I Meccanismi dell’Eredità. II

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?

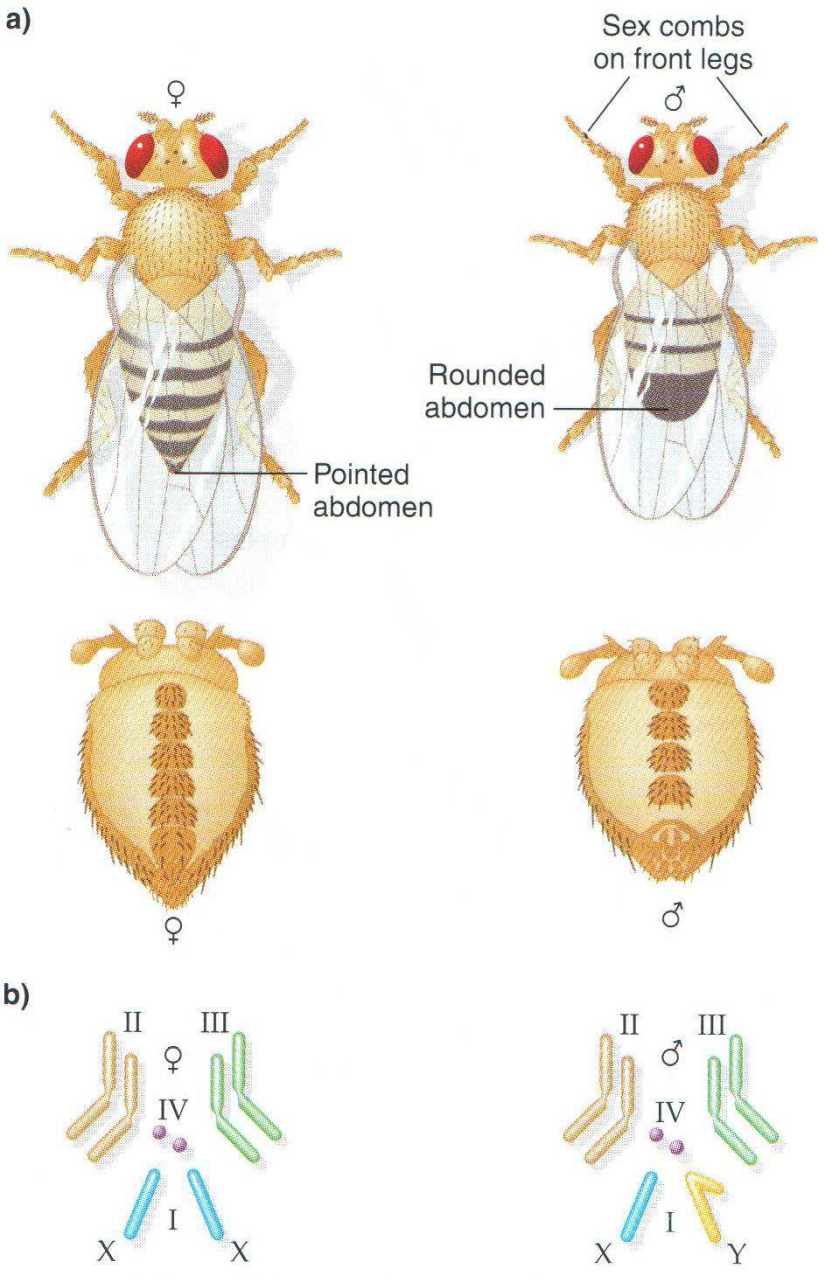
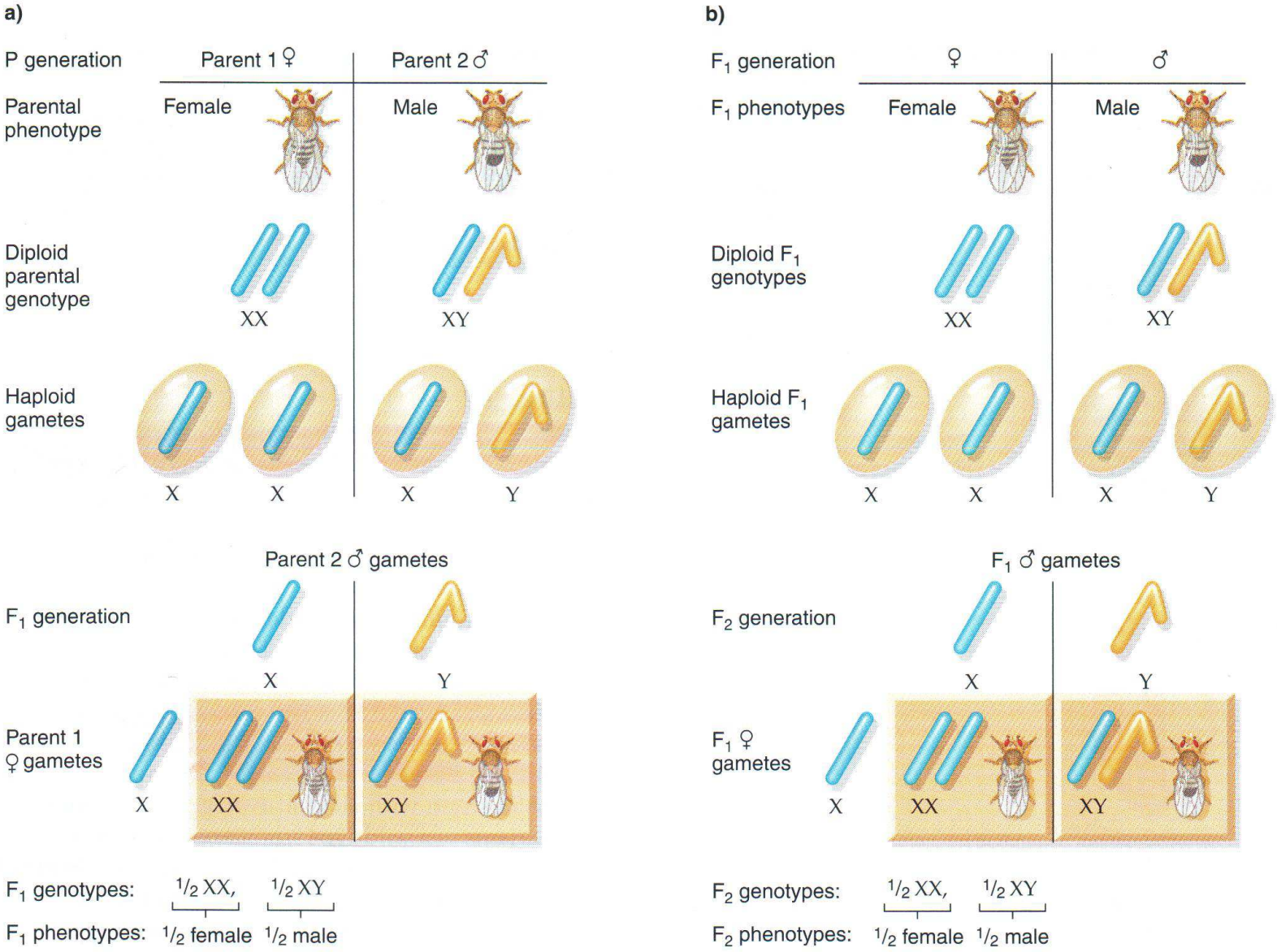
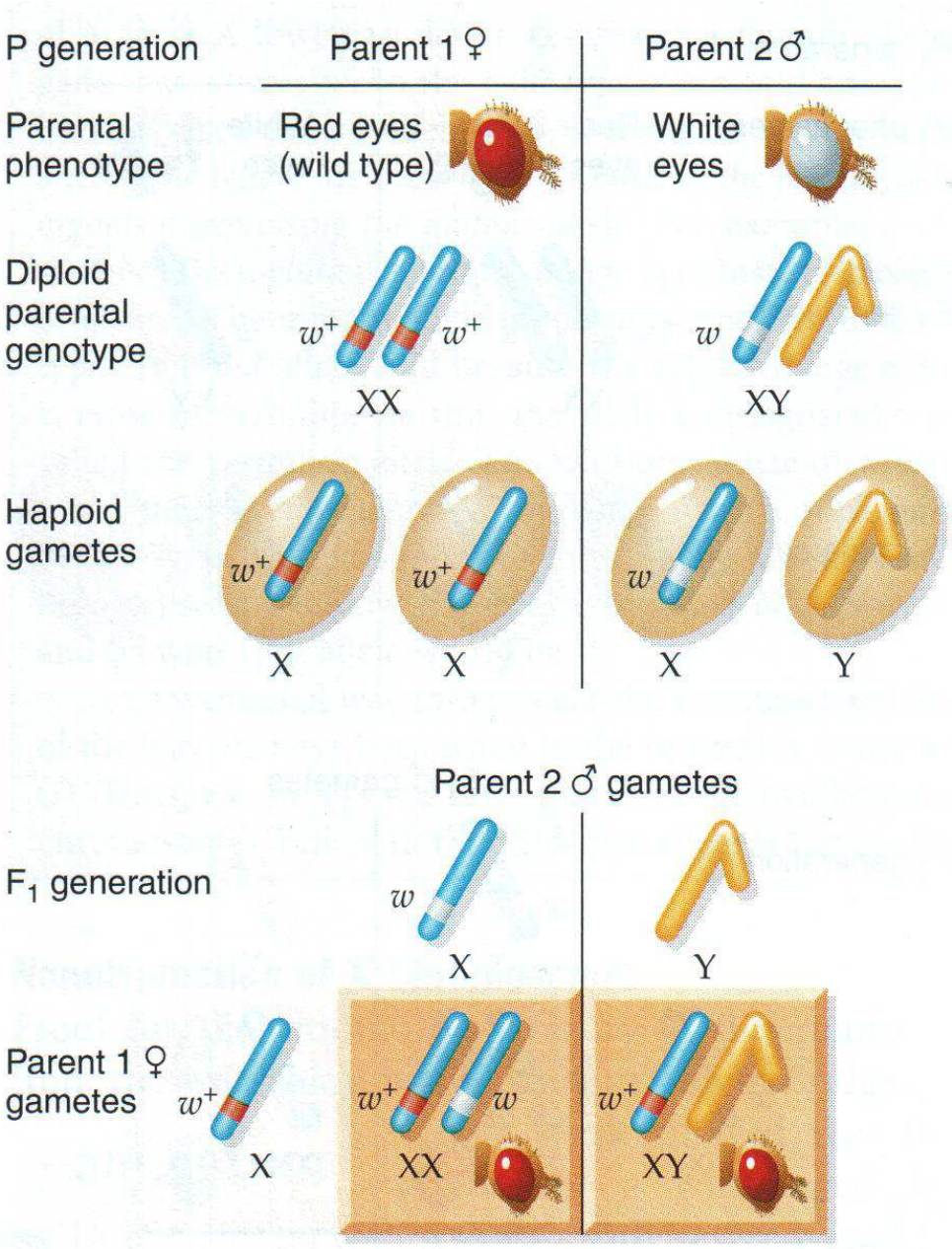

a)
Sex combs
on front legs
♀
♂
Rounded
abdomen
Pointed
abdomen
♀
♂
b)
II
III
♀
IV
I
X
X
II
III
♂
IV
I
X
Y

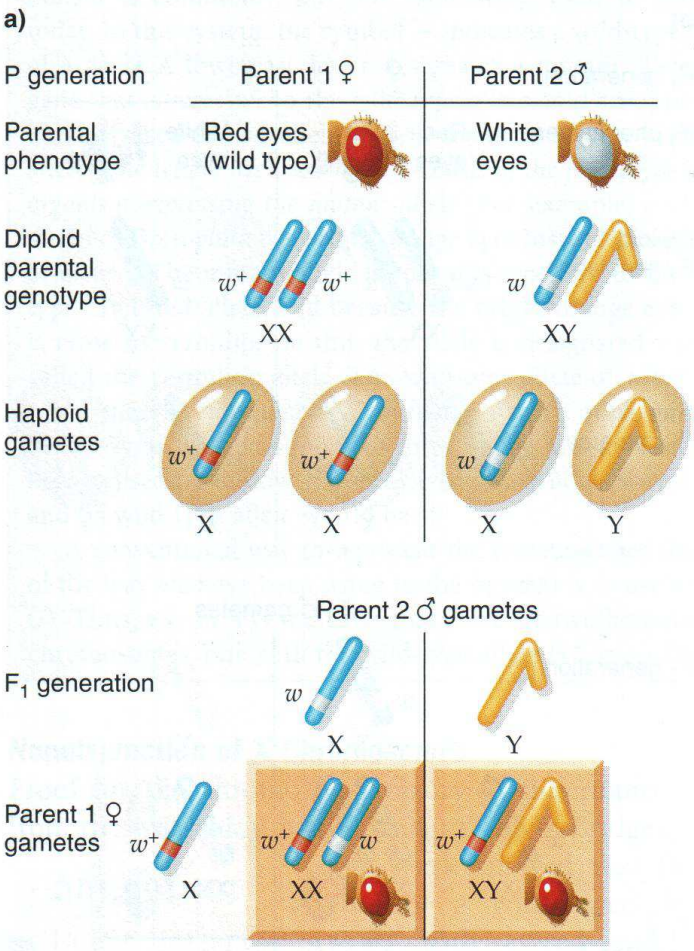
Figure 3.2

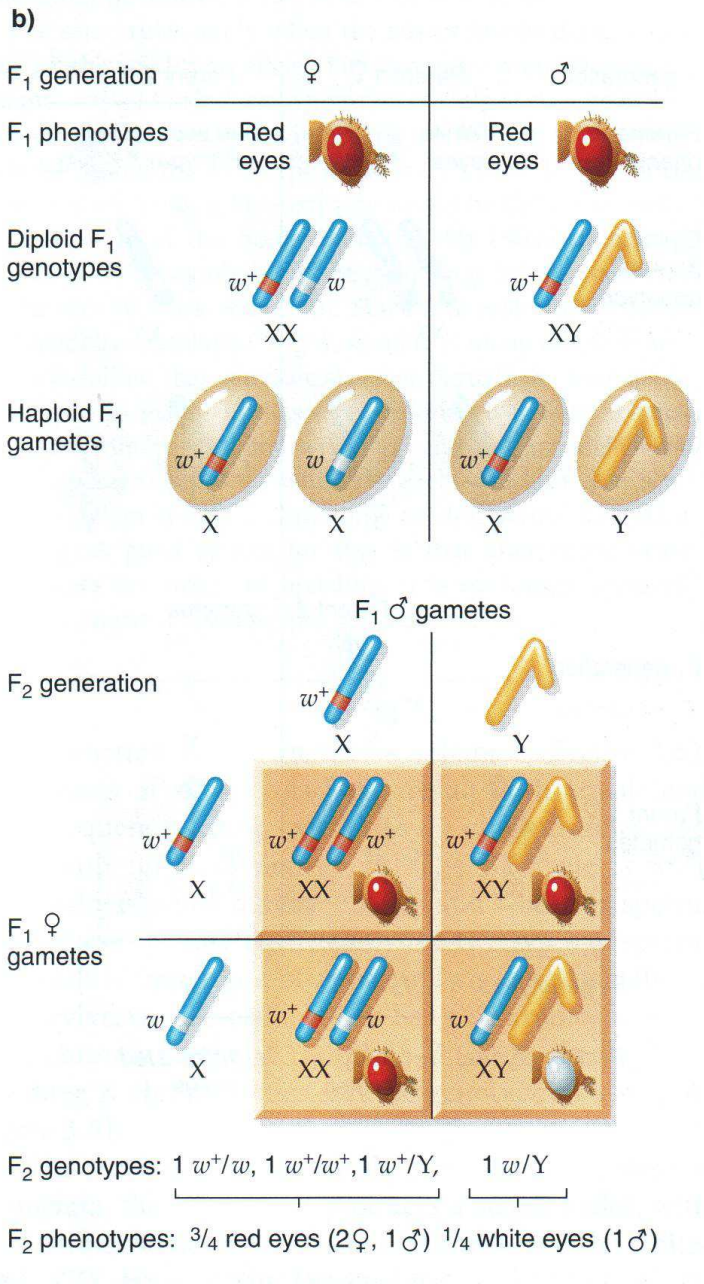
Inheritance pattern of X and Y chromosomes in organisms where the female is XX and the male is XY. (a) Production of the F_1 generation. **(b)** Production of the F_2 generation.

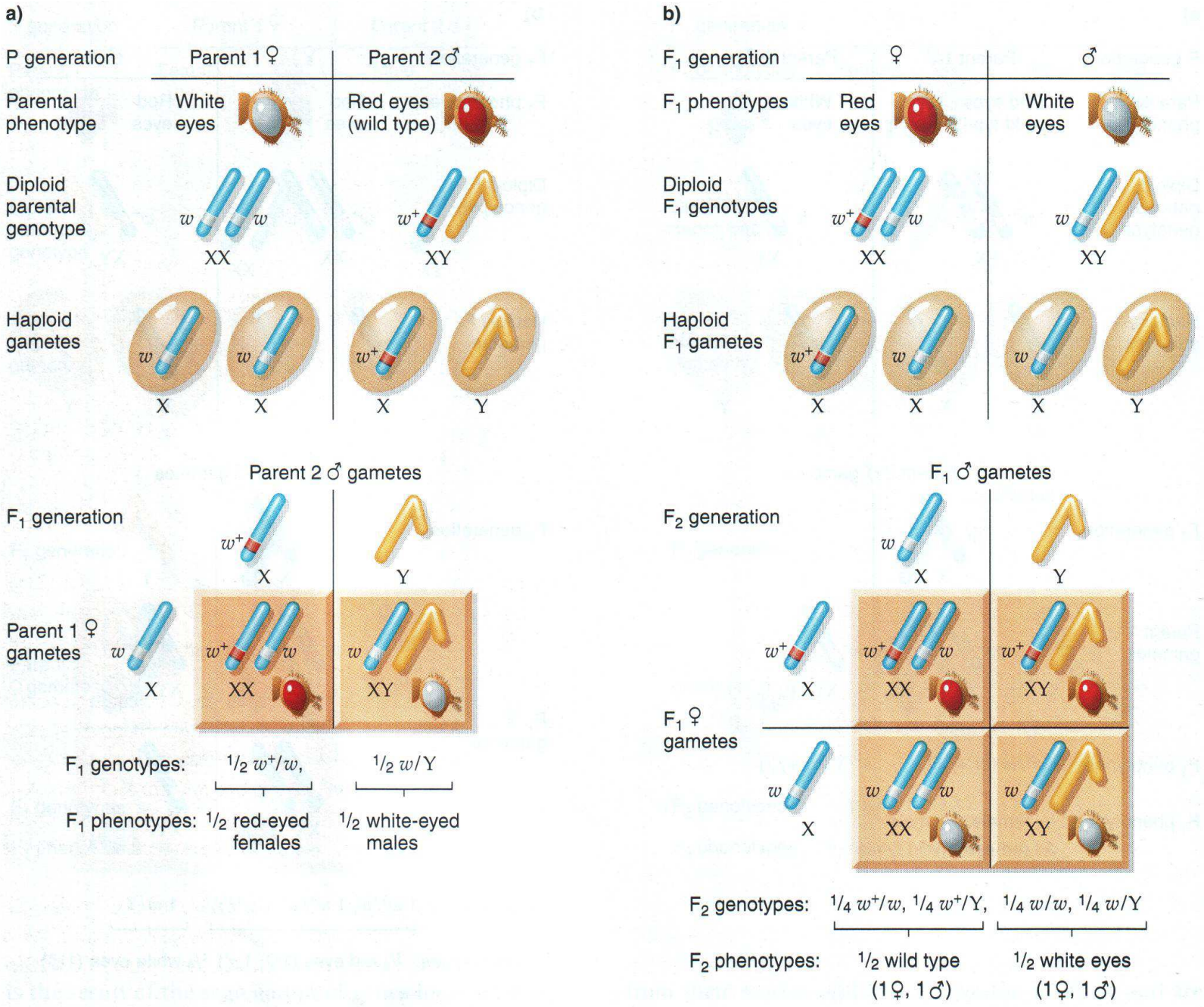


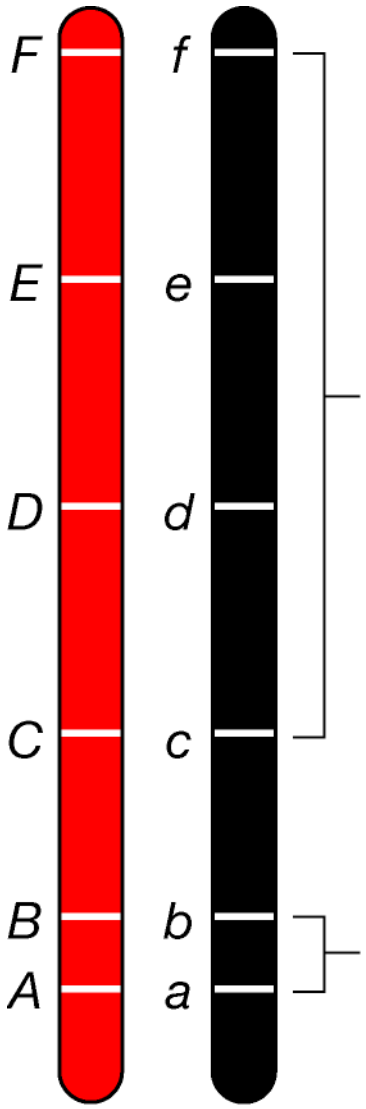


F_1 genotypes: $^1/_2$ w^+/w (or $+/w$), $^1/_2$ w^+/Y (or $+/\ $)


a)
P generation
Parent 1 ♀
Parent 2 ♂
Parental phenotype
Red eyes (wild type)
White eyes
Diploid parental genotype
w^+ w^+
XX
w
XY
Haploid gametes
w^+
w^+
w
X
X
X
Y
Parent 2 ♂ gametes
w
X
Y
F_1 generation
Parent 1 ♀ gametes
w^+
X
w^+ w
XX
w^+
XY
F_1 genotypes: 1/2 w^+/w (or +/w), 1/2 w^+/Y (or +//)
F_1 phenotypes: 1/2 female, 1/2 male
All red-eyed (wild type)


b)
F_1 generation
♀
♂
F_1 phenotypes
Red eyes
Red eyes
Diploid F_1 genotypes
w^+ w
XX
w^+
XY
Haploid F_1 gametes
w^+
w
w^+
X
X
X
Y
F_1 ♂ gametes
F_2 generation
w^+
X
Y
F_1 ♀ gametes
w^+
X
w^+ w^+
XX
w^+
XY
w
X
w^+ w
XX
w
XY
F_2 genotypes: 1 w^+/w, 1 w^+/w^+, 1 w^+/Y, 1 w/Y
F_2 phenotypes: 3/4 red eyes (2♀, 1♂) 1/4 white eyes (1♂)


a)
P generation
Parent 1 ♀
Parent 2 ♂
Parental phenotype
White eyes
Red eyes (wild type)
Diploid parental genotype
w w
XX
w+
XY
Haploid gametes
w
w
w+
X
X
X
Y
Parent 2 ♂ gametes
F1 generation
w+
X
Y
Parent 1 ♀ gametes
w
X
w+ w
XX
w
XY
F1 genotypes:
1/2 w+/w,
1/2 w/Y
F1 phenotypes:
1/2 red-eyed females
1/2 white-eyed males
b)
F1 generation
♀
♂
F1 phenotypes
Red eyes
White eyes
Diploid F1 genotypes
w+ w
XX
w
XY
Haploid F1 gametes
w+
w
w
X
X
X
Y
F1 ♂ gametes
F2 generation
w
X
Y
F1 ♀ gametes
w+
X
w+ w
XX
w+
XY
w
X
w w
XX
w
XY
F2 genotypes:
1/4 w+/w, 1/4 w+/Y,
1/4 w/w, 1/4 w/Y
F2 phenotypes:
1/2 wild type (1♀, 1♂)
1/2 white eyes (1♀, 1♂)



In media avverranno parecchi scambi nel tratto che separa questi due geni. Perciò i caratteri che essi determinano (C ed F per un cromosoma, c ed f per l'altro) non hanno maggior probabilità di essere coereditati di altri due caratteri, i cui geni siano su cromosomi separati.

È improbabile che tra questi due geni avvenga uno scambio. Perciò i caratteri che essi determinano (A e B e a e b) verranno ereditati quasi sempre insieme.

Associazione genica o Linkage

Loci localizzati su cromosomi diversi si ereditano indipendentemente (II legge di Mendel).

Loci localizzati sullo stesso cromosoma, e vicini tra loro, vengono ereditati insieme.

Distanza fisica e distanza genetica

La distanza fisica tra due loci è data dal numero di nucleotidi che li separa (es: 1 Mb = un milione di basi).

La distanza genetica è data dalla percentuale di ricombinazione, espressa in centimorgan cM. 1 cM = 1% di ricombinazione.

C’è una relazione tra distanza fisica e distanza genetica di due loci:

Un milione di basi = 1 cM

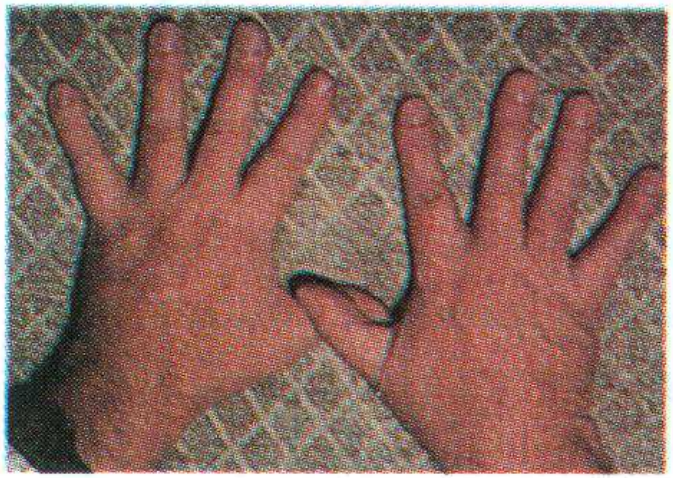
1 cM = 1% di ricombinazione

Due loci non associati, non linked, avranno un valore di cM = 50% (sono su cromosomi diversi, o ad una distanza maggiore di 50 milioni di basi)

Figure 2.15

Photographs of (a) normal hands and (b) hands exhibiting brachydactyly.

a)



b)

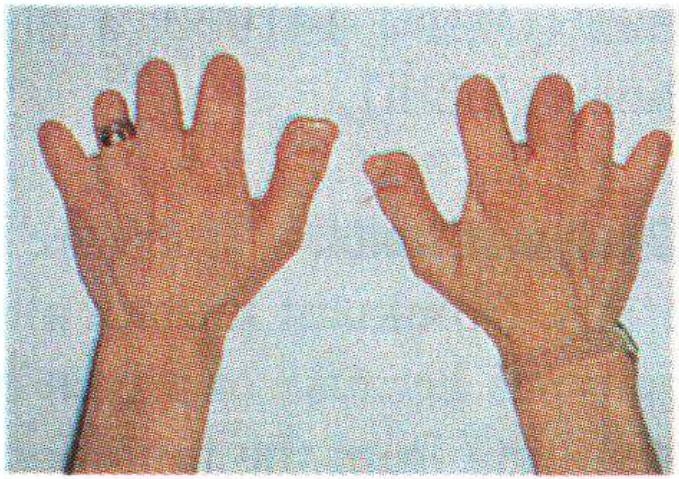


Figure 2.16

Symbols used in human pedigree analysis.

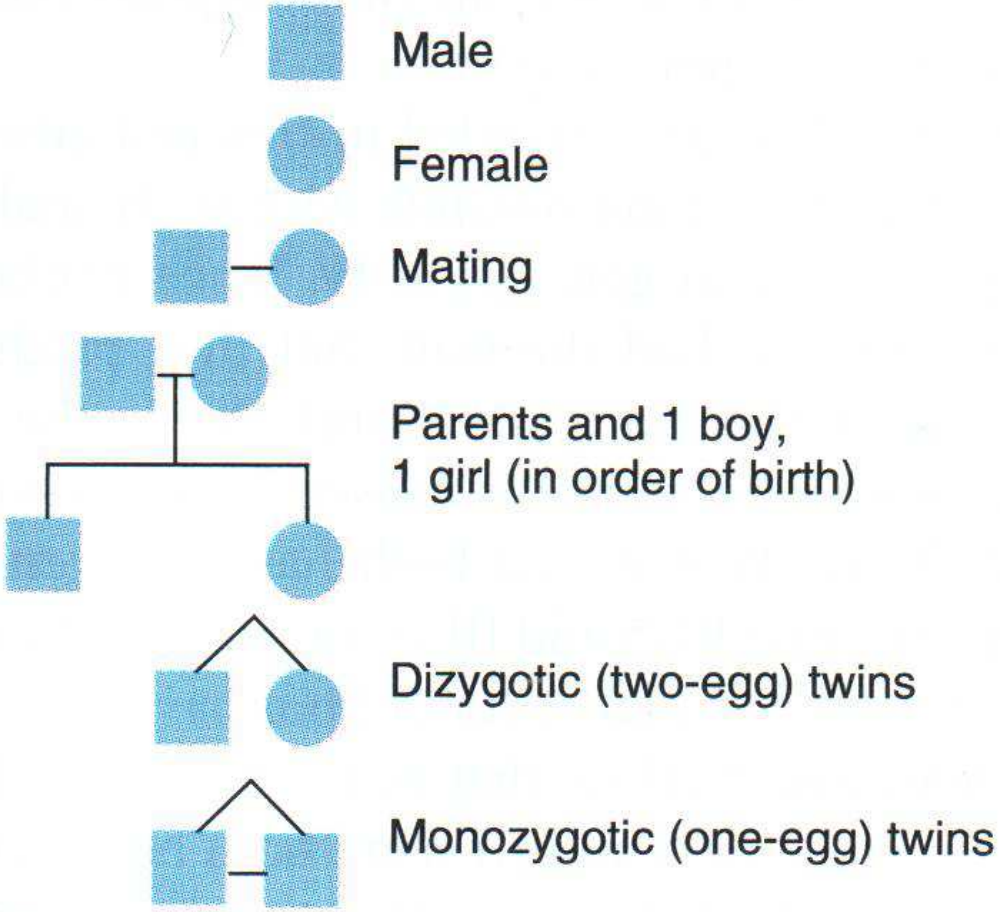
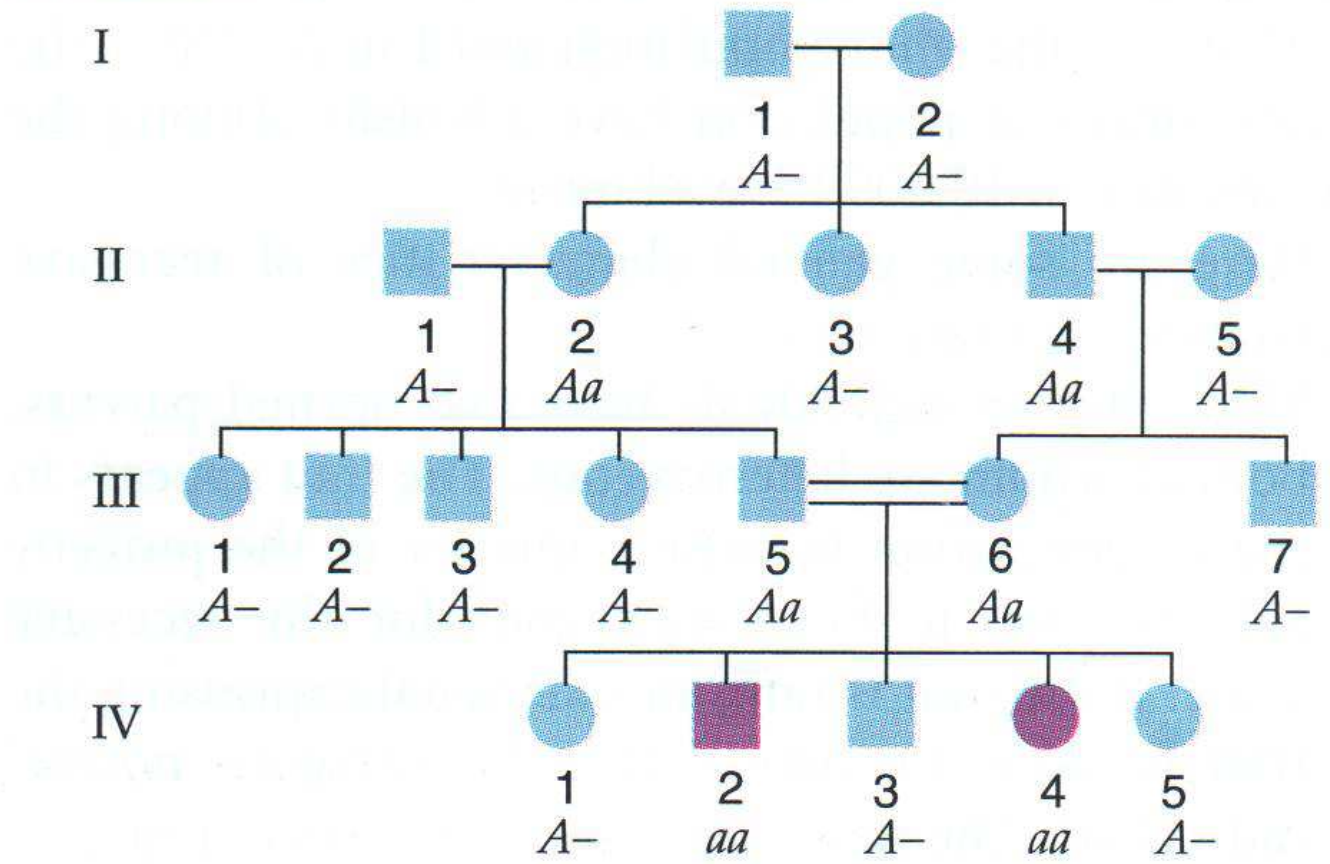


Figure 2.17

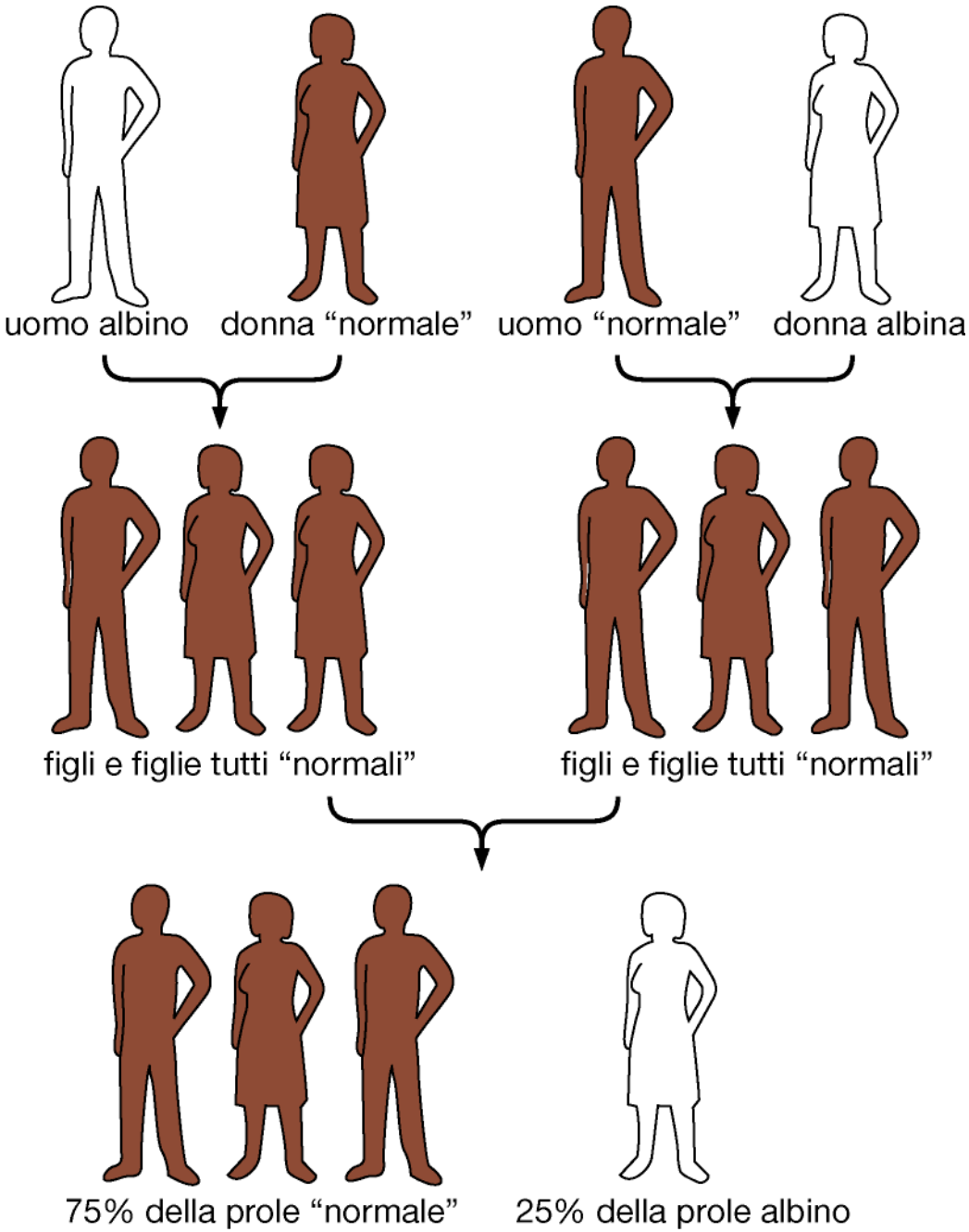
A human pedigree, illustrating the use of pedigree symbols.

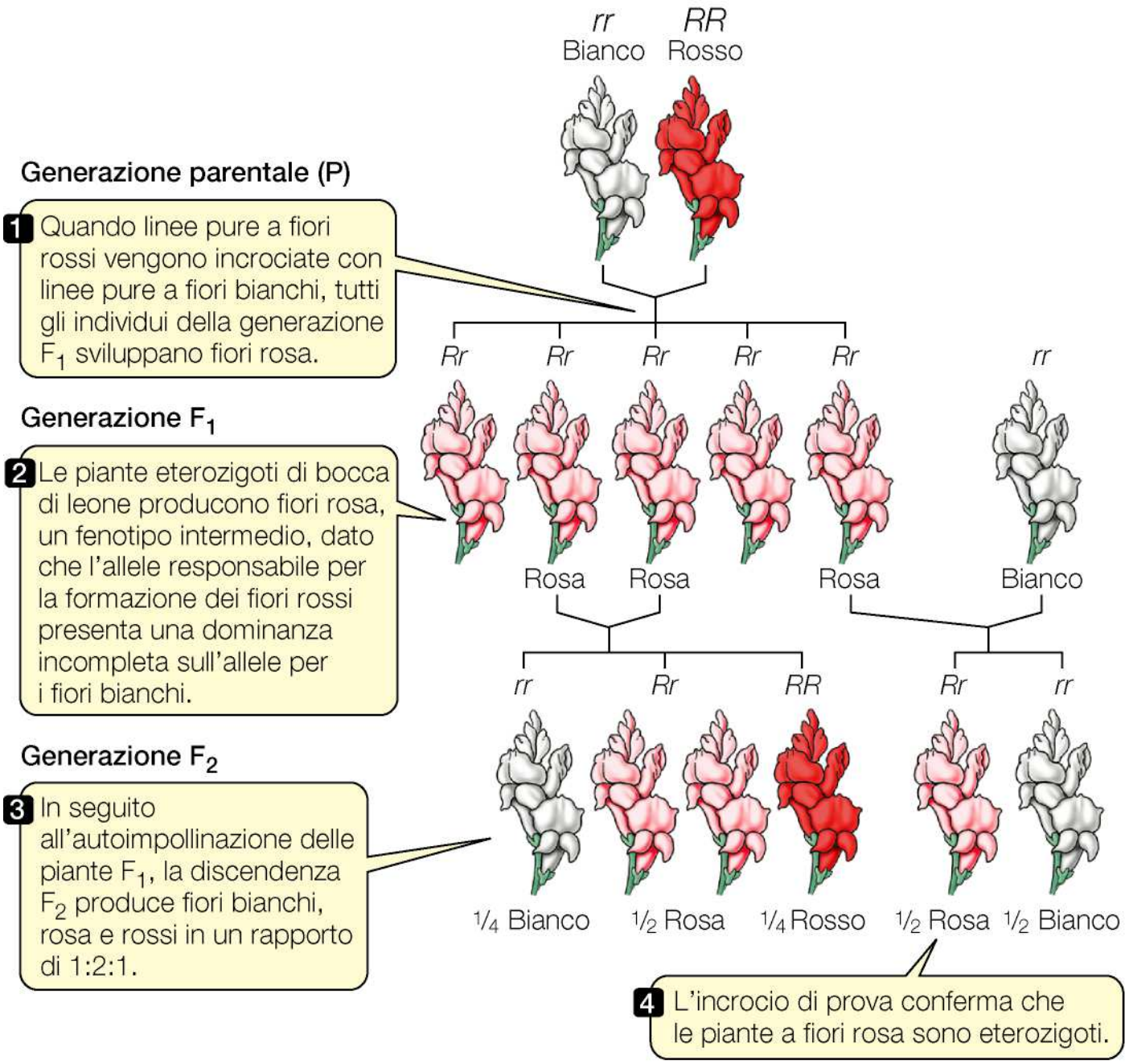


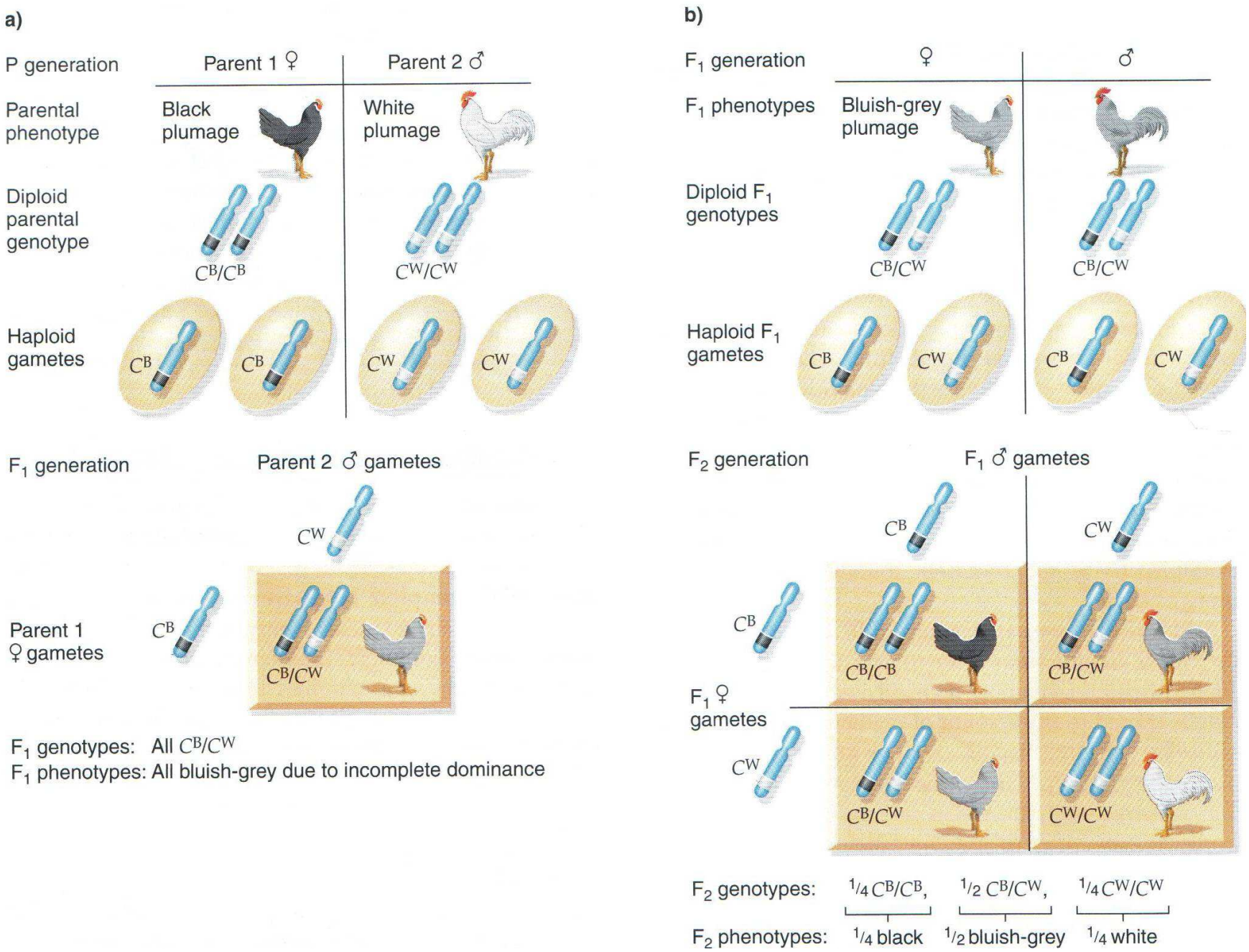
La dominanza e la recessività sono proprietà dei caratteri, **non dei geni**.

Un carattere è dominante se si manifesta nell’eterozigote, altrimenti è recessivo.

Es: anemia falciforme.

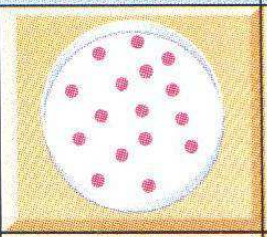
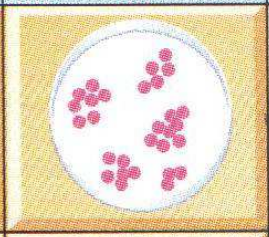
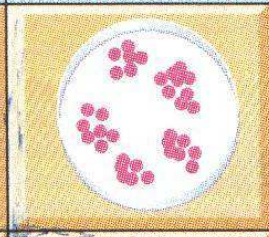
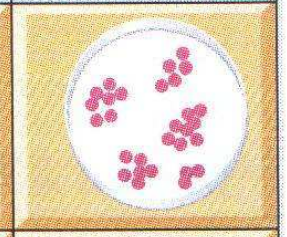
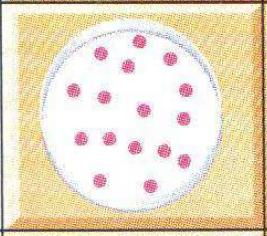
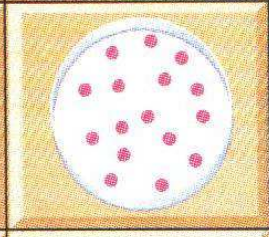
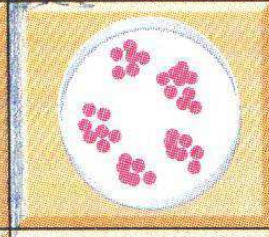
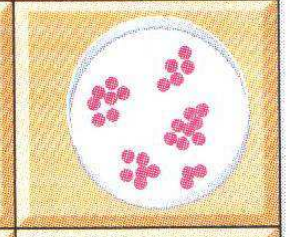
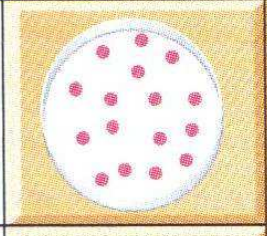
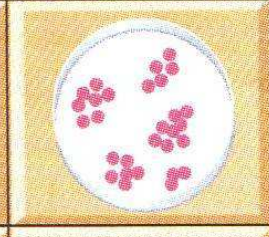
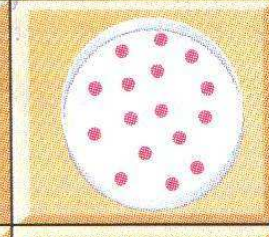
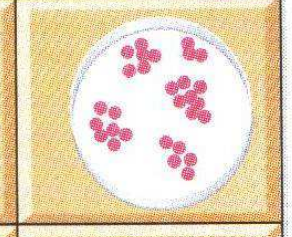
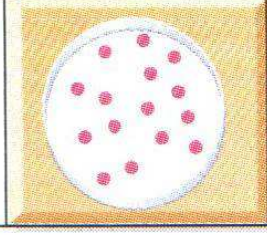
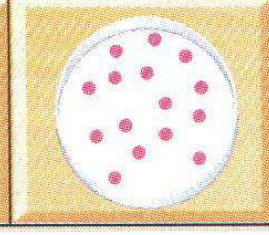
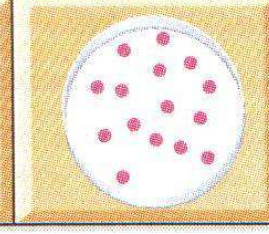
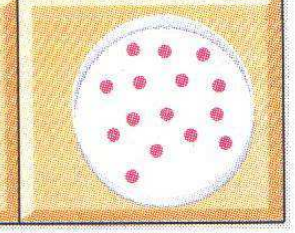

uomo albino
donna “normale”
uomo “normale”
donna albina
figli e figlie tutti “normali”
figli e figlie tutti “normali”
75% della prole “normale”
25% della prole albino


rr
Bianco
RR
Rosso
Generazione parentale (P)
1 Quando linee pure a fiori rossi vengono incrociate con linee pure a fiori bianchi, tutti gli individui della generazione F_1 sviluppano fiori rosa.
Rr
Rr
Rr
Rr
Rr
rr
Generazione F_1
2 Le piante eterozigoti di bocca di leone producono fiori rosa, un fenotipo intermedio, dato che l'allele responsabile per la formazione dei fiori rossi presenta una dominanza incompleta sull'allele per i fiori bianchi.
Rosa
Rosa
Rosa
Bianco
rr
Rr
RR
Rr
rr
Generazione F_2
3 In seguito all'autoimpollinazione delle piante F_1, la discendenza F_2 produce fiori bianchi, rosa e rossi in un rapporto di 1:2:1.
1/4 Bianco
1/2 Rosa
1/4 Rosso
1/2 Rosa
1/2 Bianco
4 L'incrocio di prova conferma che le piante a fiori rosa sono eterozigoti.


a)
P generation
Parent 1 ♀
Parent 2 ♂
Parental phenotype
Black plumage
White plumage
Diploid parental genotype
C^B/C^B
C^W/C^W
Haploid gametes
C^B
C^B
C^W
C^W
F_1 generation
Parent 2 ♂ gametes
C^W
Parent 1 ♀ gametes
C^B
C^B/C^W
F_1 genotypes: All C^B/C^W
F_1 phenotypes: All bluish-grey due to incomplete dominance
b)
F_1 generation
♀
♂
F_1 phenotypes
Bluish-grey plumage
Diploid F_1 genotypes
C^B/C^W
C^B/C^W
Haploid F_1 gametes
C^B
C^W
C^B
C^W
F_2 generation
F_1 ♂ gametes
C^B
C^W
F_1 ♀ gametes
C^B
C^W
C^B/C^B
C^B/C^W
C^B/C^W
C^W/C^W
F_2 genotypes: $^1/_4\ C^B/C^B$, $^1/_2\ C^B/C^W$, $^1/_4\ C^W/C^W$
F_2 phenotypes: $^1/_4$ black $^1/_2$ bluish-grey $^1/_4$ white

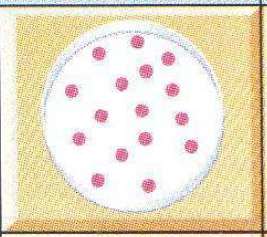
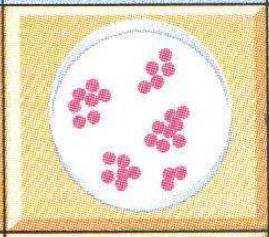
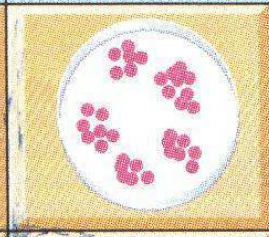
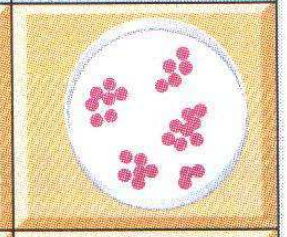
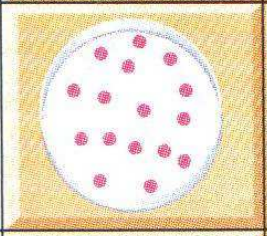
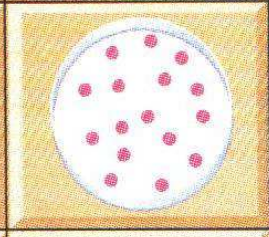
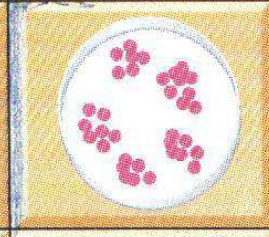
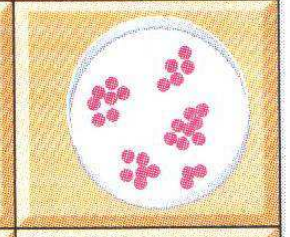
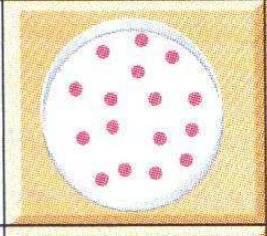
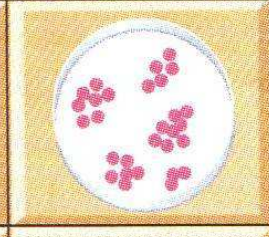
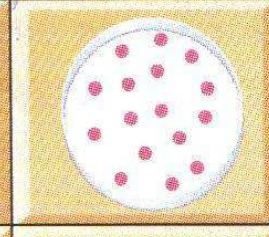
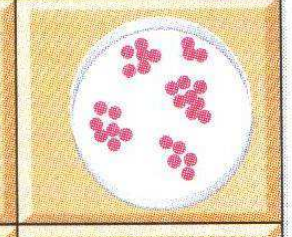
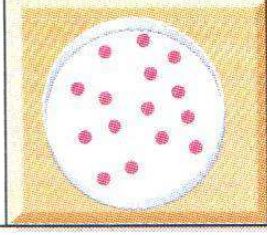
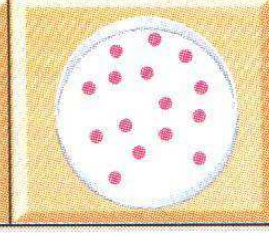
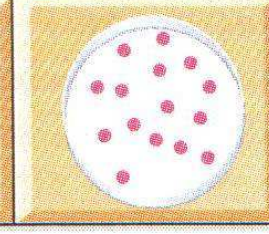
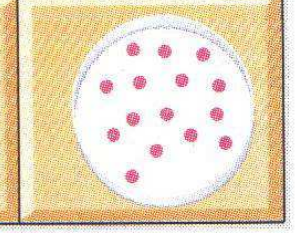
Dominanza incompleta il fenotipo dell'eterozigote è intermedio rispetto a quello degli omozigoti

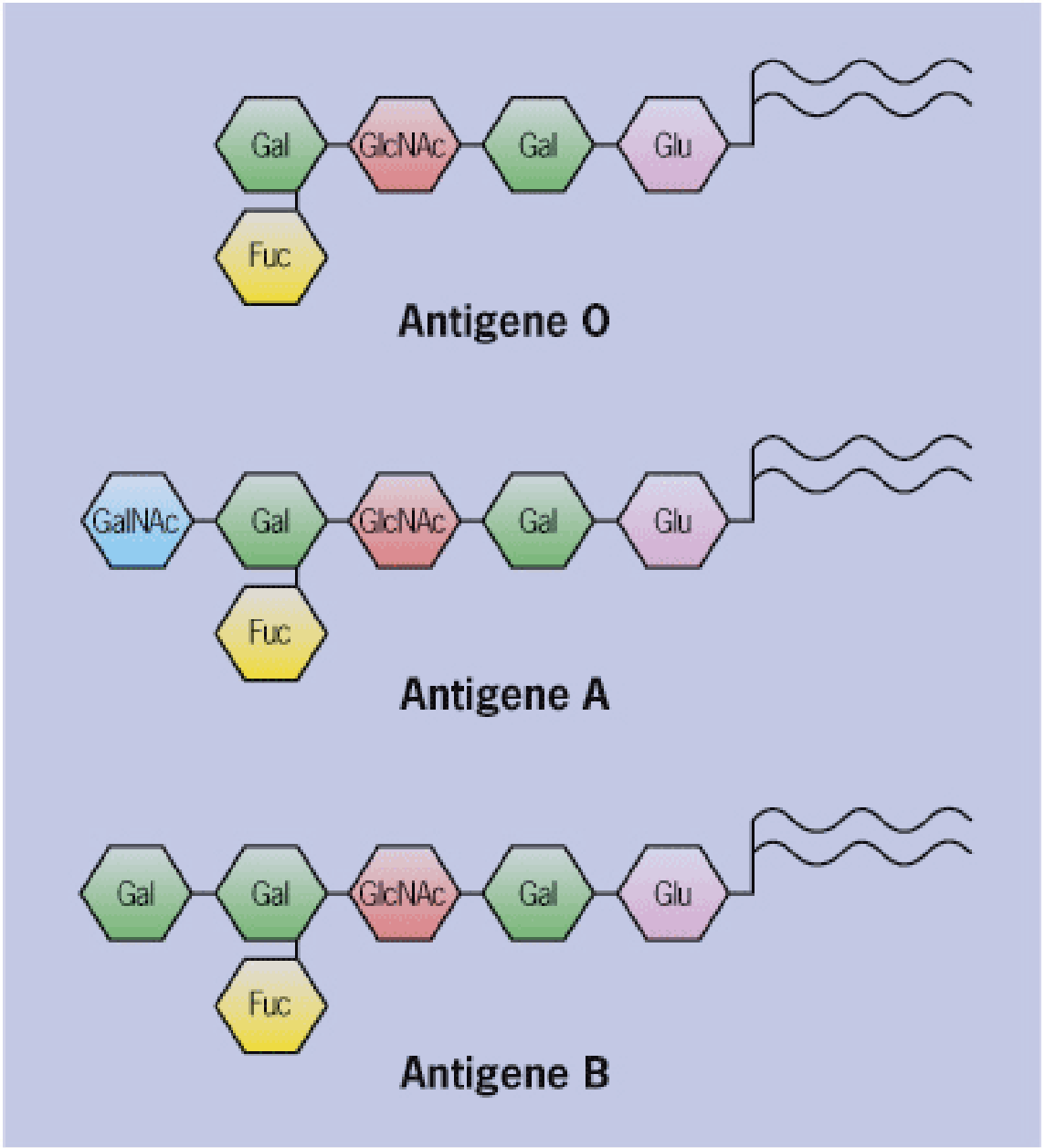
Co-dominanza l'eterozigote mostra i fenotipi di **entrambi** gli omozigoti

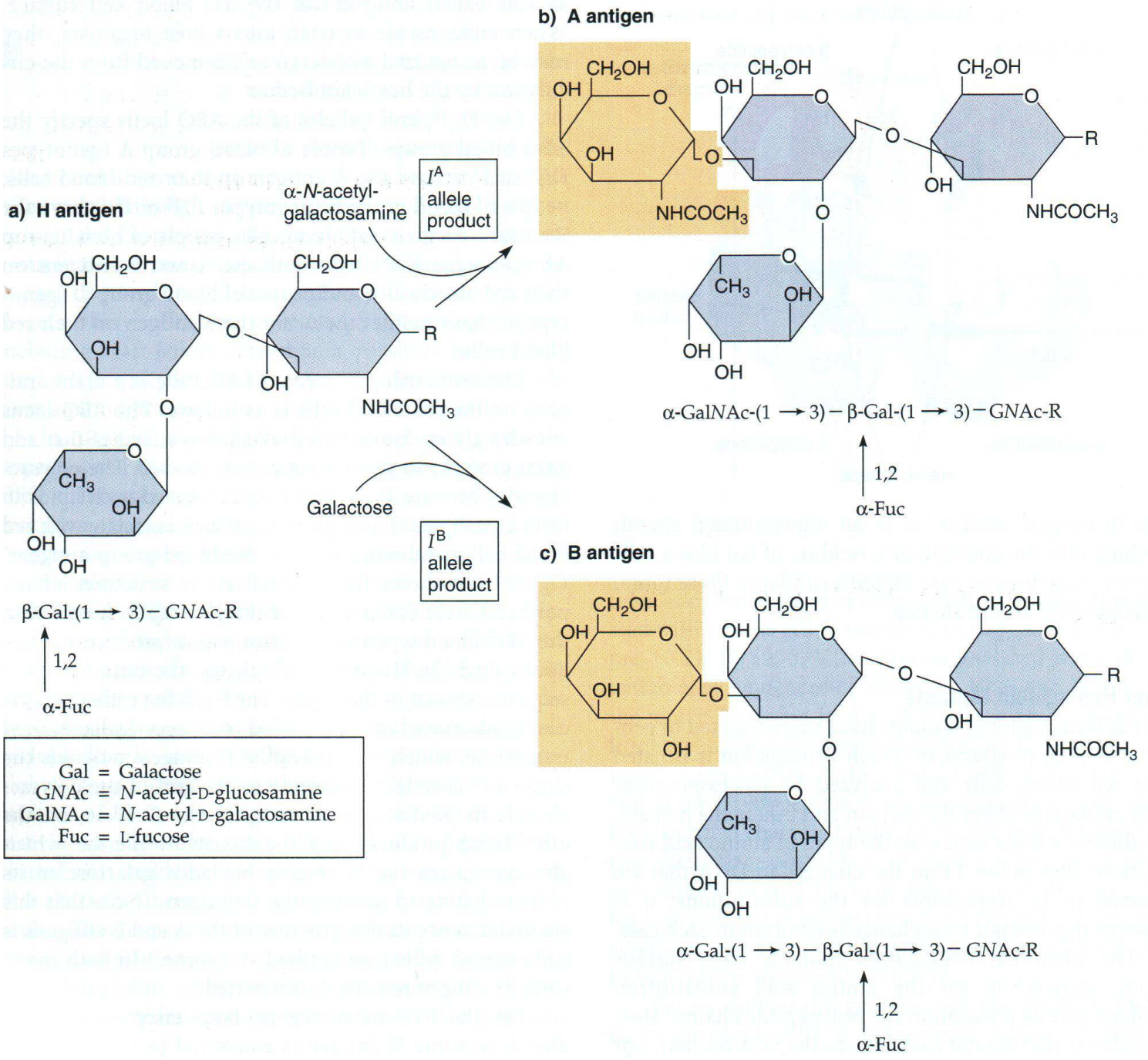
Serum from blood type	Antibodies present in serum	Cells from blood type			
		O	A	B	AB
O	Anti-A Anti-B				
A	Anti-B				
B	Anti-A				
AB	—				

Nella co-dominanza, entrambi gli alleli presenti nell’eterozigote esprimono un prodotto genico. Per cui, spesso si osservano i fenotipi degli omozigoti.

Nella dominanza incompleta, l’eterozigote esprime il prodotto di un allele soltanto. In questi casi la dose dimezzata di prodotto rispetto all’omozigote può non essere sufficiente.

Serum from blood type	Antibodies present in serum	Cells from blood type			
		O	A	B	AB
O	Anti-A Anti-B				
A	Anti-B				
B	Anti-A				
AB	—				

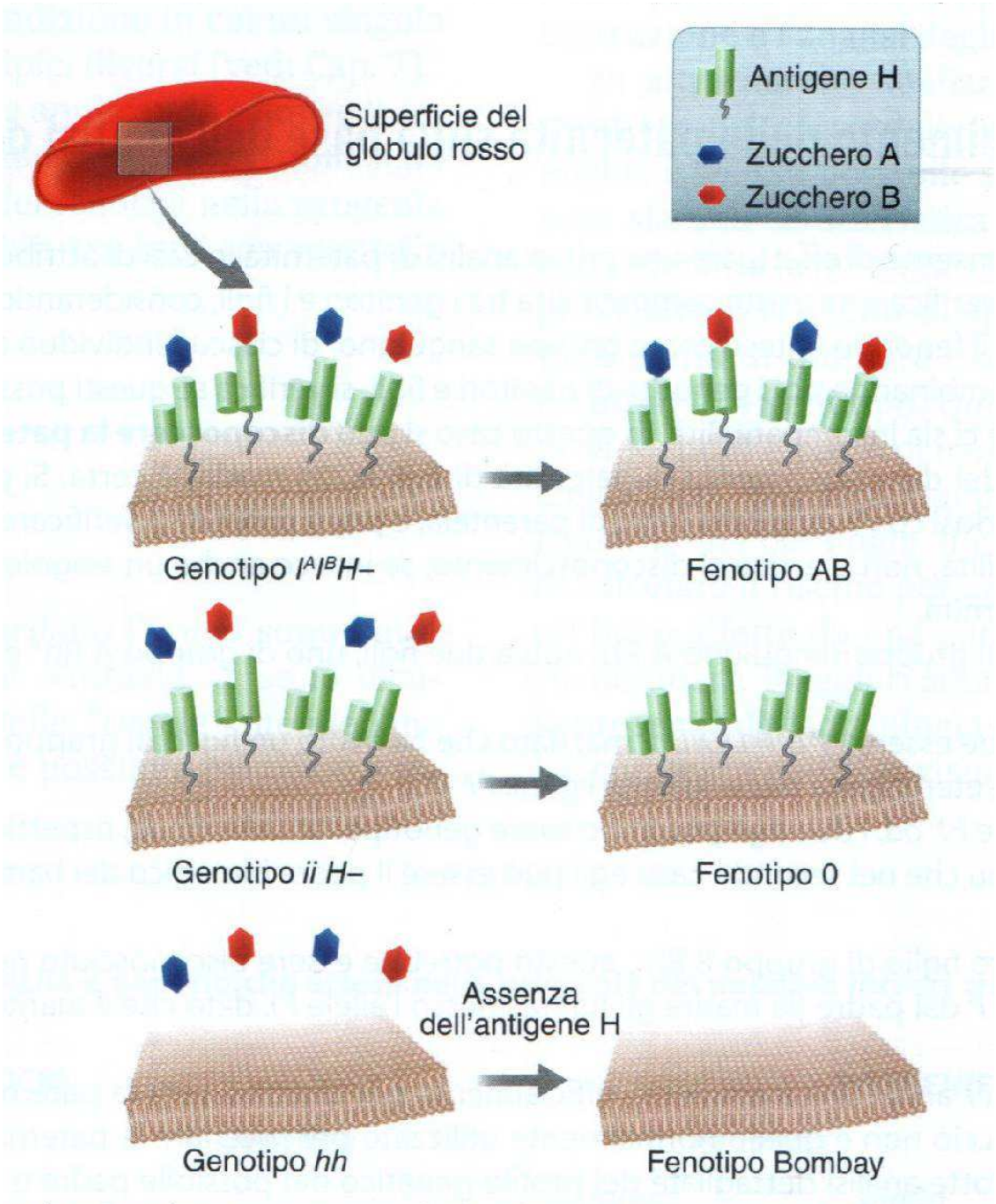

Gal
GlcNAc
Gal
Glu
Fuc
Antigene O
GalNAc
Gal
GlcNAc
Gal
Glu
Fuc
Antigene A
Gal
Gal
GlcNAc
Gal
Glu
Fuc
Antigene B


a) H antigen
α-N-acetyl-galactosamine
I^A allele product
Galactose
I^B allele product
β-Gal-(1 → 3)– GNAc-R
1,2
α-Fuc
b) A antigen
α-GalNAc-(1 → 3)– β-Gal-(1 → 3)–GNAc-R
1,2
α-Fuc
c) B antigen
α-Gal-(1 → 3)– β-Gal-(1 → 3)– GNAc-R
1,2
α-Fuc
Gal = Galactose
GNAc = N-acetyl-D-glucosamine
GalNAc = N-acetyl-D-galactosamine
Fuc = L-fucose

La **epistasi** è una forma di interazione genica in cui un gene maschera l'espressione fenotipica di un altro.

Un gene che maschera l'espressione di un altro gene è definito **epistatico**.

Un gene la cui espressione è mascherata è definito **ipostatico**.


Antigene H
Zucchero A
Zucchero B
Superficie del globulo rosso
Genotipo I^AI^BH-
Fenotipo AB
Genotipo *ii H–*
Fenotipo 0
Assenza dell'antigene H
Genotipo *hh*
Fenotipo Bombay

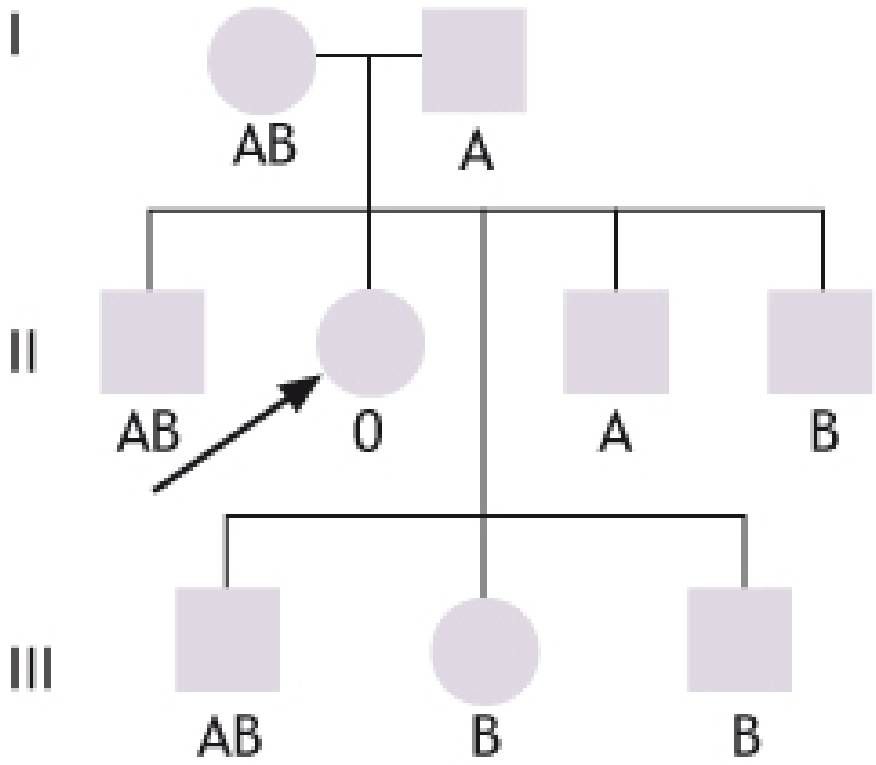


Figura 12.42 Fenotipo Bombay. Albero genealogico in cui una donna (II.2) mostra il fenotipo Bombay. Fenotipicamente appare di gruppo 0, possiede la sostanza H incompleta perché l'allele recessivo *h*, in *omozigosi*, impedisce la sintesi di una fucosiltransferasi funzionante.

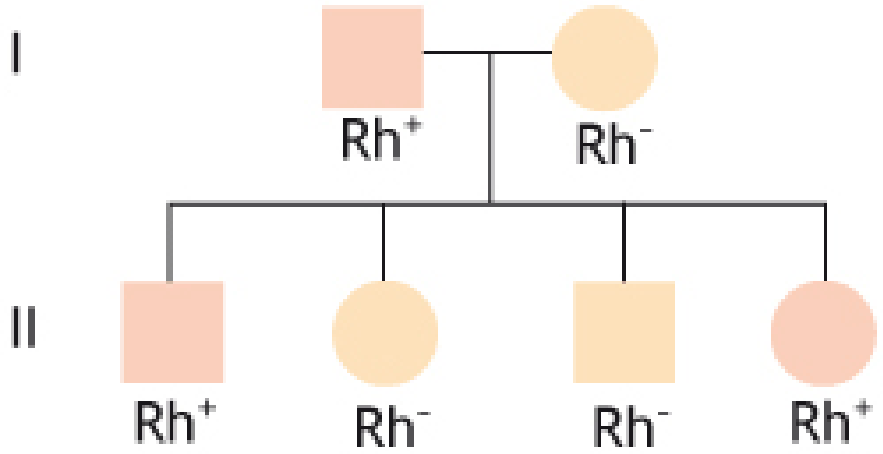


Figura 12.43 Condizione di incompatibilità materno-fetale da Rh. In quest'albero genealogico la situazione risulta tecnicamente rischiosa per la figlia II.4.