from generation to generation.

4. He carefully controlled his matings to ensure that the progeny he observed really resulted from the specific fertilizations he intended.

- **He worked with large numbers of plants, counted all offspring, subjected his findings to quantitative analysis, and compared his results with predictions.**
- **He was a brilliant practical experimentalist.**

Monohybrid crosses reveal units of inheritance and law of segregation

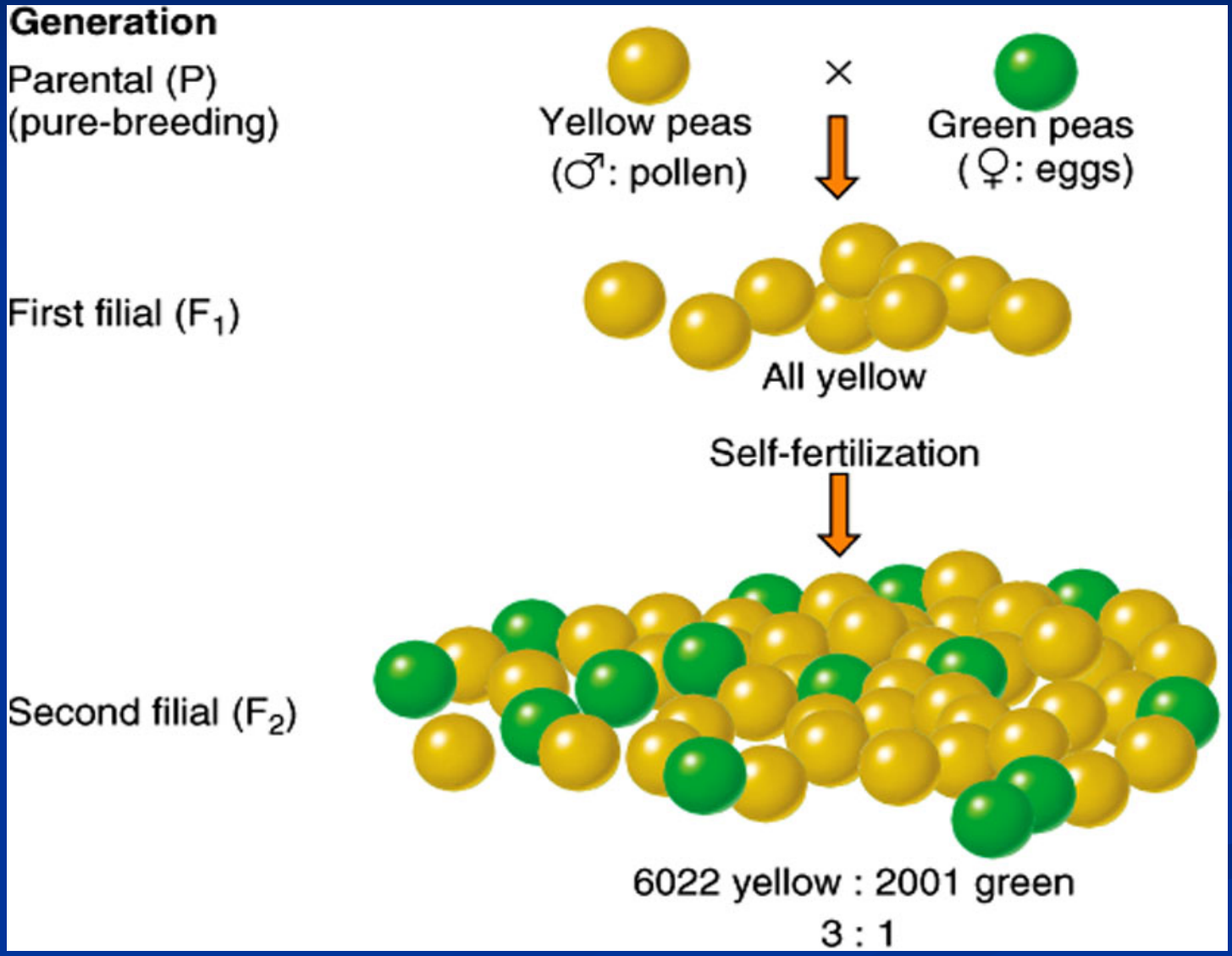
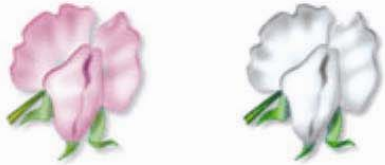


Fig. 2.9

Character	Contrasting traits		F_1 results	F_2 results	F_2 ratio
Seed shape	round/wrinkled		all round	5474 round 1850 wrinkled	2.96:1
Seed color	yellow/green		all yellow	6022 yellow 2001 green	3.01:1
Pod shape	full/constricted		all full	882 full 299 constricted	2.95:1
Pod color	green/yellow		all green	428 green 152 yellow	2.82:1
Flower color	violet/white		all violet	705 violet 224 white	3.15:1
Flower position	axial/terminal		all axial	651 axial 207 terminal	3.14:1
Stem height	tall/dwarf		all tall	787 tall 277 dwarf	2.84:1

- **The F1 yellow peas consist of two types. One breeds true, the other can produce some green peas.**
- **One form must be hidden when plants with each trait are interbred.**
  - **Trait that appears in F1 is *dominant*.**
  - **Trait that is hidden in F1 is *recessive*.**
- **Disappearance of traits in F1 generation and reappearance in the F2 generation disproves the hypothesis that traits blend.**

To explain his observations, Mendel proposed:

- Each unit of inheritance comes in two alternative forms, ***alleles***.
- For each trait, every plant carries two copies of a unit of inheritance, one inherited from the mother and the other from the father.

Alleles are alternative forms of a single gene.

Mendel's first law: Law of segregation

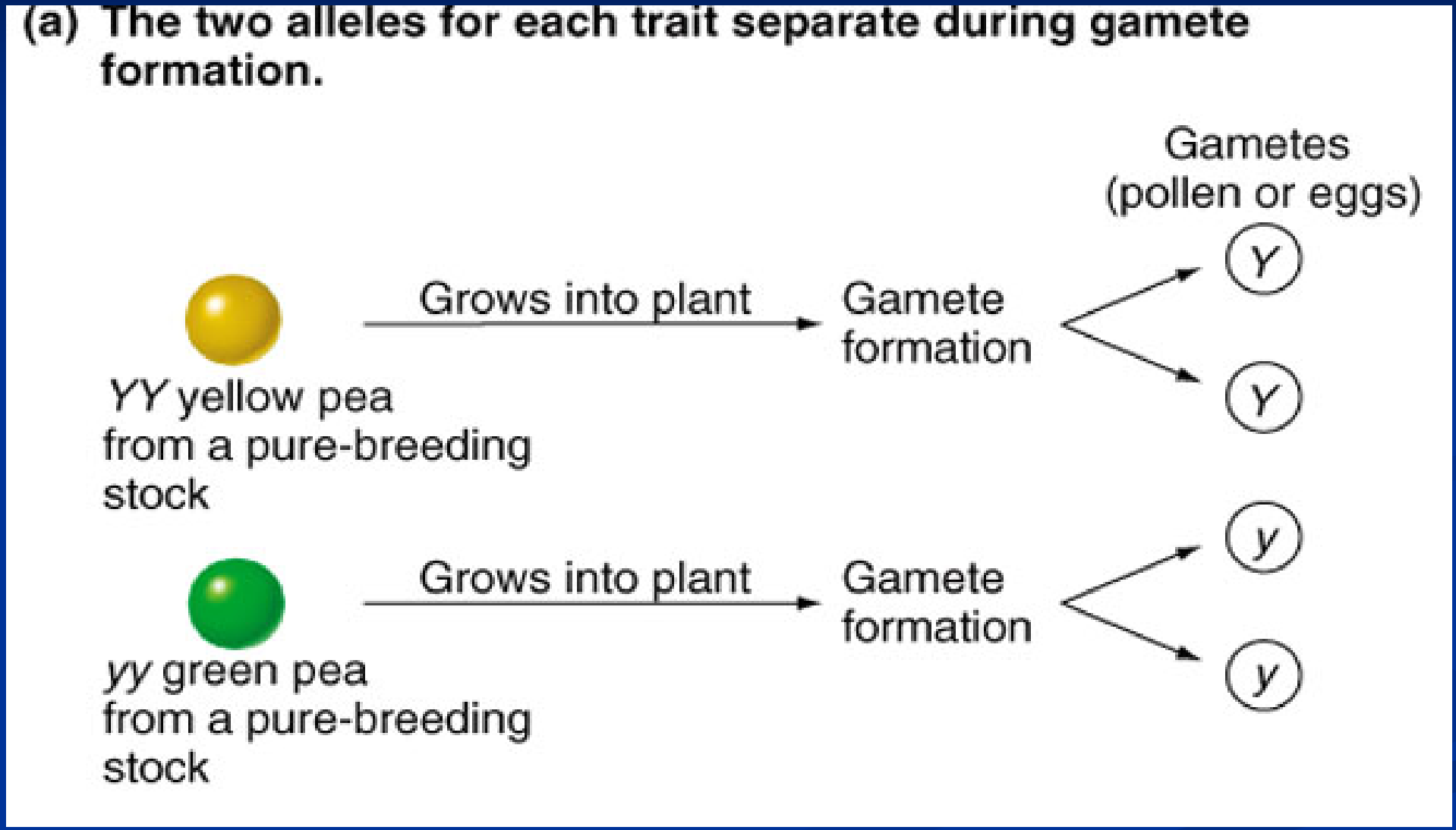
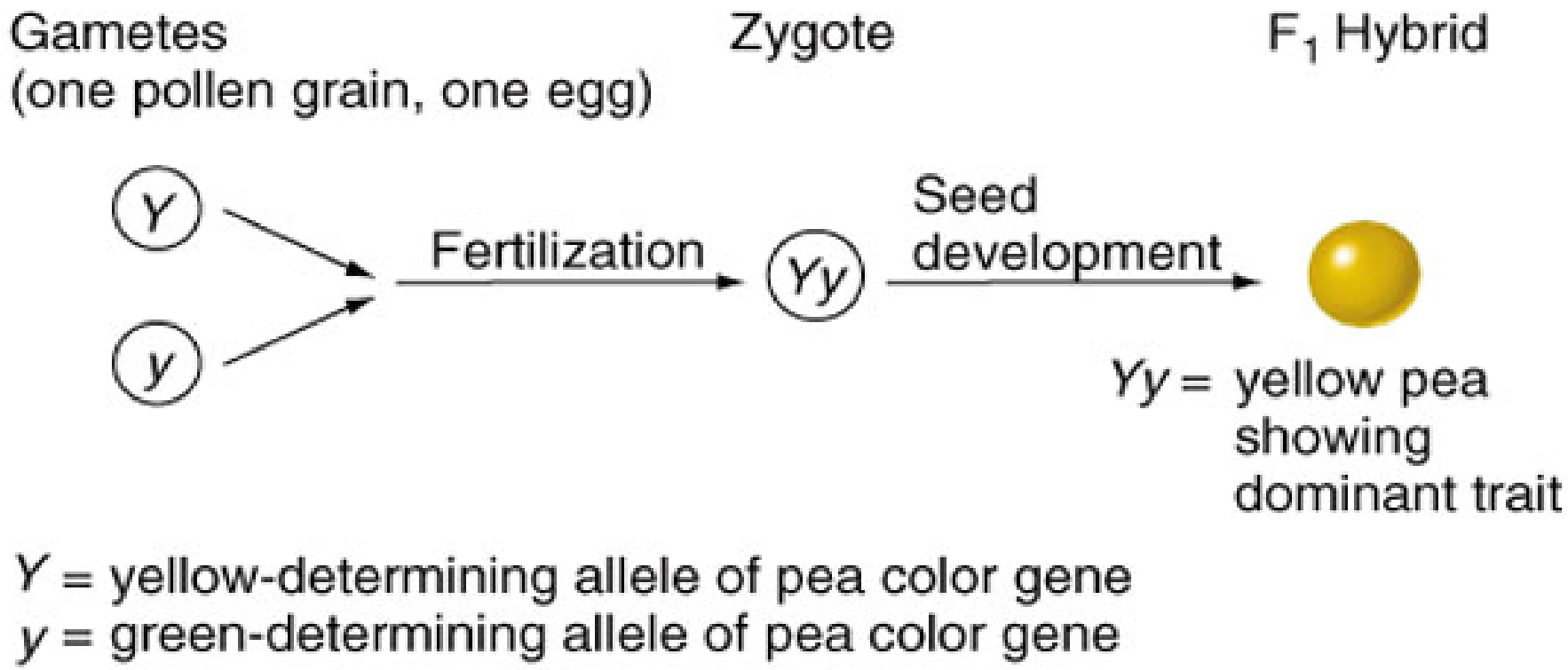


Fig. 2.10a

(b) Two gametes, one from each parent, unite at random at fertilization.



Y = yellow-determining allele of pea color gene
y = green-determining allele of pea color gene

Fig. 2.10b

The Punnet square: visual summary of a cross

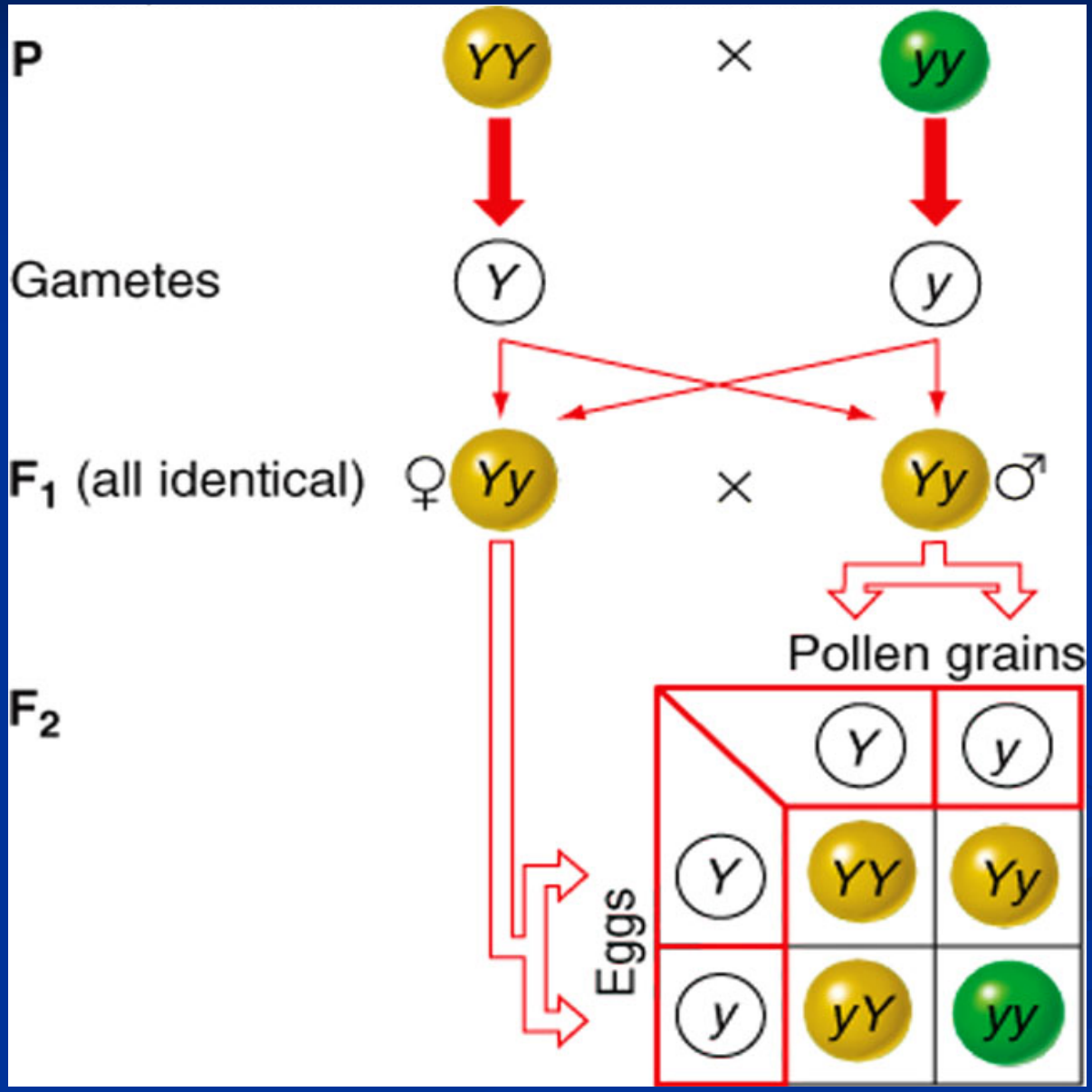


Fig. 2.11

Mendel's results reflect basic rules of probability

The Product Rule:

The probability of two **independent events** occurring together

= probability of event A × probability of event B

The Sum Rule:

The probability of either of two **mutually exclusive events** occurring

= probability of event A + probability of event B

- Cross Yy ×Yy pea plants.
  - Chance of YY offspring
    - Chance of sperm with Y allele: 1/2
    - Chance of egg with Y allele: 1/2
    - Chance of YY = 1/2 x 1/2= 1/4
  - Chance of Yy offspring
    - Chance of sperm with Y allele and egg with y allele: 1/4
    - Chance of sperm with y allele and egg with Y allele: 1/4
    - Chance of Yy = 1/4 + 1/4 = 1/2

Additional crosses confirm predicted ratios

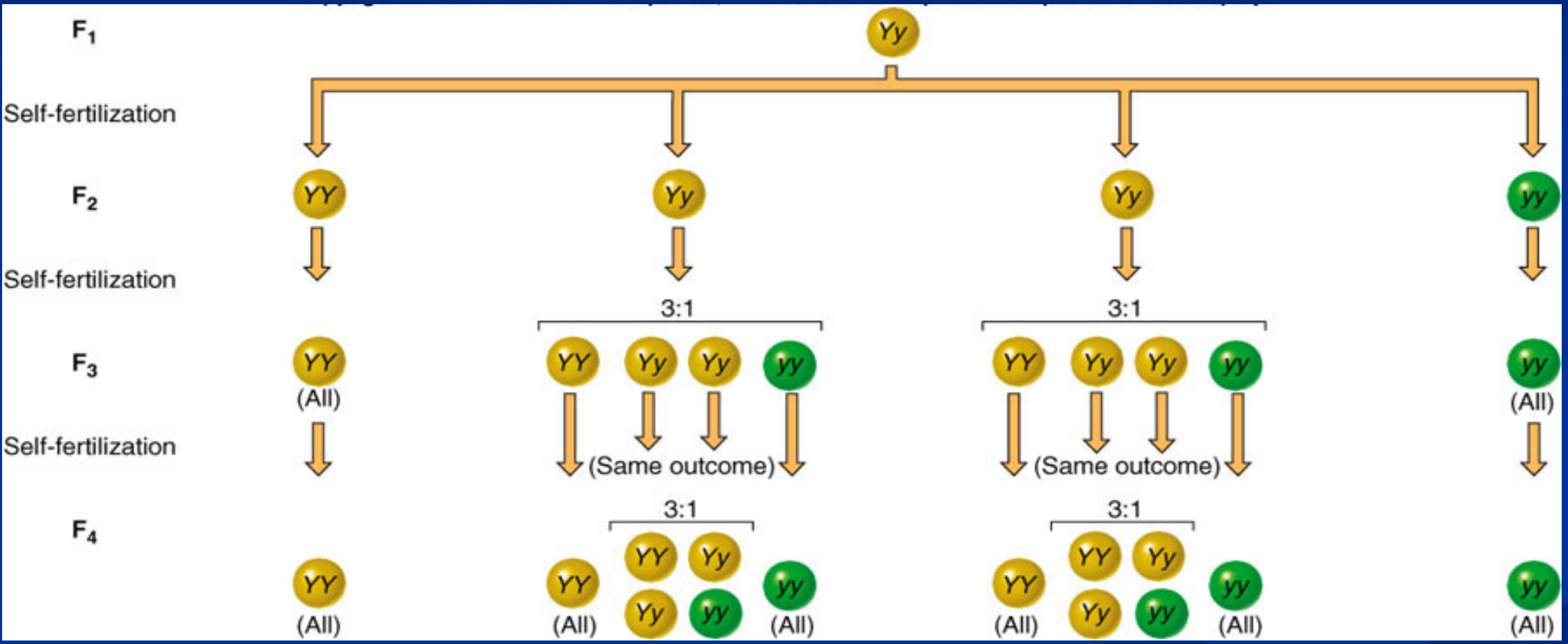
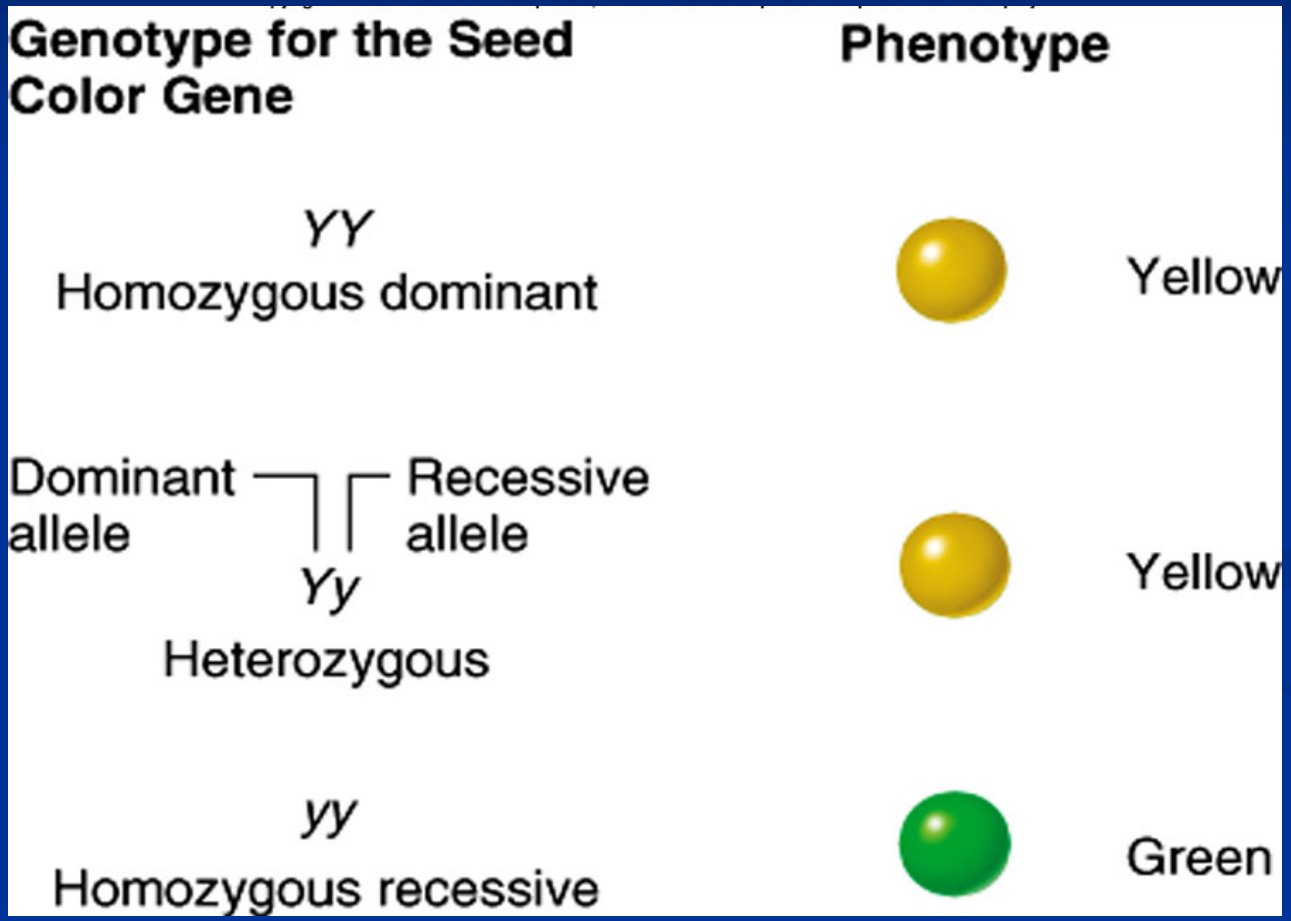


Fig. 2.12

Phenotypes and genotypes

- **Phenotype:** Observable characteristic of an organism.
- **Genotype:** Pair of alleles present in an individual.
- **Homozygous:** Two alleles of trait are the same (YY or yy).
- **Heterozygous:** Two alleles of trait are different (Yy).

Phenotypes versus genotypes



Yy × Yy

1:2:1
YY:Yy:yy

3:1
yellow: green

Fig. 2.13

Test cross reveals unknown genotype

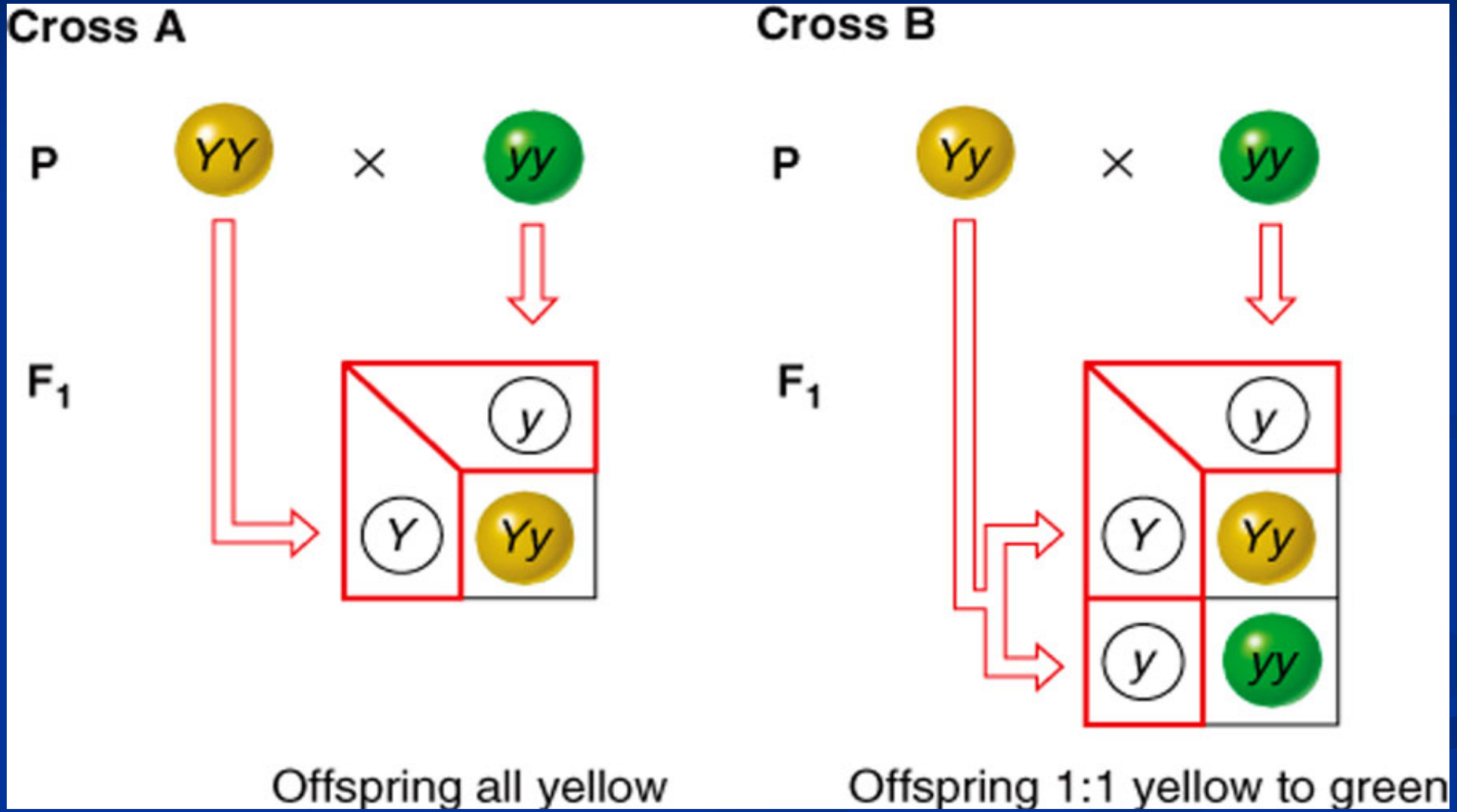


Fig. 2.14

Dihybrid crosses

- A **dihybrid** is an individual that is heterozygous at two genes.
- Mendel designed experiments to determine if two genes segregate independently of one another in dihybrids.

- **First constructed true-breeding lines for both traits, crossed them to produce dihybrid offspring, and examined the F2 for parental or recombinant types.**

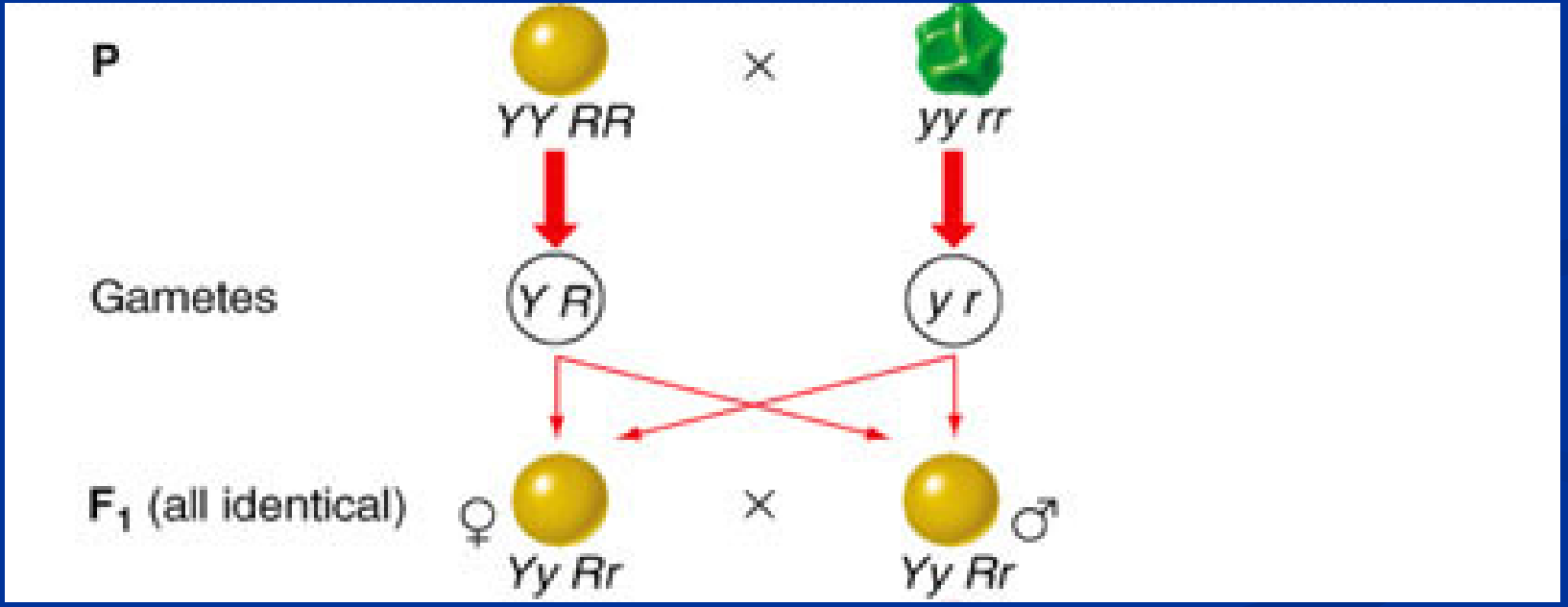


Fig. 2.15

F2

Type	Genotype		Phenotype	Number	Phenotypic ratio
Parental	*Y– R–*		yellow round	315	9/16
Recombinant	*yy R–*		green round	108	3/16
Recombinant	*Y– rr*		yellow wrinkled	101	3/16
Parental	*yy rr*		green wrinkled	32	1/16

Ratio of yellow (dominant) to green (recessive) = 12:4 or 3:1

Ratio of round (dominant) to wrinkled (recessive) = 12:4 or 3:1

Fig. 2.15

Mendel's second law: Law of independent assortment

During gamete formation, different pairs of alleles segregate independently of each other.

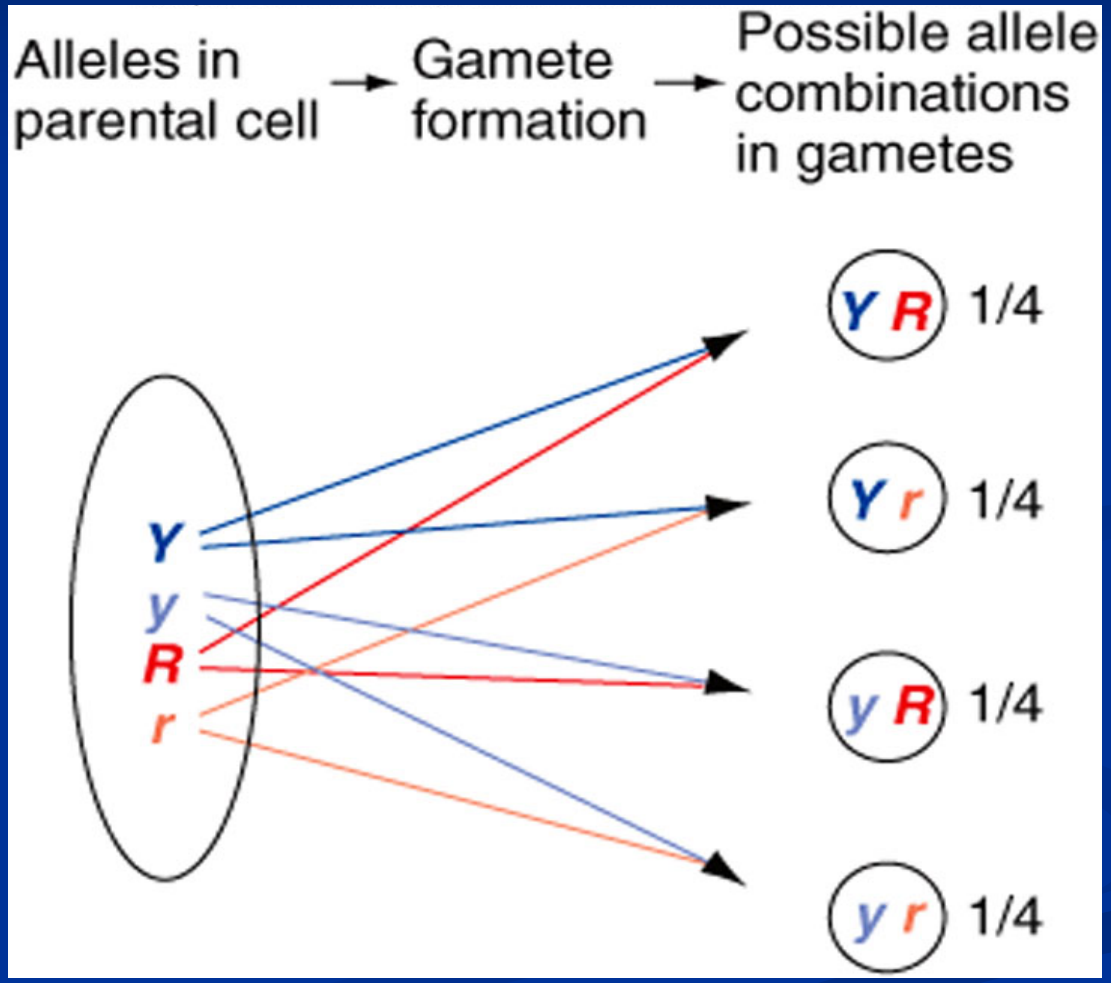
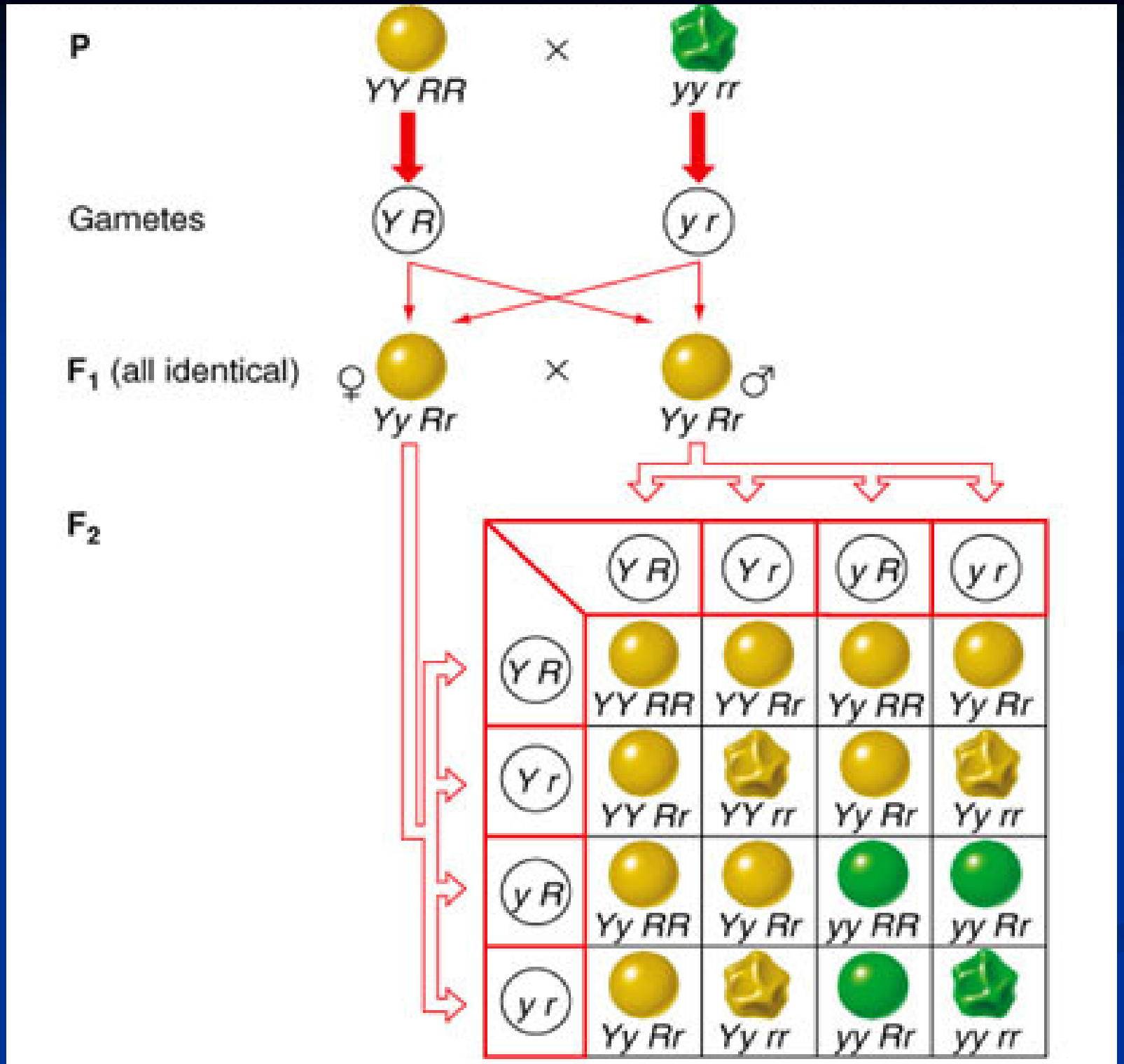


Fig. 2.16



9 genotypes
= 3^2

4 phenotypes
= 2^2

Fig. 2.15

Following crosses with branched-line diagrams

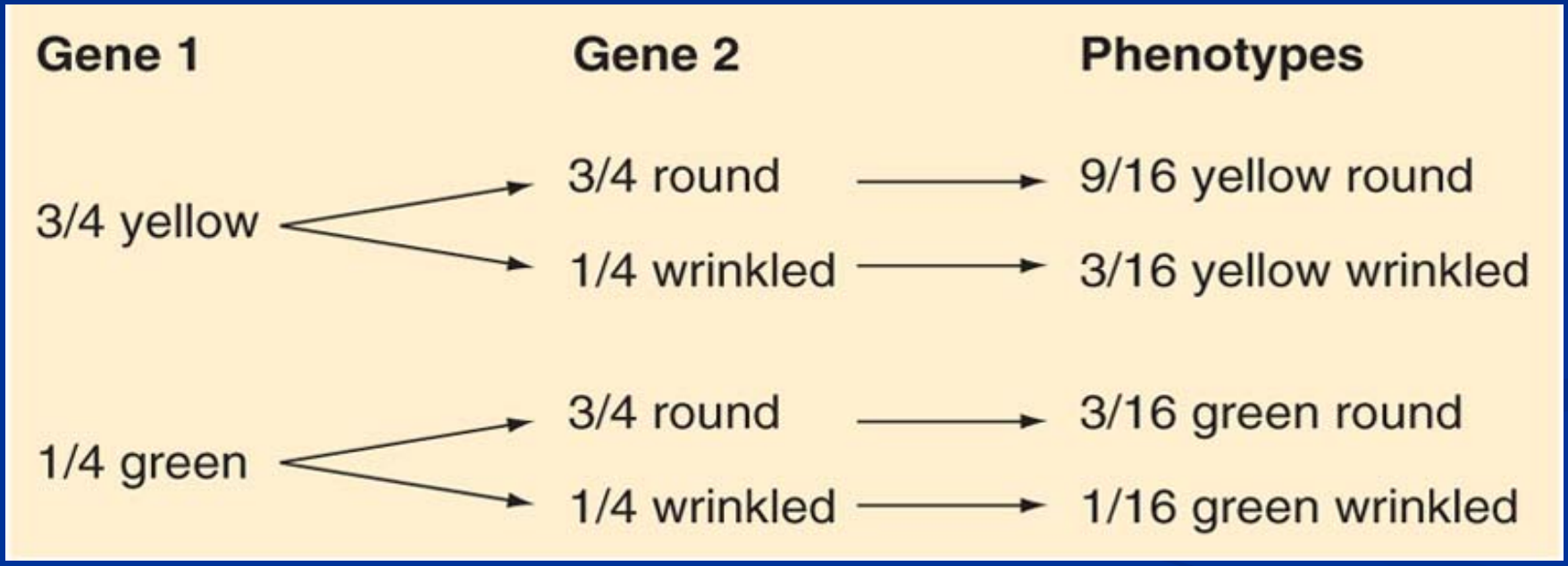


Fig. 2.17

Summary of Mendel's work

- Inheritance is particulate, not blending.
- There are two copies of units of inheritance for each trait in a germ cell.
- Gametes contain only one copy of the unit of inheritance for each trait.
- Alleles (different forms of the trait) segregate randomly during gamete formation.
- Alleles are dominant or recessive, which leads to the difference between genotype and phenotype.
- Different pairs of alleles for different traits assort independently during gamete formation.

Mendel solved the three basic questions of genetics

1. What is inherited?

Mendel: Alleles of genes.

2. How is it inherited?

Mendel: According to the laws of segregation and independent assortment.

3. What is the role of chance in heredity?

Mendel: Inheritance is determined by chance for each individual, but it is governed by defined probabilities in a population.

Mendel's success relied on a scientific approach to solve problems

1. Observe a genetic process.
2. Analyze data.
3. Develop a working model.

Rediscovery of Mendel's work

- Mendel's work was unappreciated and remained dormant for 34 years.
- In the late 1800's, Darwin's theory was viewed with skepticism because he could not explain the persistence of variation in organisms.

- **In 1900, 16 years after Mendel died, three scientists rediscovered and confirmed Mendel's laws, giving birth to the science of genetics.**

(a) Gregor Mendel

(b) Carl Correns

(c) Hugo de Vries

(d) Eric von Tschermak

Fig. 2.19

Four themes in Mendelian genetics

1. Variation is widespread in nature.
2. Observable variation is essential for following genes.
3. Variation is inherited according to genetic laws and not solely by chance.
4. Mendel's laws apply to **all** sexually reproducing organisms.

2.3 Mendelian inheritance in humans

- Most traits in humans are due to the interaction of multiple genes and do not show a simple Mendelian pattern of inheritance.

- A fraction of traits (4300, in 2009) represent single-genes traits, such as sickle-cell anemia, cystic fibrosis, Tay-Sachs disease, and Huntington's disease.

TABLE 2.1 Some of the Most Common Single-Gene Traits in Humans

Disease	Effect	Incidence of Disease
Caused by a Recessive Allele		
Thalassemia (chromosome 16 or 11)	Reduced amounts of hemoglobin; anemia, bone and spleen enlargement	1/10 in parts of Italy
Sickle-cell anemia (chromosome 11)	Abnormal hemoglobin; sickle-shaped red cells, anemia, blocked circulation; increased resistance to malaria	1/625 African-Americans
Cystic fibrosis (chromosome 7)	Defective cell membrane protein; excessive mucous production; digestive and respiratory failure	1/2000 Caucasians
Tay-Sachs disease (chromosome 15)	Missing enzyme; buildup of fatty deposit in brain; buildup destroys mental development	1/3000 Eastern European Jews
Phenylketonuria (PKU) (chromosome 12)	Missing enzyme; mental deficiency	1/10,000 Caucasians
Caused by a Dominant Allele		
Hypercholesterolemia (chromosome 19)	Missing protein that removes cholesterol from the blood; heart attack by age 50	1/122 French Canadians
Huntington disease (chromosome 4)	Progressive mental and neurological damage; neurologic disorders by ages 40–70	1/25,000 Caucasians

In humans, we must use pedigree to study inheritance

- **Pedigree**, a family history, is an orderly diagram of a family's relevant genetic features extending through multiple generations.
- Pedigrees help us infer if a trait is from a single gene and if the trait is dominant or recessive.

Symbols used in pedigree analysis

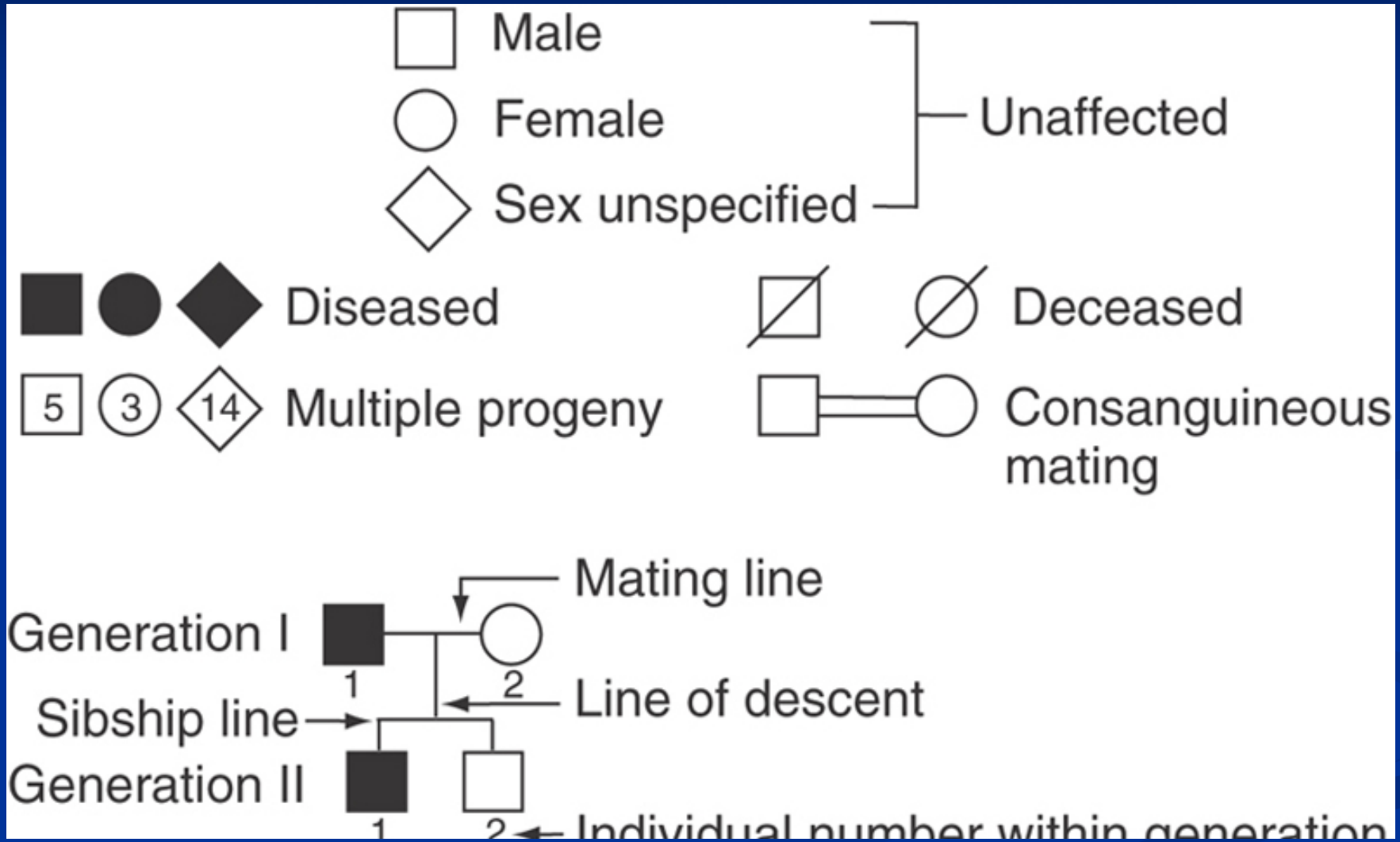
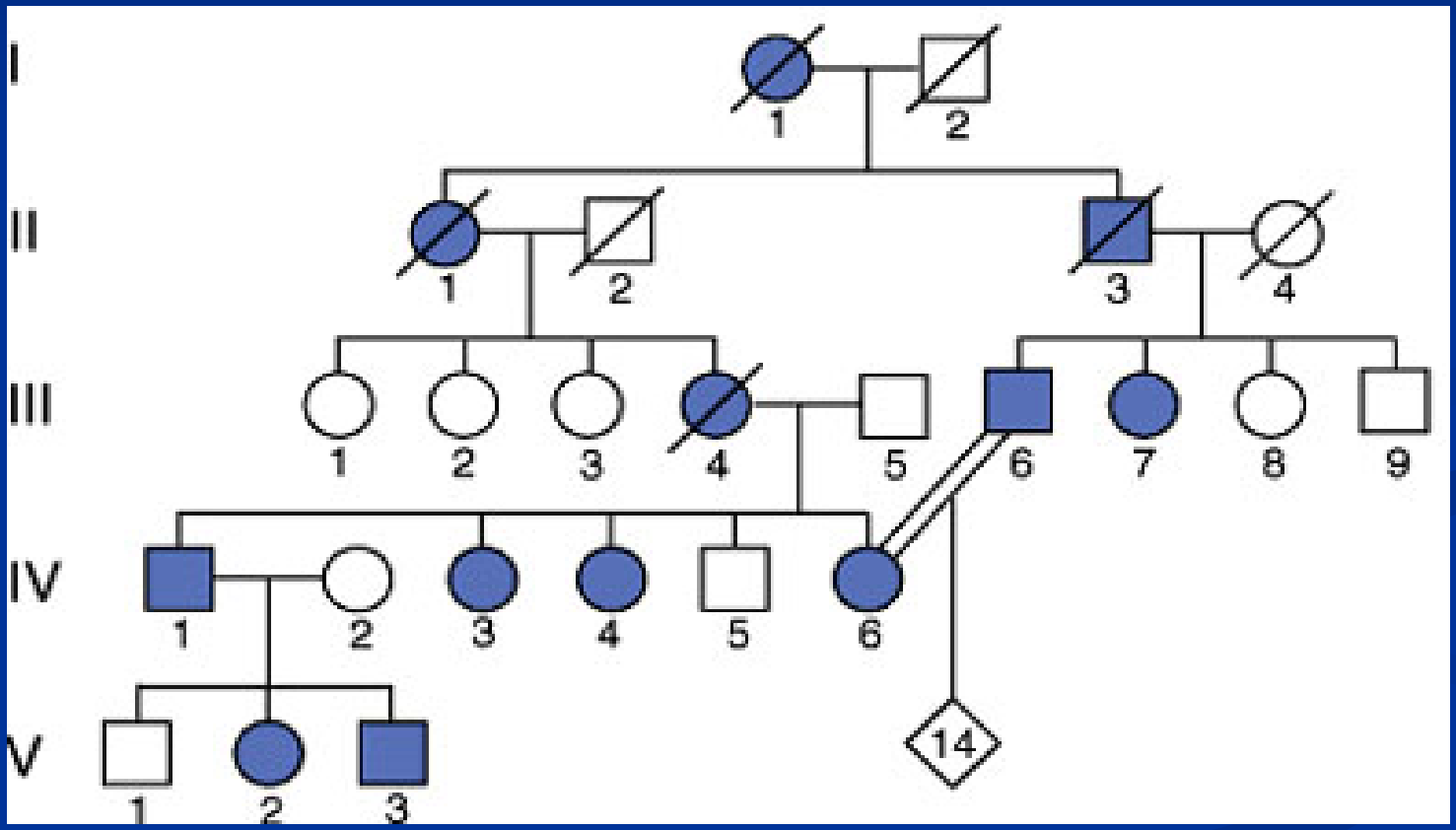


Fig. 2.20

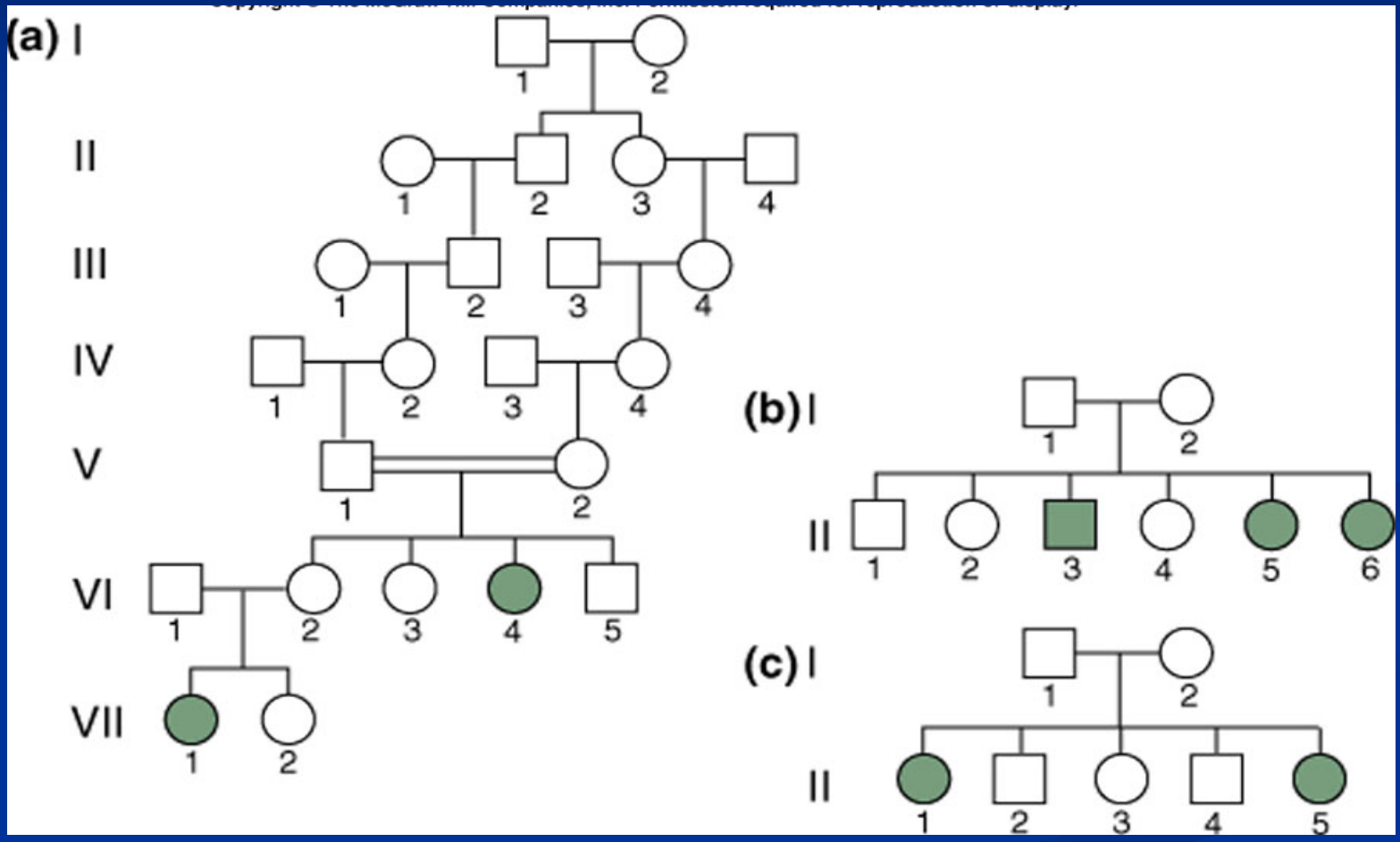
A vertical pattern of inheritance indicates a rare dominant trait



Huntington's disease: A rare dominant disease.

Fig. 2.21

A horizontal pattern of inheritance indicates a rare recessive trait



Cystic fibrosis: a recessive disease

Fig. 2.22

TABLE 2.2 How to Recognize Dominant and Recessive Traits in Pedigrees

Dominant Traits

1. Affected children always have at least one affected parent.
2. As a result, dominant traits show a *vertical pattern* of inheritance: the trait shows up in every generation.
3. Two affected parents can produce unaffected children, if both parents are heterozygotes.

Recessive Traits

1. Affected individuals can be the children of two unaffected carriers, particularly as a result of consanguineous matings.
2. All the children of two affected parents should be affected.
3. *Rare* recessive traits show a *horizontal pattern* of inheritance: the trait first appears among several members of one generation and is not seen in earlier generations.
4. Recessive traits may show a vertical pattern of inheritance if the trait is extremely common in the population.