I Meccanismi dell’Eredità. I

G. Camelopardalis

Teorie uniparentali

Teoria del mescolamento

Genitori

Cavallo
($2n = 64$)

Asino
($2n = 62$)

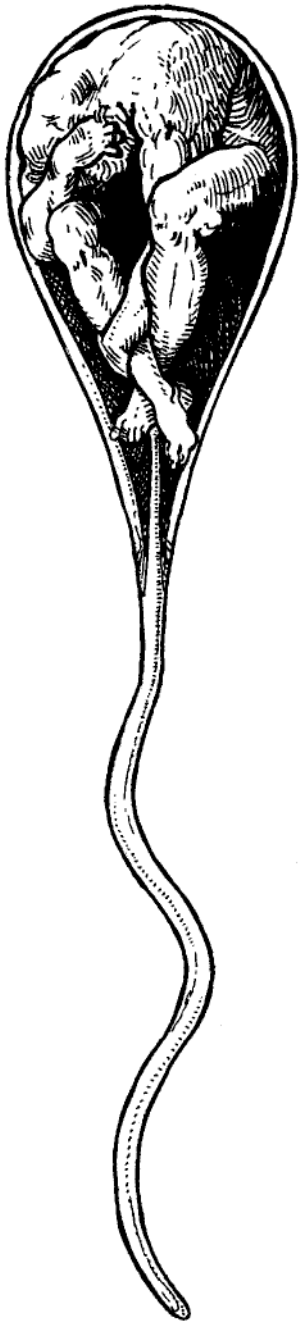
Cavallo ♀ × Asino ♂ →

Mulo
($2n = 63$)

Progenie

Cavallo ♂ × Asino ♀ →

Bardotto
($2n = 63$)



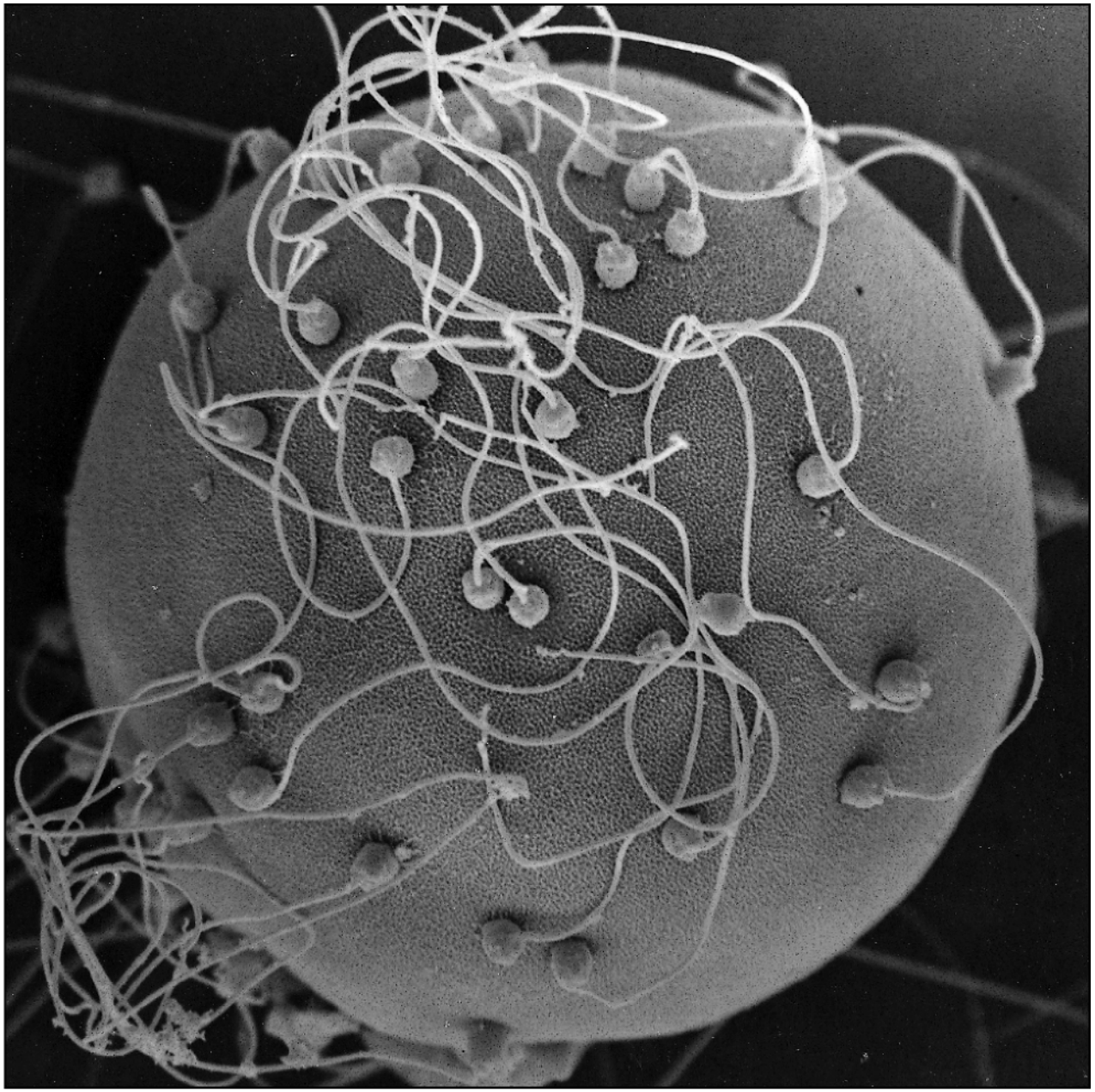

50 µm

Figure 2.2

Gregor Johann Mendel, founder of the science of geneti

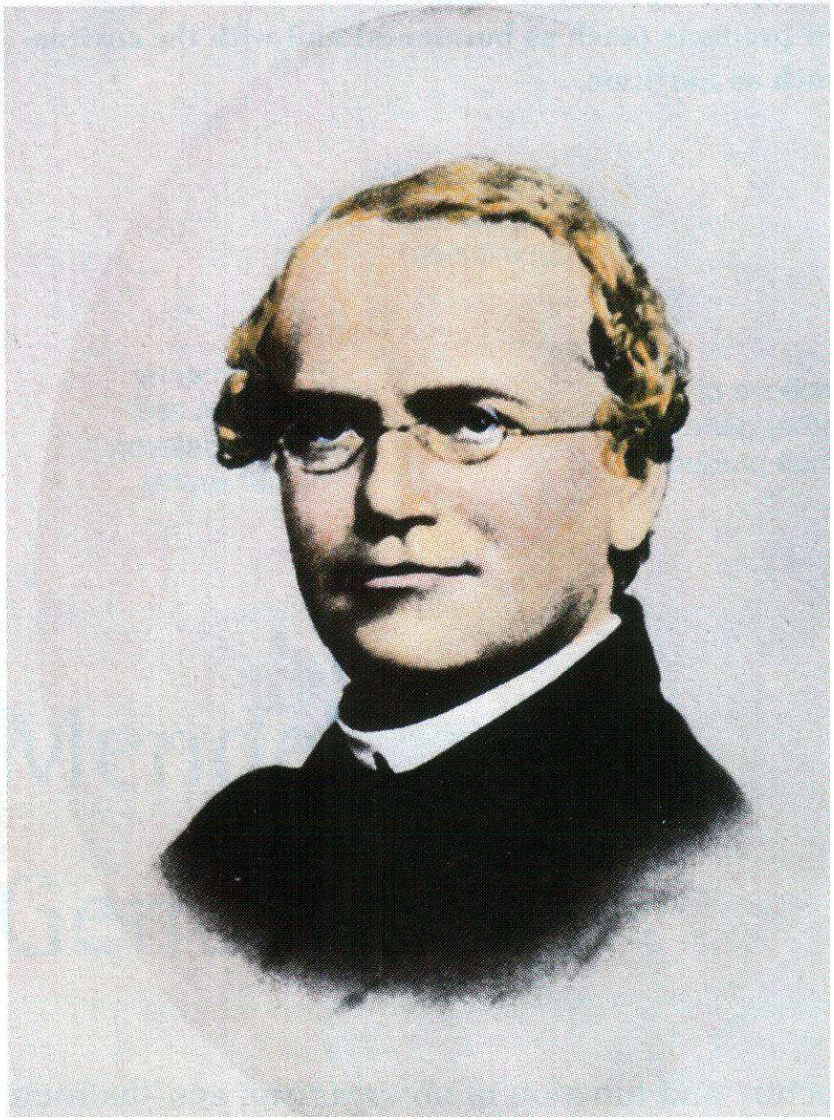


Figure 1.25

Generalized structure of a flower.

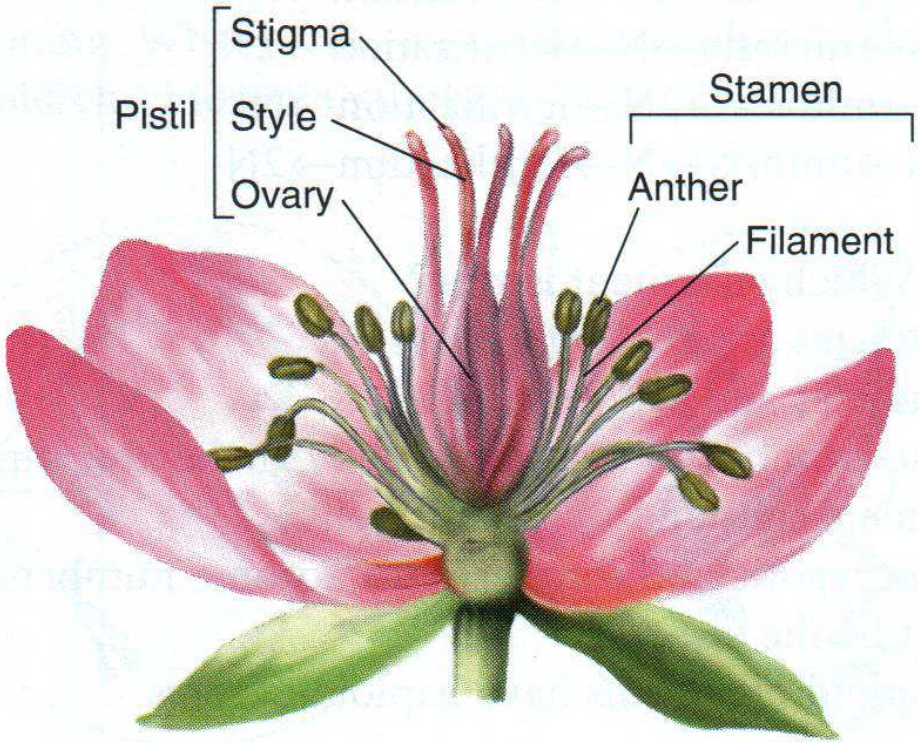
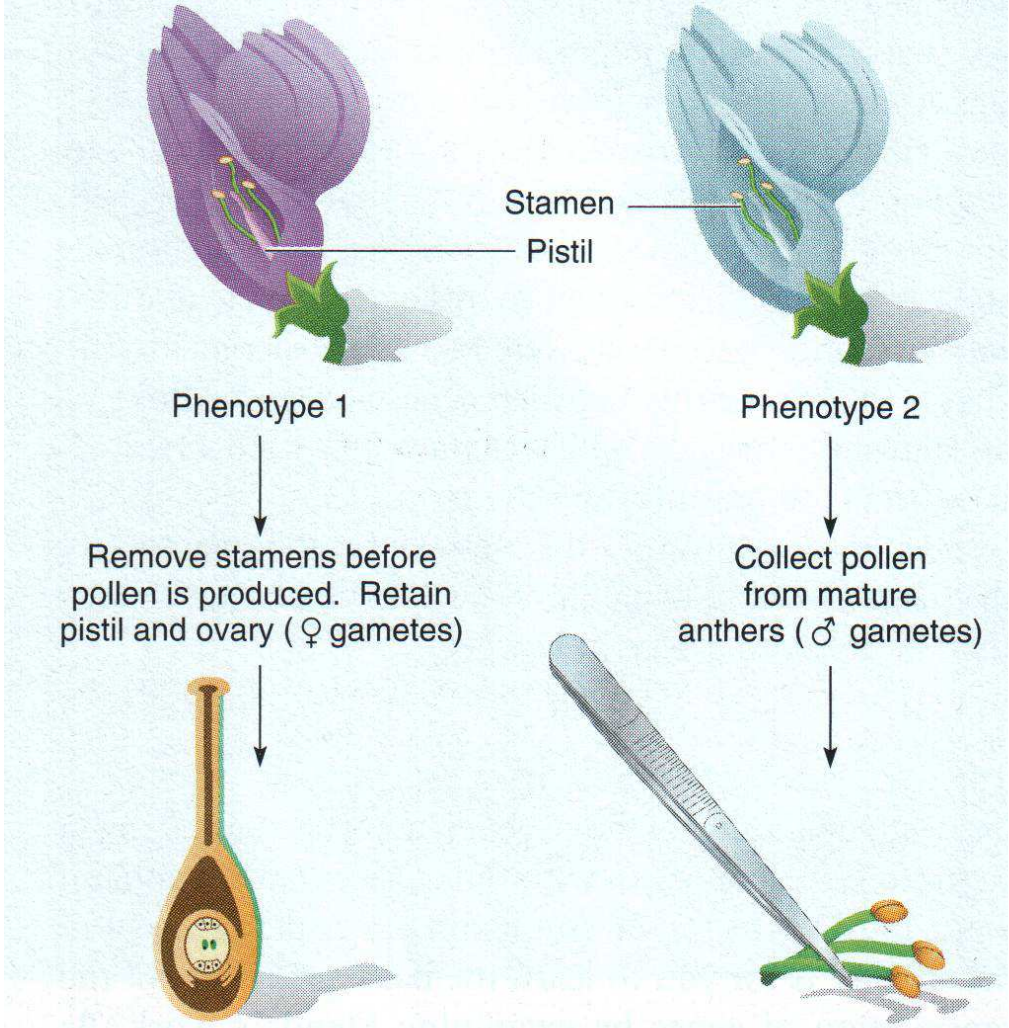
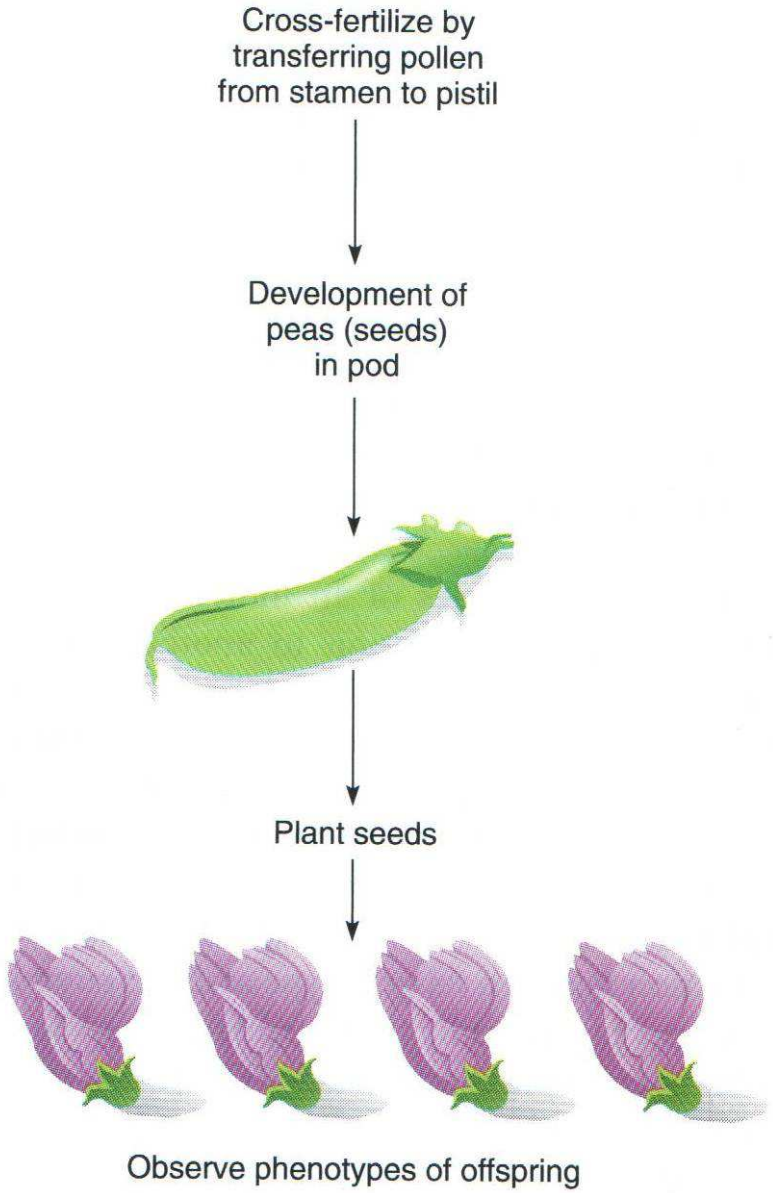


Figure 2.3

Procedure for crossing pea plants.




Cross-fertilize by transferring pollen from stamen to pistil
Development of peas (seeds) in pod
Plant seeds
Observe phenotypes of offspring

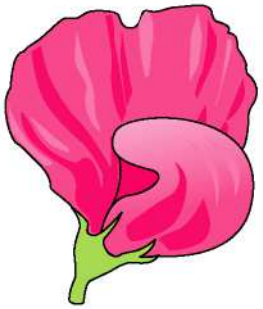
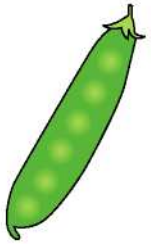
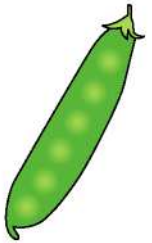
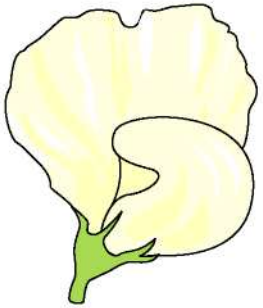
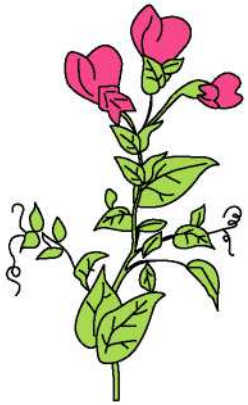
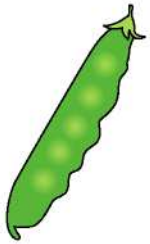
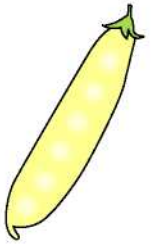
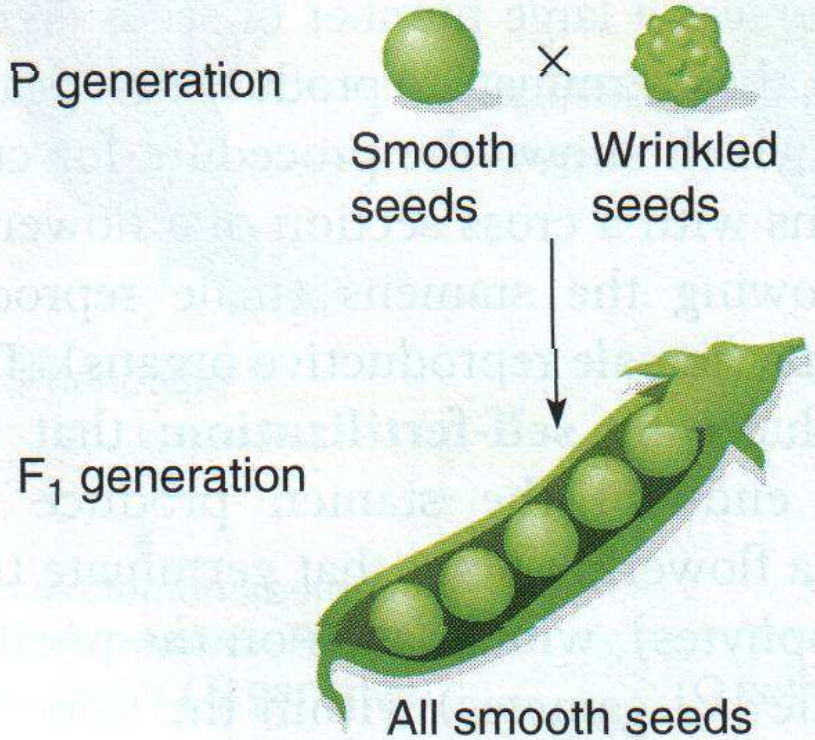
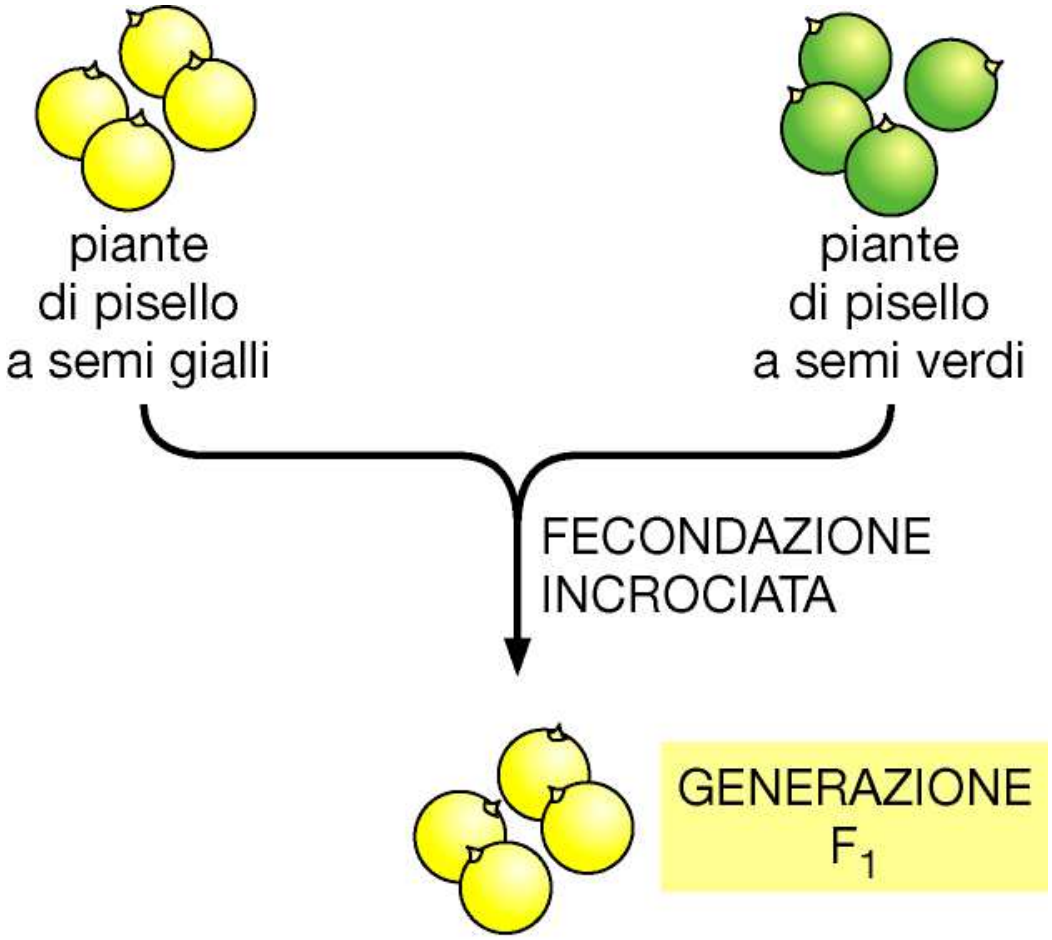
	Forma del seme	Colore del seme	Colore del fiore	Posizione del fiore	Forma del baccello	Colore del baccello	Altezza della pianta
Prima variante del carattere (dominante)							
rotondo (*R*)	giallo (*Y*)	rosso violetto	fiori assiali	turgido	verde	alta	
Seconda variante del carattere (recessiva)							
rugoso (*r*)	verde (*y*)	bianco	fiori terminali	raggrinzito	giallo	bassa	

Figure 2.5

Results of one of Mendel's breeding crosses. In the parental generation, he crossed a true-breeding pea strain that produced smooth seeds with one that produced wrinkled seeds. All the F_1 progeny seeds were smooth.

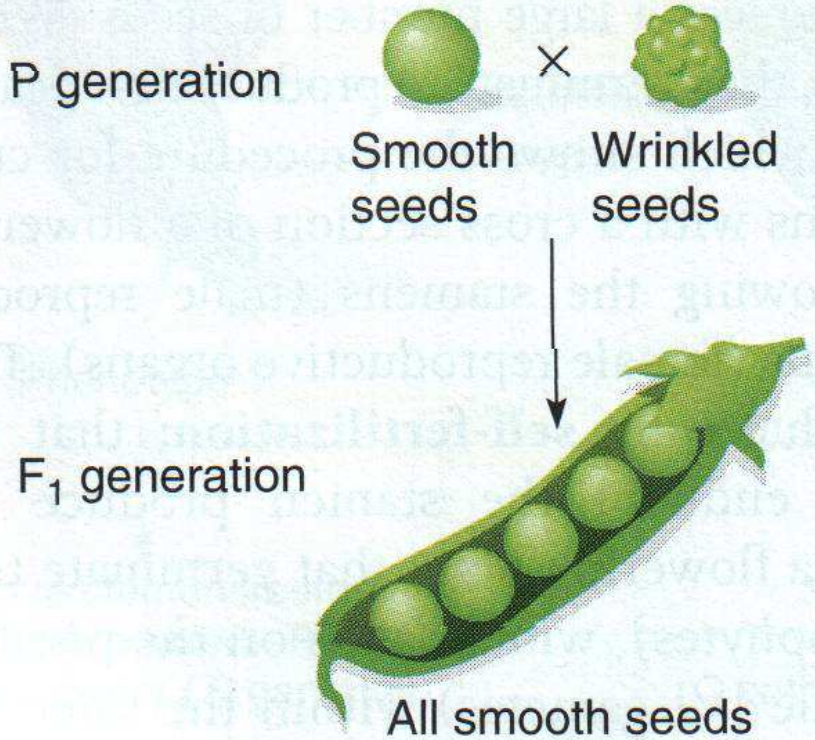


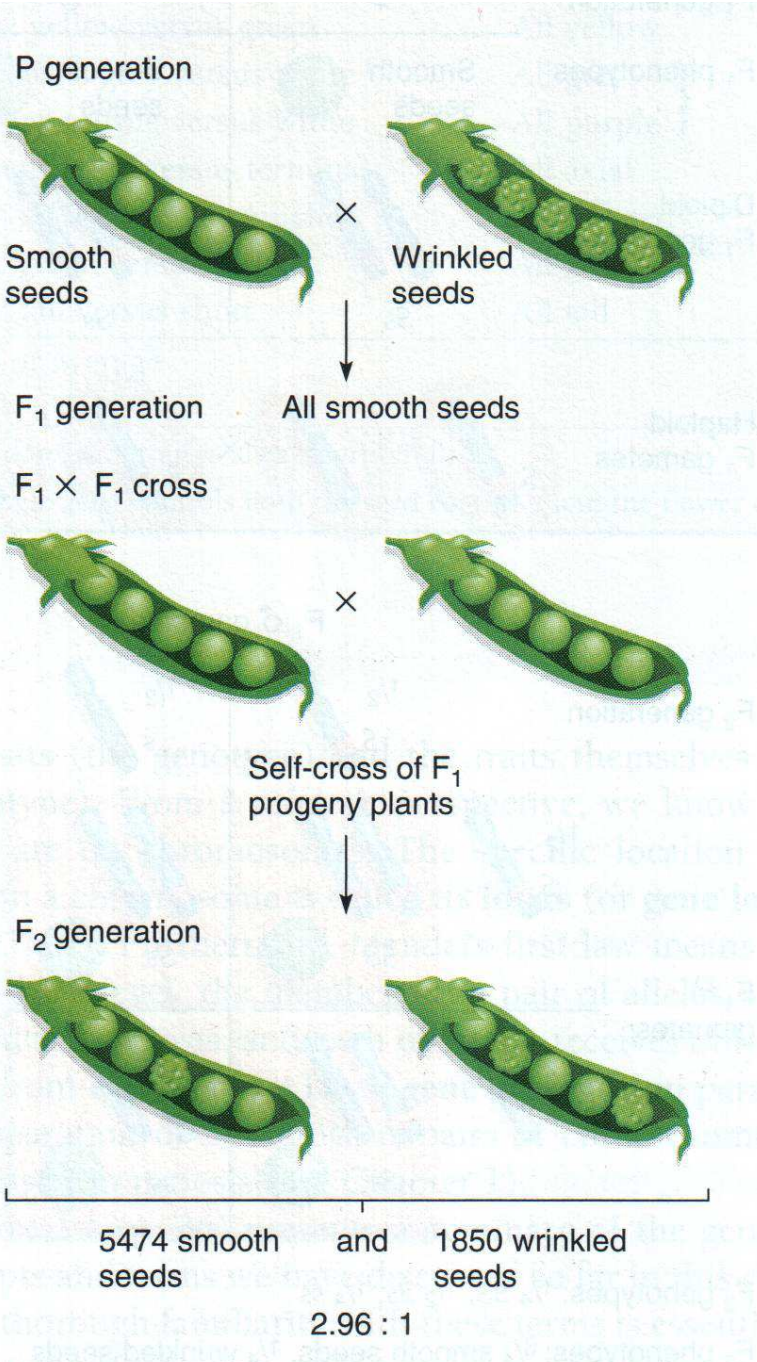

piante
di pisello
a semi gialli
piante
di pisello
a semi verdi
FECONDAZIONE
INCROCIATA
GENERAZIONE
F_1
100% di piante a semi gialli

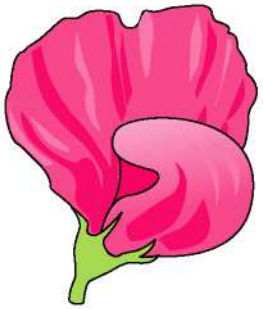
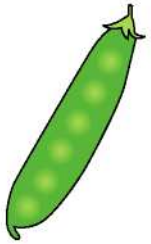
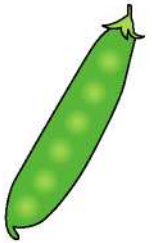
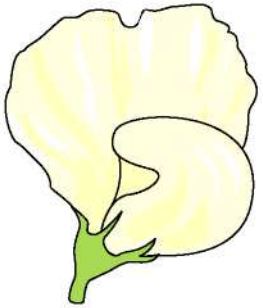
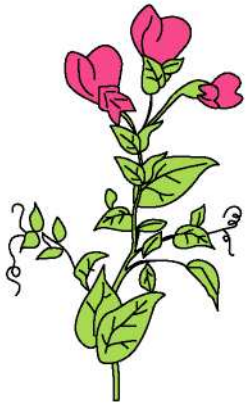
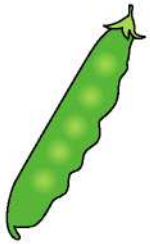
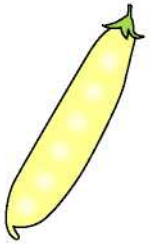
Character[a]	$\mathbf{F_1}$
Seeds: smooth versus wrinkled	All smooth
Seeds: yellow versus green	All yellow
Seed coats: grey versus white [b]	All grey
Flowers: purple versus white	All purple
Flowers: axial versus terminal	All axial
Pods: inflated versus pinched	All inflated
Pods: green versus yellow	All green
Stem: tall versus short	All tall

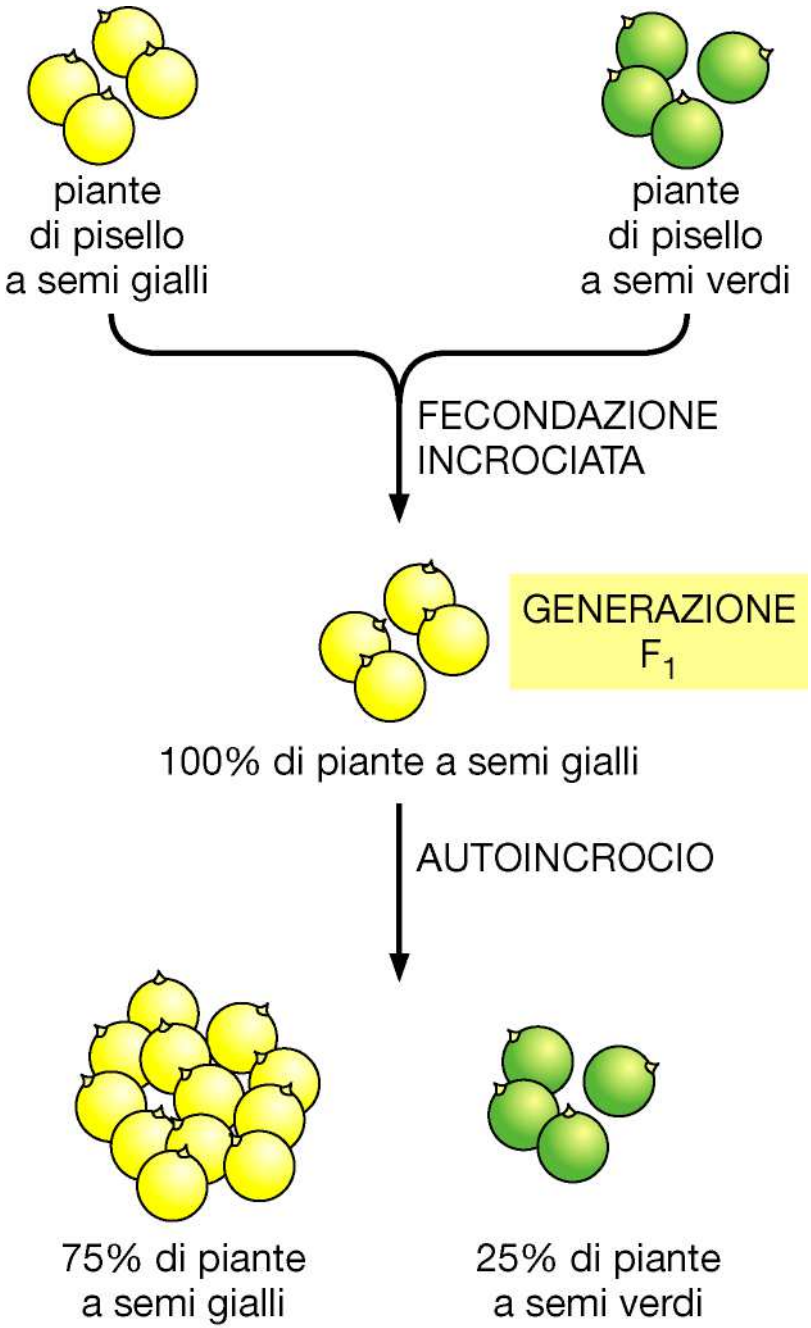
Figure 2.5

Results of one of Mendel's breeding crosses. In the parental generation, he crossed a true-breeding pea strain that produced smooth seeds with one that produced wrinkled seeds. All the F_1 progeny seeds were smooth.




P generation
Smooth seeds
×
Wrinkled seeds
F1 generation
All smooth seeds
F1 × F1 cross
×
Self-cross of F1 progeny plants
F2 generation
5474 smooth seeds
and
1850 wrinkled seeds
2.96 : 1

	Forma del seme	Colore del seme	Colore del fiore	Posizione del fiore	Forma del baccello	Colore del baccello	Altezza della pianta
Prima variante del carattere (dominante)							
rotondo (*R*)	giallo (*Y*)	rosso violetto	fiori assiali	turgido	verde	alta	
Seconda variante del carattere (recessiva)							
rugoso (*r*)	verde (*y*)	bianco	fiori terminali	raggrinzito	giallo	bassa	


piante
di pisello
a semi gialli
piante
di pisello
a semi verdi
FECONDAZIONE
INCROCIATA
GENERAZIONE
F_1
100% di piante a semi gialli
AUTOINCROCIO
75% di piante
a semi gialli
25% di piante
a semi verdi

Character[a]	F_1	F_2 (Number) Dominant	Recessive	Total	F_2 (Ratio) Dominant : Recessive
Seeds: smooth versus wrinkled	All smooth	5,474	1,850	7,324	2.96:1
Seeds: yellow versus green	All yellow	6,022	2,001	8,023	3.01:1
Seed coats: grey versus white [b]	All grey	705	224	929	3.15:1
Flowers: purple versus white	All purple				
Flowers: axial versus terminal	All axial	651	207	858	3.14:1
Pods: inflated versus pinched	All inflated	882	299	1,181	2.95:1
Pods: green versus yellow	All green	428	152	580	2.82:1
Stem: tall versus short	All tall	787	277	1,064	2.84:1
Total or average		14,949	5,010	19,959	2.98:1

Box 2.1 Genetics Terminology

Alleles: alternative forms of a gene. For example, the alleles *S* and *s* represent the smoothness and wrinkledness of the pea seed. (Like gene symbols, allele symbols are underlined or *italicized*.)

Cross: a mating between two individuals, leading to the fusion of gametes.

Diploid: a eukaryotic cell or organism with two homologous sets of chromosomes.

Gamete: a mature reproductive cell that is specialized for sexual fusion. Each gamete is haploid and fuses with a cell of similar origin but of opposite sex to produce a diploid zygote.

Gene (Mendelian factor): the determinant of a characteristic of an organism. Gene symbols are underlined or *italicized*.

Genotype: the genetic constitution of an organism. A diploid organism in which both alleles are the same at a given gene locus is said to be **homozygous** for that allele. Homozygotes produce only one gametic type with respect to that locus. For example, true-breeding smooth-seeded peas have the genotype *SS*, and true-breeding wrinkle-seeded peas have the genotype *ss*; both are homozygous. The smooth parent is **homozygous dominant;** the wrinkled parent is **homozygous recessive.**

Diploid organisms that have two different alleles at a specific gene locus are said to be **heterozygous.** Thus, F_1 hybrid plants from the cross of *SS* and *ss* parents have one *S* allele and one *s* allele. Individuals heterozygous for two allelic forms of a gene produce two kinds of gametes (*S* and *s*).

Haploid: a cell or an individual with one copy of each chromosome.

Locus (gene locus; plural = loci): the specific place on a chromosome where a gene is located.

Phenotype: the physical manifestation of a genetic trait that results from a specific genotype and its interaction with the environment. In our example, the *S* allele was dominant to the *s* allele, so in the heterozygous condition the seed is smooth. Therefore, both the homozygous dominant *SS* and the heterozygous *Ss* seeds have the same phenotype (smooth), even though they differ in genotype.

Zygote: the cell produced by the fusion of male and female gametes.

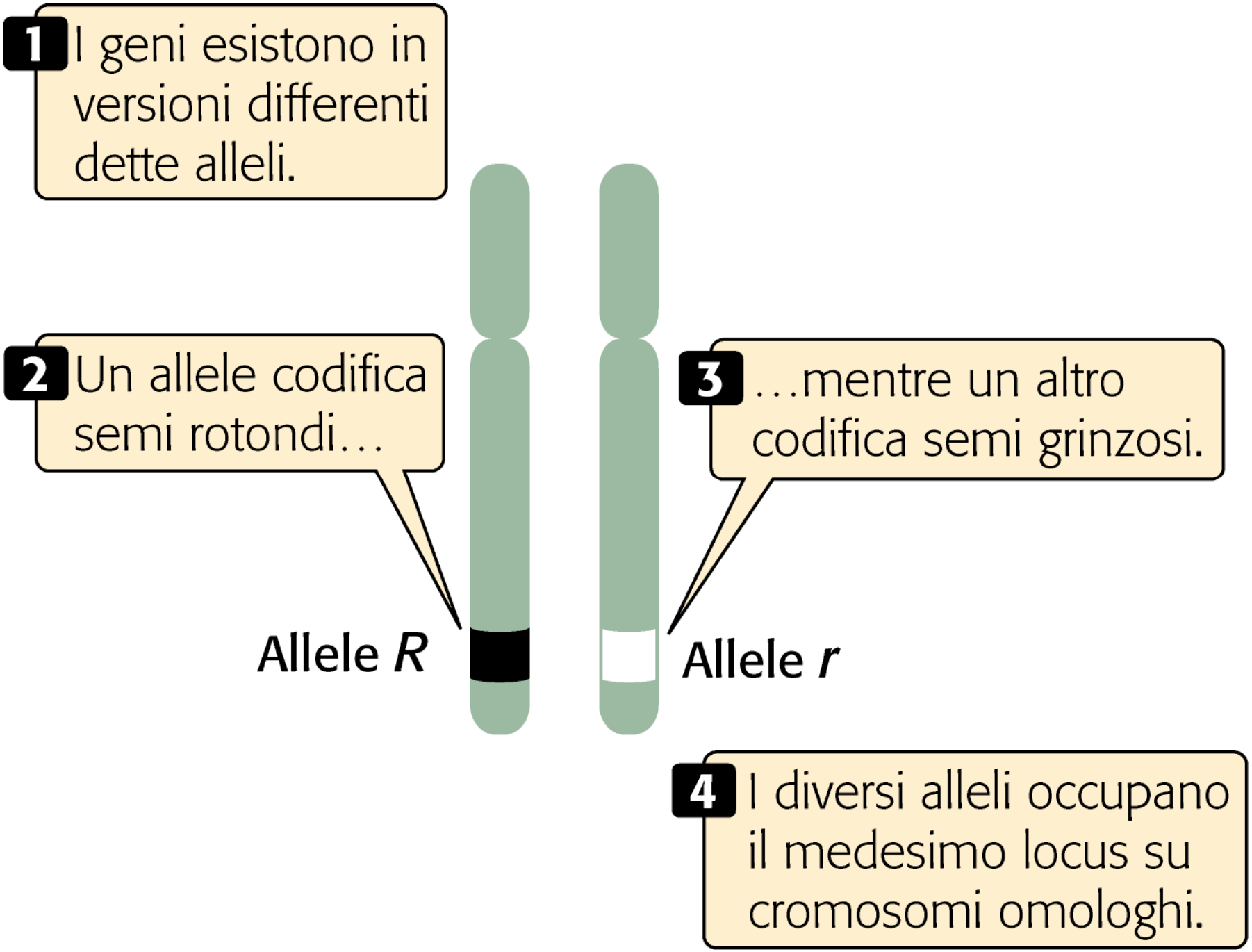
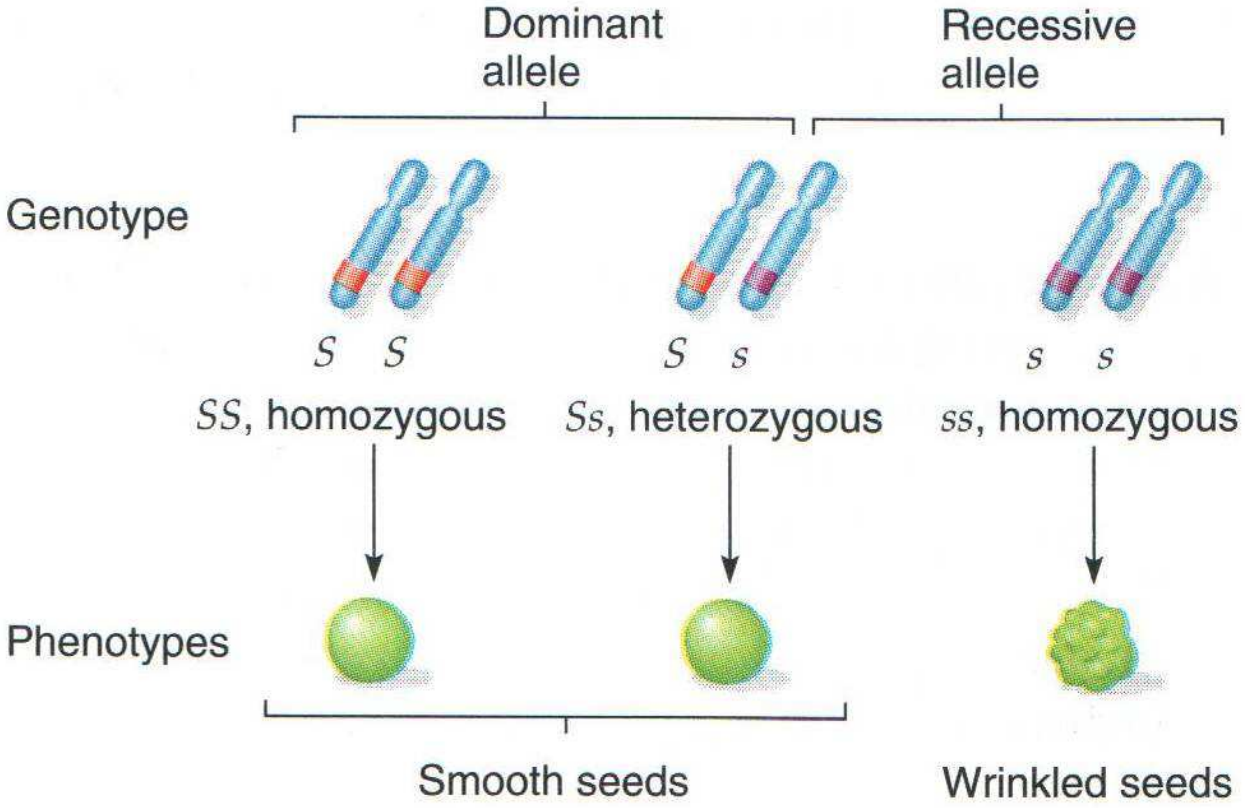
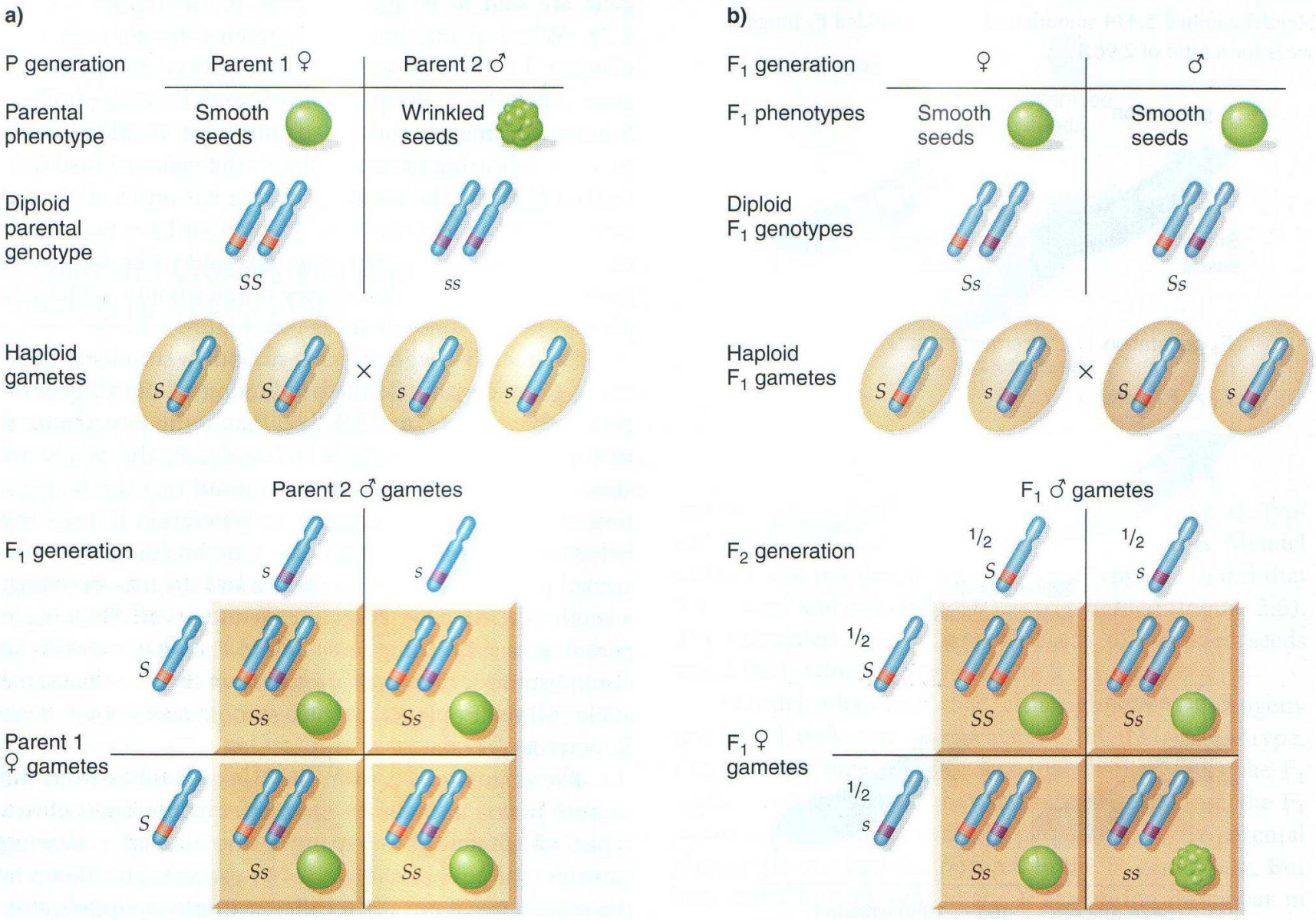

1 I geni esistono in versioni differenti dette alleli.
2 Un allele codifica semi rotondi…
3 …mentre un altro codifica semi grinzosi.
Allele R
Allele r
4 I diversi alleli occupano il medesimo locus su cromosomi omologhi.

Figure 2.7

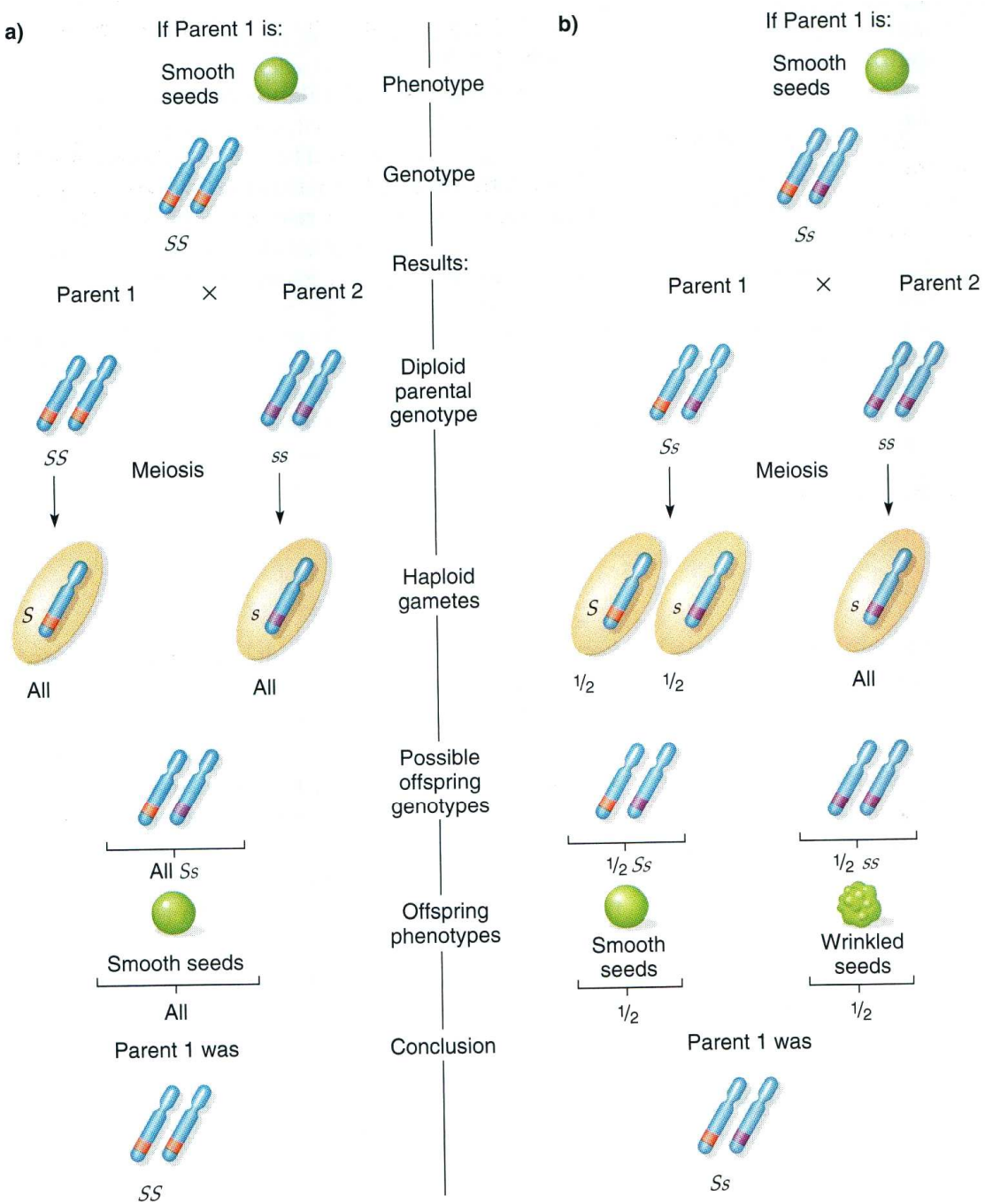
Dominant and recessive alleles of a gene for seed shape in peas.




a)
P generation
Parent 1 ♀
Parent 2 ♂
Parental phenotype
Smooth seeds
Wrinkled seeds
Diploid parental genotype
SS
ss
Haploid gametes
S
S
×
s
s
Parent 2 ♂ gametes
F1 generation
s
s
S
Ss
Ss
Parent 1 ♀ gametes
S
Ss
Ss
F1 genotypes: all Ss
F1 phenotypes: all smooth (smooth is dominant to wrinkled)
b)
F1 generation
♀
♂
F1 phenotypes
Smooth seeds
Smooth seeds
Diploid F1 genotypes
Ss
Ss
Haploid F1 gametes
S
s
×
S
s
F1 ♂ gametes
F2 generation
1/2
S
1/2
s
1/2
S
SS
Ss
F1 ♀ gametes
1/2
s
Ss
ss
F2 genotypes: 1/4 SS, 1/2 Ss, 1/4 ss
F2 phenotypes: 3/4 smooth seeds, 1/4 wrinkled seeds

Il test del **re-incrocio** consiste nell’incrociare un individuo con genotipo non noto con un omozigote recessivo.

L’analisi del test consente di determinare il genotipo non noto.


a)
If Parent 1 is:
Smooth seeds
Phenotype
Genotype
SS
Results:
Parent 1
×
Parent 2
Diploid parental genotype
SS
Meiosis
ss
S
s
Haploid gametes
All
All
All Ss
Possible offspring genotypes
Smooth seeds
All
Offspring phenotypes
Parent 1 was
Conclusion
SS
b)
If Parent 1 is:
Smooth seeds
Ss
Parent 1
×
Parent 2
Ss
Meiosis
ss
S
s
s
1/2
1/2
All
1/2 Ss
1/2 ss
Smooth seeds
Wrinkled seeds
1/2
1/2
Parent 1 was
Ss

Prima Legge di Mendel:

(Principio della Segregazione)

Le due copie di un gene (gli alleli), segregano durante la formazione dei gameti.

La segregazione degli alleli è data dalla separazione dei cromosomi omologhi durante l'anafase della prima divisione meiotica.

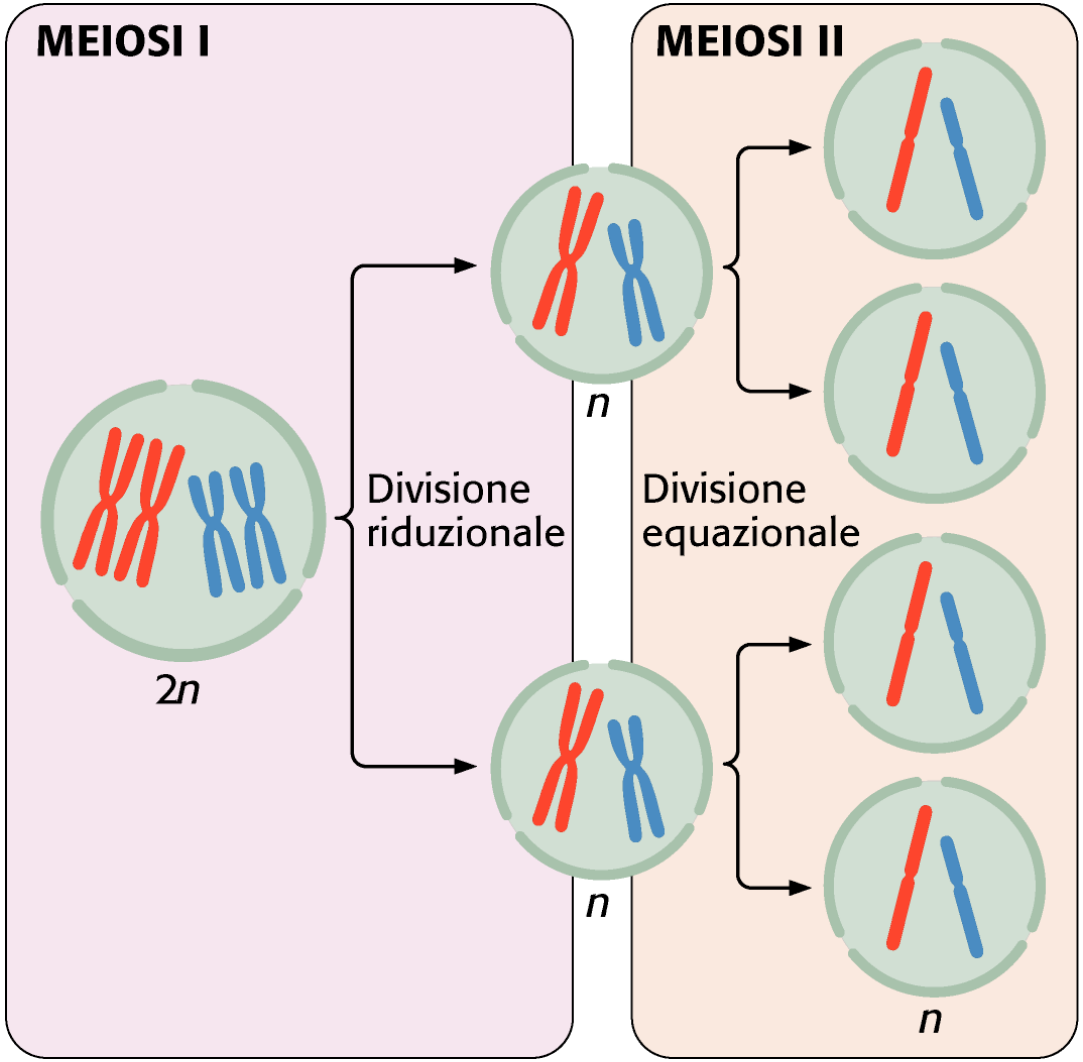
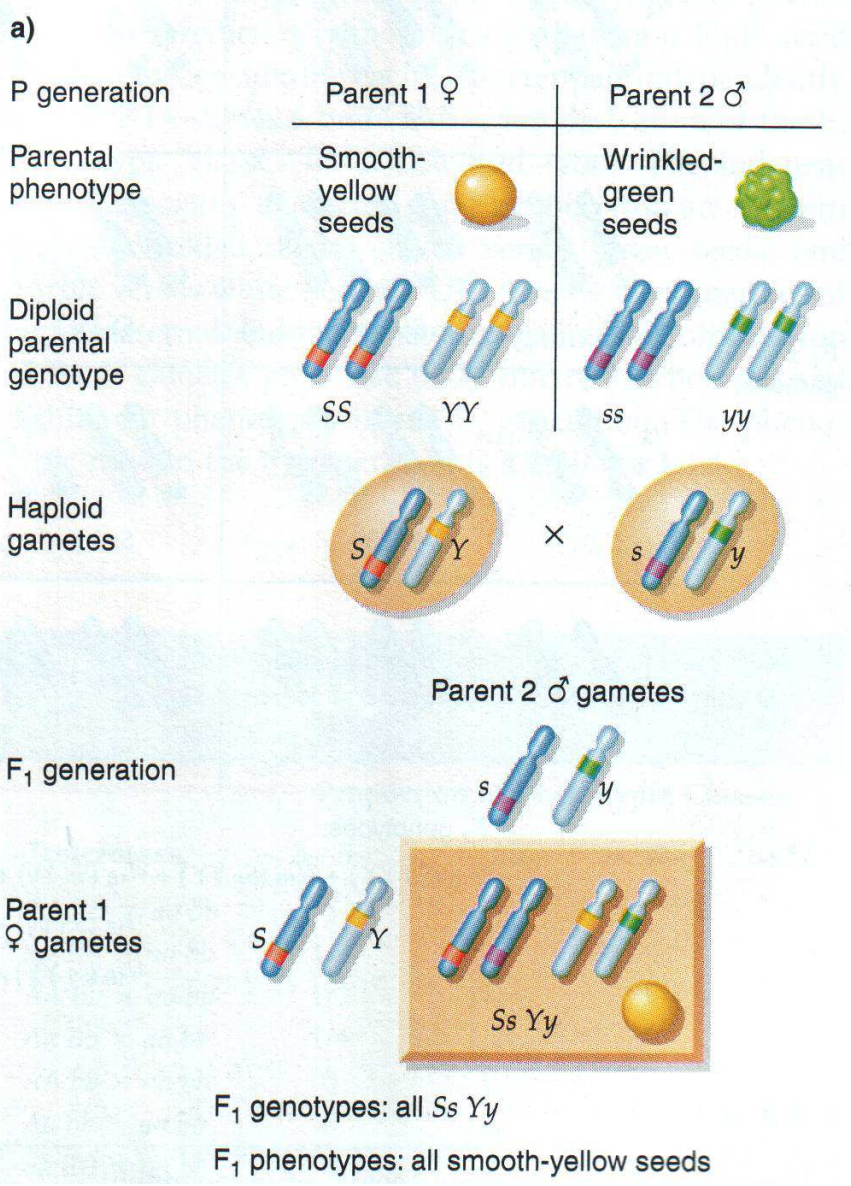
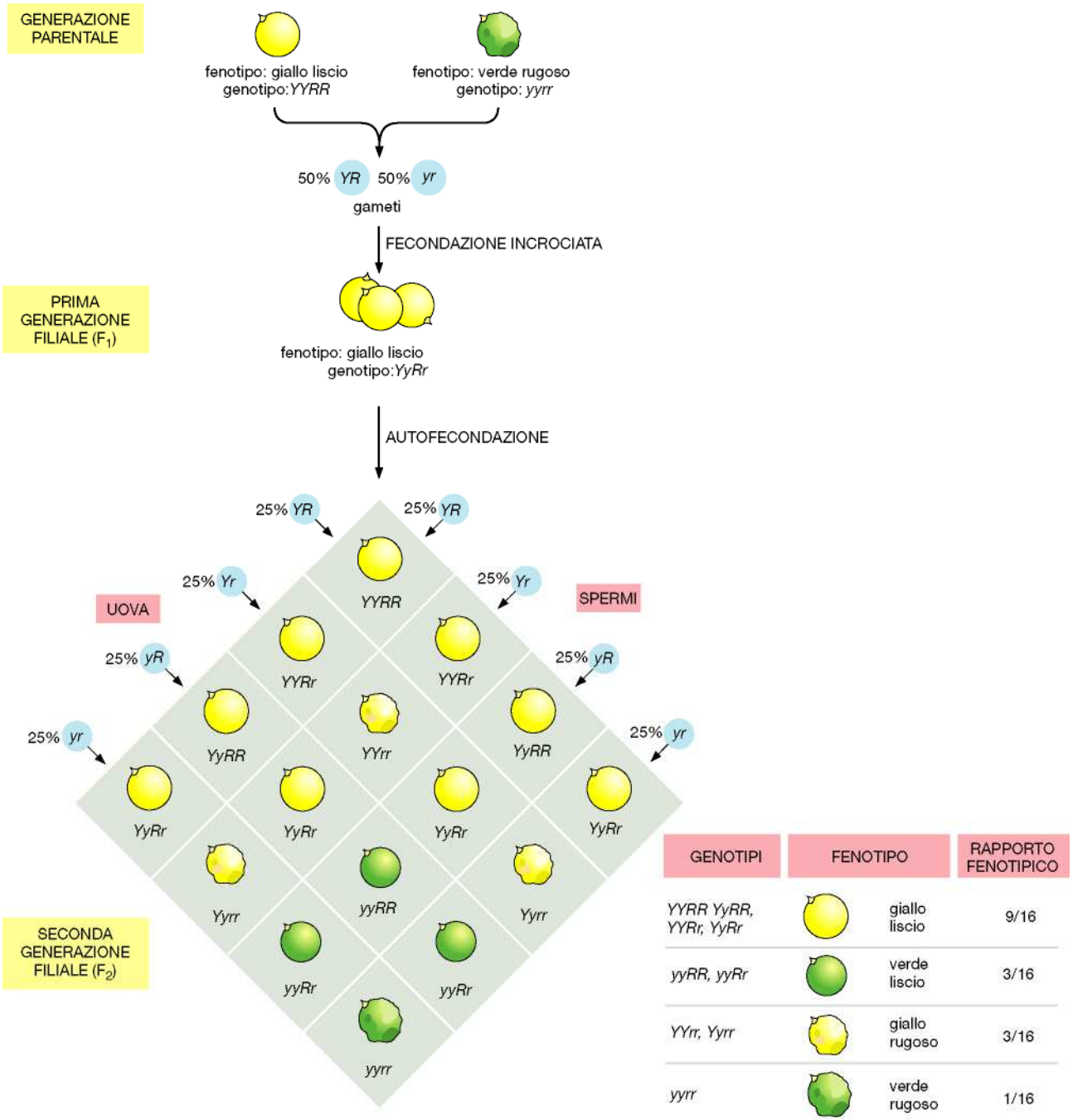
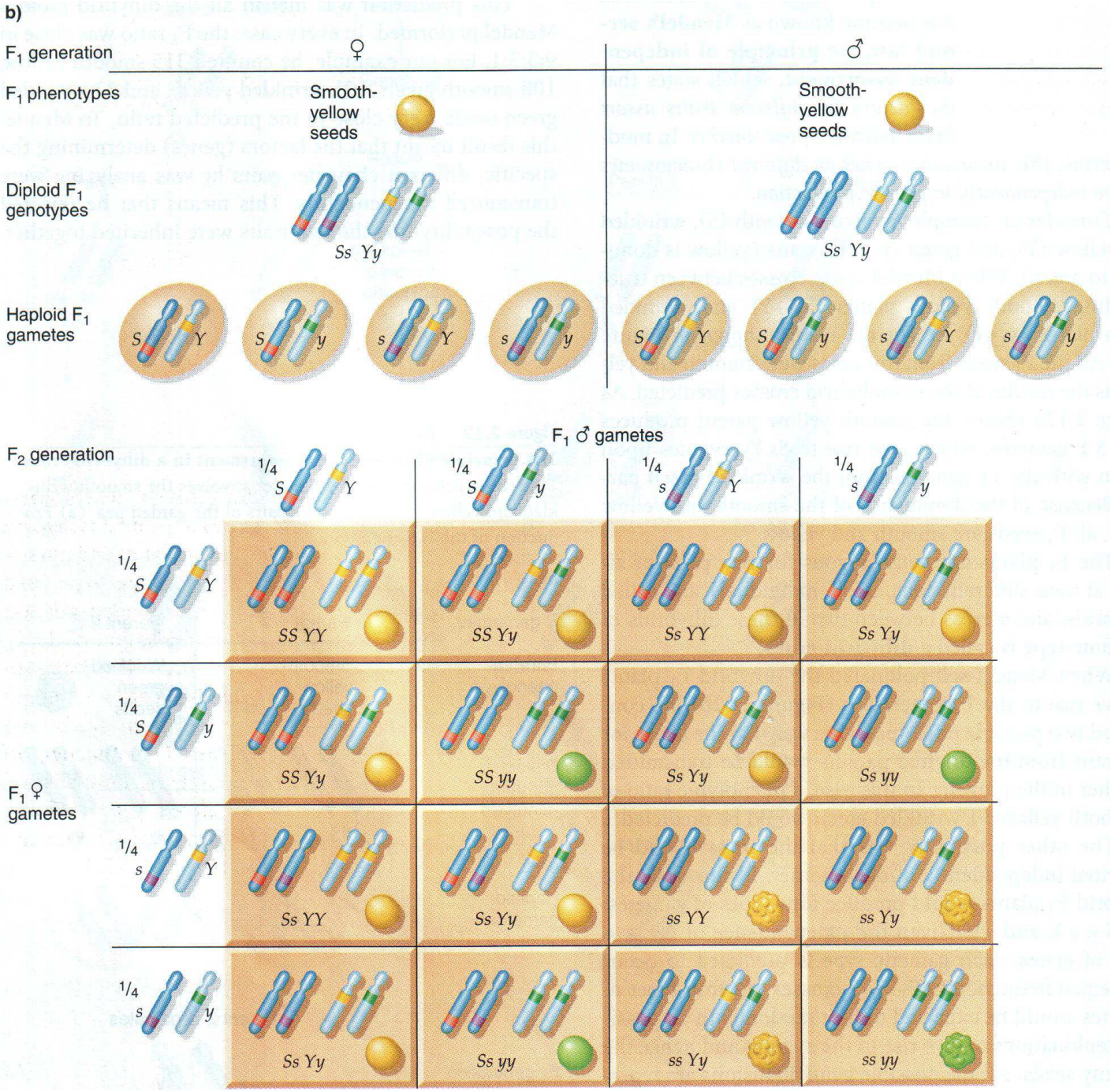

MEIOSI I
MEIOSI II
2n
Divisione riduzionale
n
n
Divisione equazionale
n

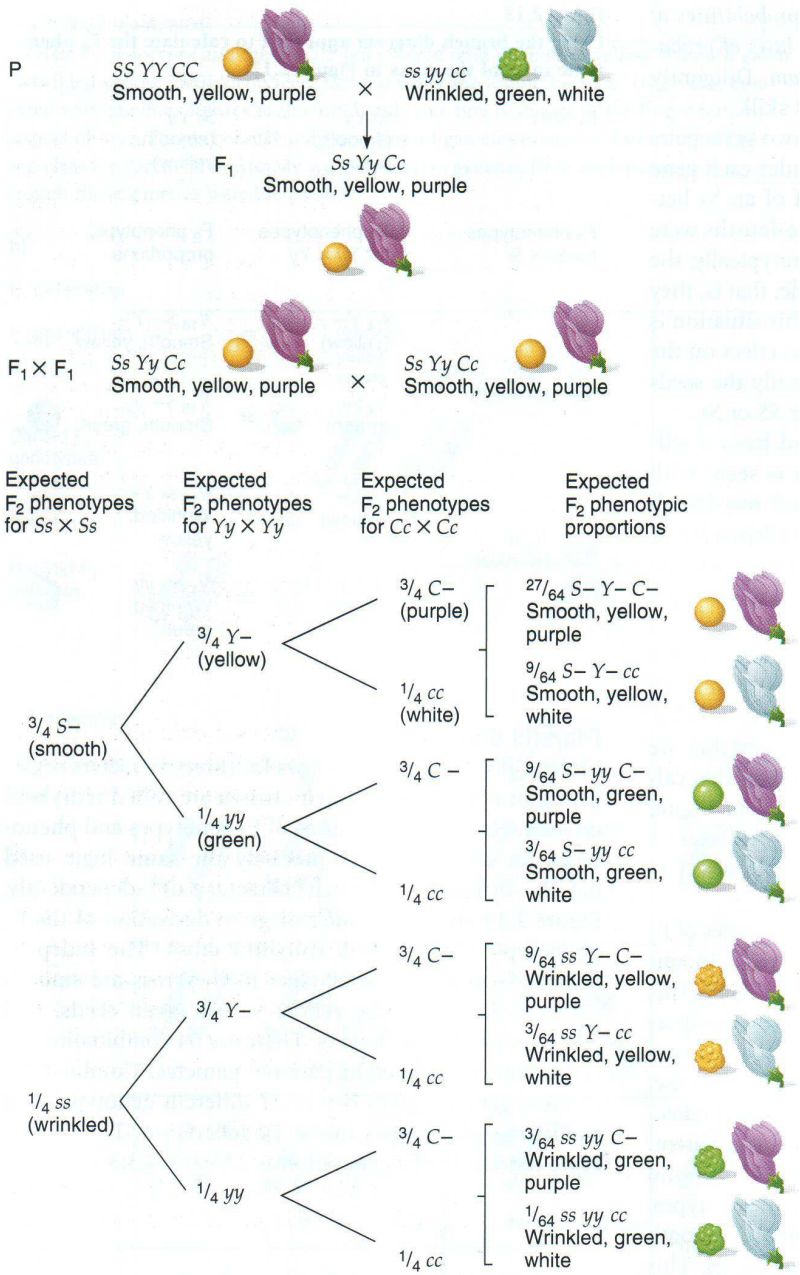
Figure 2.12

The principle of independent assortment in a dihybrid cross. This cross, actually done by Mendel, involves the smooth/wrinkled and yellow/green character pairs of the garden pea. **(a)** Production of the F_1 generation.




GENERAZIONE PARENTALE
fenotipo: giallo liscio
genotipo:YYRR
fenotipo: verde rugoso
genotipo: yyrr
50% YR
50% yr
gameti
FECONDAZIONE INCROCIATA
PRIMA GENERAZIONE FILIALE (F1)
fenotipo: giallo liscio
genotipo:YyRr
AUTOFECONDAZIONE
UOVA
SPERMI
25% YR
25% Yr
25% yR
25% yr
YYRR
YYRr
YyRR
YYrr
YyRr
Yyrr
yyRR
yyRr
yyrr
SECONDA GENERAZIONE FILIALE (F2)
GENOTIPI
FENOTIPO
RAPPORTO FENOTIPICO
YYRR YyRR, YYRr, YyRr
giallo liscio
9/16
yyRR, yyRr
verde liscio
3/16
YYrr, Yyrr
giallo rugoso
3/16
yyrr
verde rugoso
1/16


b)
F1 generation
♀
♂
F1 phenotypes
Smooth-yellow seeds
Smooth-yellow seeds
Diploid F1 genotypes
Ss Yy
Ss Yy
Haploid F1 gametes
S Y
S y
s Y
s y
S Y
S y
s Y
s y
F2 generation
F1 ♂ gametes
1/4 S Y
1/4 S y
1/4 s Y
1/4 s y
F1 ♀ gametes
1/4 S Y
SS YY
SS Yy
Ss YY
Ss Yy
1/4 S y
SS Yy
SS yy
Ss Yy
Ss yy
1/4 s Y
Ss YY
Ss Yy
ss YY
ss Yy
1/4 s y
Ss Yy
Ss yy
ss Yy
ss yy

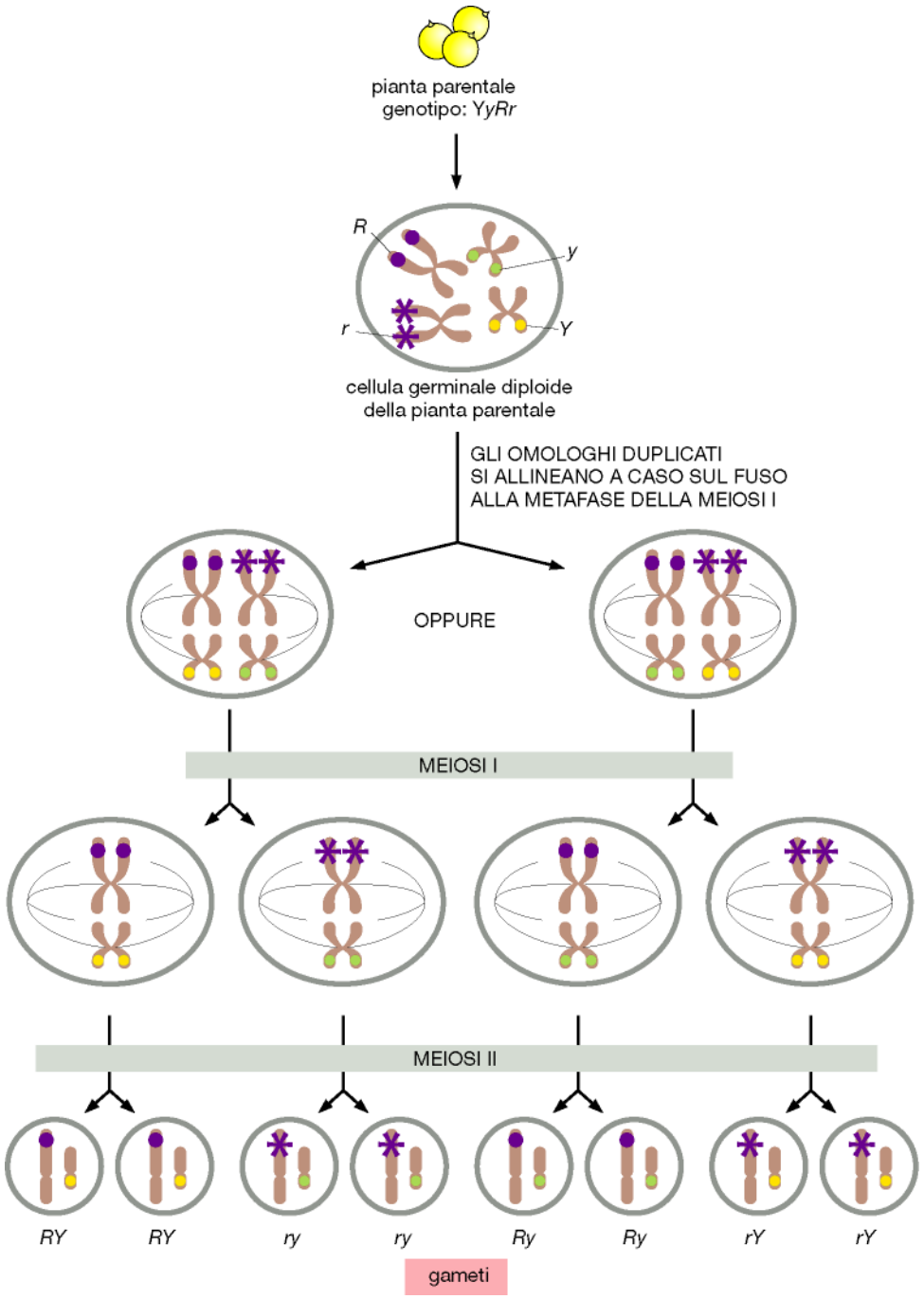

P
SS YY CC
Smooth, yellow, purple
×
ss yy cc
Wrinkled, green, white
F1
Ss Yy Cc
Smooth, yellow, purple
F1 × F1
Ss Yy Cc
Smooth, yellow, purple
×
Ss Yy Cc
Smooth, yellow, purple
Expected F2 phenotypes for Ss × Ss
Expected F2 phenotypes for Yy × Yy
Expected F2 phenotypes for Cc × Cc
Expected F2 phenotypic proportions
3/4 S– (smooth)
1/4 ss (wrinkled)
3/4 Y– (yellow)
1/4 yy (green)
3/4 Y–
1/4 yy
3/4 C– (purple)
1/4 cc (white)
3/4 C –
1/4 cc
3/4 C–
1/4 cc
3/4 C–
1/4 cc
27/64 S– Y– C– Smooth, yellow, purple
9/64 S– Y– cc Smooth, yellow, white
9/64 S– yy C– Smooth, green, purple
3/64 S– yy cc Smooth, green, white
9/64 ss Y– C– Wrinkled, yellow, purple
3/64 ss Y– cc Wrinkled, yellow, white
3/64 ss yy C– Wrinkled, green, purple
1/64 ss yy cc Wrinkled, green, white

Seconda Legge di Mendel:

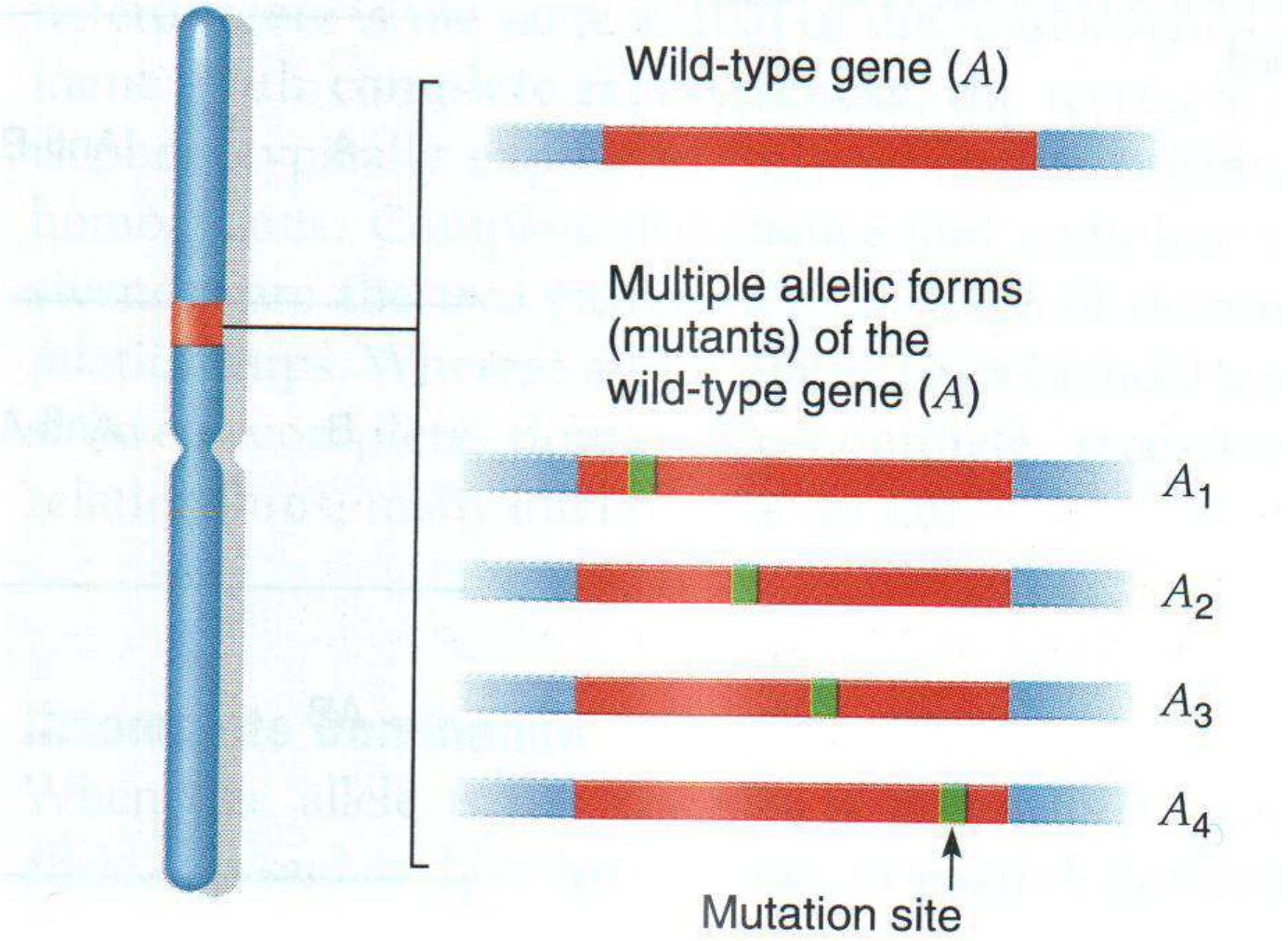
(Principio dell'Assortimento Indipendente)

Alleli di geni differenti si assortiscono indipendentemente l'uno dall'altro.

Alleli di geni localizzati su cromosomi diversi si comportano indipendentemente durante la formazione dei gameti.

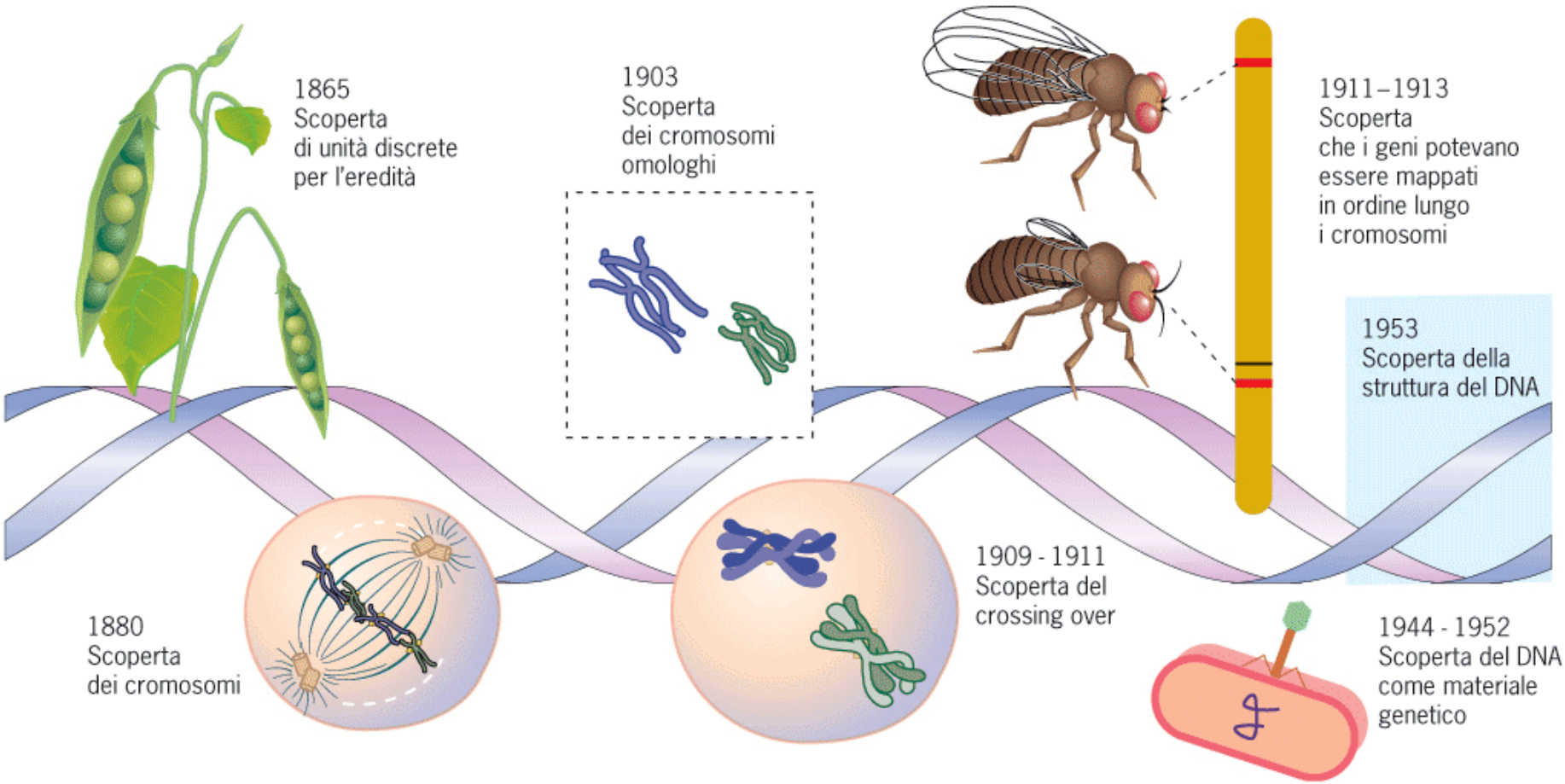

pianta parentale
genotipo: YyRr
R
y
r
Y
cellula germinale diploide
della pianta parentale
GLI OMOLOGHI DUPLICATI
SI ALLINEANO A CASO SUL FUSO
ALLA METAFASE DELLA MEIOSI I
OPPURE
MEIOSI I
MEIOSI II
RY
RY
ry
ry
Ry
Ry
rY
rY
gameti

Allelic forms of a gene.



La “riscoperta” dei principi di Mendel

•Carl Correns	mais, piselli
•Hugo de Vries	piante
•Erich von Tschermak	piselli
•William Bateson	fagiani


1865
Scoperta
di unità discrete
per l'eredità
1903
Scoperta
dei cromosomi
omologhi
1911–1913
Scoperta
che i geni potevano
essere mappati
in ordine lungo
i cromosomi
1953
Scoperta della
struttura del DNA
1880
Scoperta
dei cromosomi
1909 - 1911
Scoperta del
crossing over
1944 - 1952
Scoperta del DNA
come materiale
genetico

La Teoria Cromosomica dell'Eredità

W. Sutton e T. Boveri (1903)

La trasmissione dei cromosomi da una generazione all'altra correla con la trasmissione dei fattori mendeliani.

I fattori mendeliani (geni) sono localizzati sui cromosomi?