

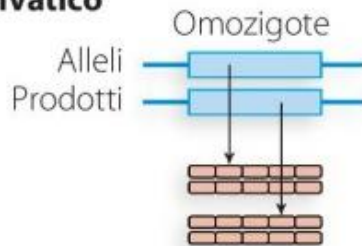
# Estensioni della genetica mendeliana

Mendel proseguì le sue ricerche su altre piante per ottenere conferme alle sue leggi, ma trovò tante e tali contraddizioni che (si dice) cadde in depressione, abbandonò la ricerca e si mise a curare l'amministrazione del convento...

Solo diverso tempo dopo si scoprì che queste contraddizioni solo APPARENTEMENTE erano incompatibili con i risultati ottenuti sul *Pisum sativum*.

# Basi molecolari della dominanza e mutazioni

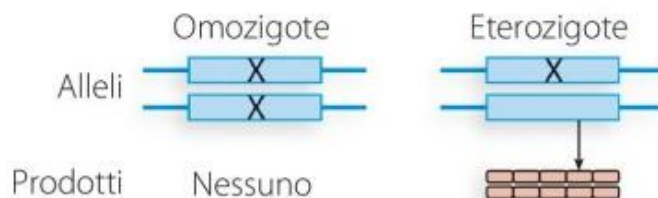
## (a) Tipo selvatico



**Figura 4.1** Conseguenze funzionali delle mutazioni. (a) Tipo selvatico. (b), (c) e (d) mutazioni a perdita di funzione. (e) e (f) mutazioni a guadagno di funzione.

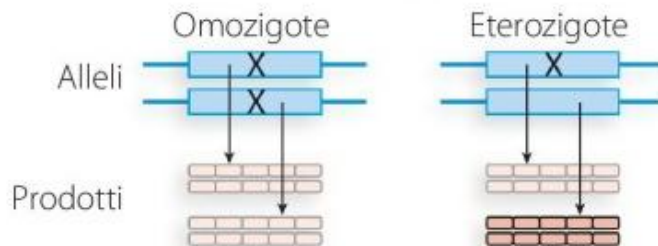
L'espressione dei prodotti di alleli di tipo selvatico produce il fenotipo selvatico.

## (b) Perdita di funzione: mutazione amorfica/nulla



L'allele nullo non produce alcun prodotto funzionale. Gli organismi omozigoti nulli hanno fenotipo mutante (amorfico) a causa dell'assenza di prodotto genico. Organismi eterozigoti producono prodotto genico meno funzionale di organismi omozigoti di tipo selvatico e possono avere fenotipo mutante. Vedete il testo per la discussione sulle differenze tra mutazioni dominanti e recessive.

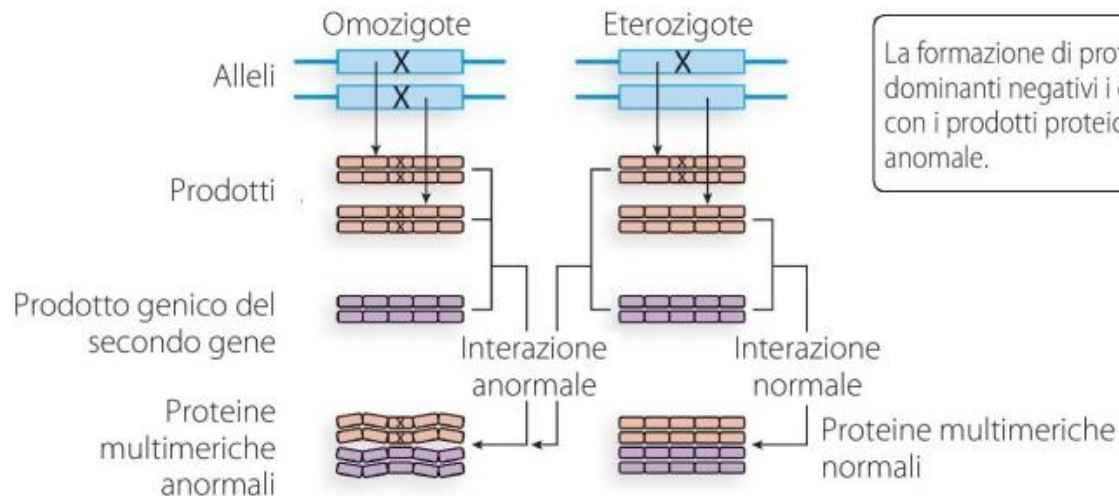
## (c) Perdita di funzione: mutazione leaky/ipomorfica



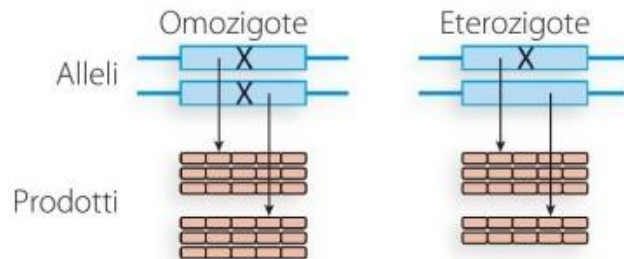
Gli alleli mutanti ipomorfici producono una piccola quantità di prodotto genico di tipo selvatico. Gli organismi omozigoti hanno un fenotipo mutante (ipomorfico). Gli organismi eterozigoti possono essere anche mutanti.

# Basi molecolari della dominanza e mutazioni

## (d) **Perdita di funzione:** mutazione dominante negativa

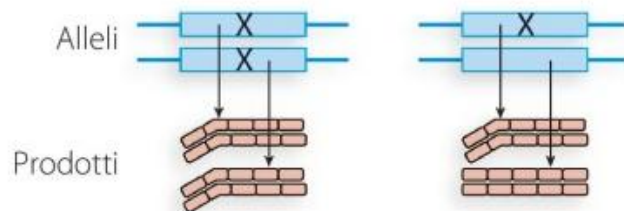


## (e) **Guadagno di funzione:** mutazione ipermorfica



Un'eccessiva espressione del prodotto genico porta ad un'eccessiva attività del gene. Il fenotipo mutante può essere più grave, o letale, nel genotipo omozigote che in quello eterozigote.

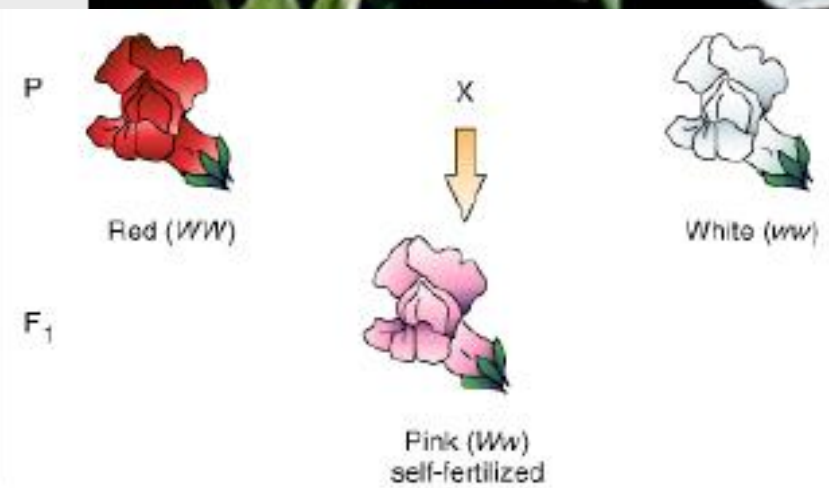
## (f) **Guadagno di funzione:** mutazione neomorfica



L'allele mutante ha una nuova funzione che produce un fenotipo mutante in organismi omozigoti ed eterozigoti e può essere più grave in organismi omozigoti.

# Variazioni della dominanza

In molte piante è possibile trovare delle varianti a fiori colorati e delle varianti a fiori bianchi. Tra queste la bocca di leone e la *Mirabilis jalapa* (bella di notte). In queste piante, incrociare individui a fiori rossi con individui a fiori bianchi dà risultati inattesi...



|                                                                                    | <u>F<sub>2</sub> Phenotype</u> | <u>Observed<br/>number</u> | <u>Expected<br/>number</u>                  |
|------------------------------------------------------------------------------------|--------------------------------|----------------------------|---------------------------------------------|
|   | Red                            | 62                         | $(1/4) \times 250 = 62.5$                   |
|   | Pink                           | 131                        | $(1/2) \times 250 = 125$                    |
|  | White                          | <u>57</u>                  | <u><math>(1/4) \times 250 = 62.5</math></u> |
|                                                                                    | <b>Total</b>                   | <b>250</b>                 | <b>250</b>                                  |

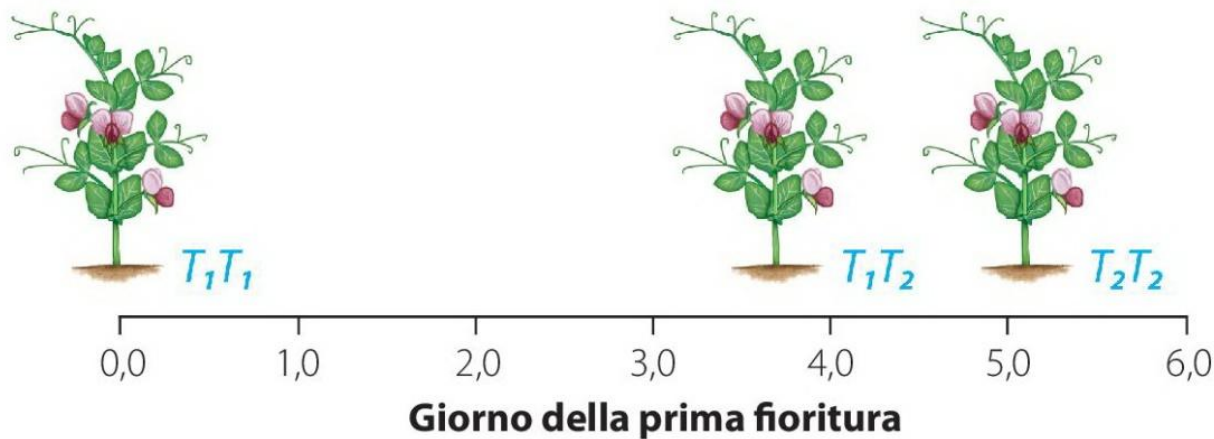
ANIMAZIONE

Completa  
corrispondenza tra  
genotipi e fenotipi: gli  
eterozigoti  $A/a$  sono  
facilmente distinguibili  
dagli individui  $A/A$ .

Quando il fenotipo  
degli eterozigoti è  
**intermedio** tra quello  
dei genitori:  
**dominanza  
incompleta.**

# Dominanza incompleta

(a)



(b)

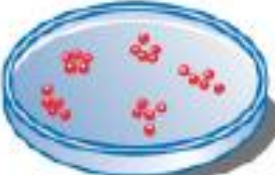
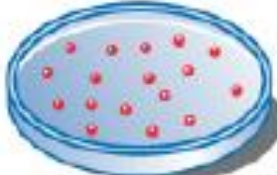
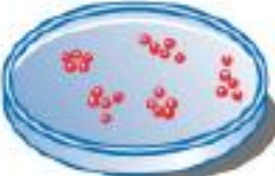
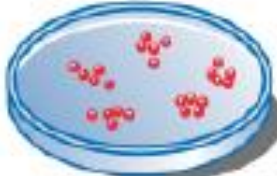
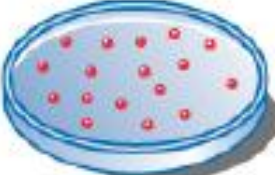
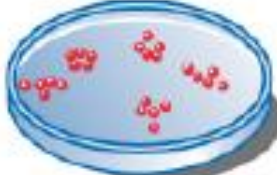
$T_1T_2 \times T_1T_2$

|       |          |          |
|-------|----------|----------|
|       | $T_1$    | $T_2$    |
| $T_1$ | $T_1T_1$ | $T_1T_2$ |
| $T_2$ | $T_1T_2$ | $T_2T_2$ |

$\frac{1}{4}$  Fioritura precoce (Giorno 0,0)  
 $\frac{1}{2}$  Fioritura intermedia (Giorno 3,7)  
 $\frac{1}{4}$  Fioritura tardiva (Giorno 5,2)

**Figura 4.2 Dominanza incompleta nel tempo di fioritura delle piante di pisello.** (a) L'allele  $T_2$  esercita dominanza incompleta sull'allele  $T_1$ , come indicato dal tardivo periodo di fioritura delle piante  $T_1T_2$ . (b) Segregazione degli alleli  $T_1$  e  $T_2$ .

# Variazioni della dominanza II codominanza

| <u>Genotype</u> | <u>Blood type<br/>(antigen present)</u> | <u>Reactions with anti-sera</u>                                                      |                                                                                       |
|-----------------|-----------------------------------------|--------------------------------------------------------------------------------------|---------------------------------------------------------------------------------------|
|                 |                                         | Anti-M serum                                                                         | Anti-N serum                                                                          |
| $L^M L^M$       | M                                       |    |    |
| $L^M L^N$       | M N                                     |   |   |
| $L^N L^N$       | N                                       |  |  |

Copyright 2000 John Wiley and Sons, Inc.

Gruppo  
sanguigno MN  
Su un locus  
autosomico chr 4

# Alleli letali

Topi gialli x topi selvatici

$Y^{+}$   $\downarrow$   $++$

topi gialli topi selvatici

$Y^{+}$   $++$

1 : 1

$Y > +$

topi gialli F1 x topi gialli F1

$Y^{+}$   $Y^{+}$

$\downarrow$

2/3 gialli

1/3 selvatici

Invece di 3/4 gialli

1/4 selvatici

# Alleli letali

$Y +$   
genitore 1

x

$Y +$   
genitore 2

Gameti genitore 1

$Y \frac{1}{2}$

$+ \frac{1}{2}$

Gameti genitore 2

$Y \frac{1}{2}$

$+ \frac{1}{2}$

|                                                                                    |                         |
|------------------------------------------------------------------------------------|-------------------------|
| <del> <math>Y Y</math><br/>                     Topo giallo                 </del> | $Y +$<br>Topo giallo    |
| $y +$<br>Topo giallo                                                               | $+ +$<br>Topo selvatico |

Rapporto  
alterato  
1:2:0

$\frac{2}{3}$  gialli

$\frac{1}{3}$  selvatici

# Alleli letali

$Y$  è un gene che per il fenotipo del colore del pelo si comporta da dominante, ma è recessivo per la **letalità**

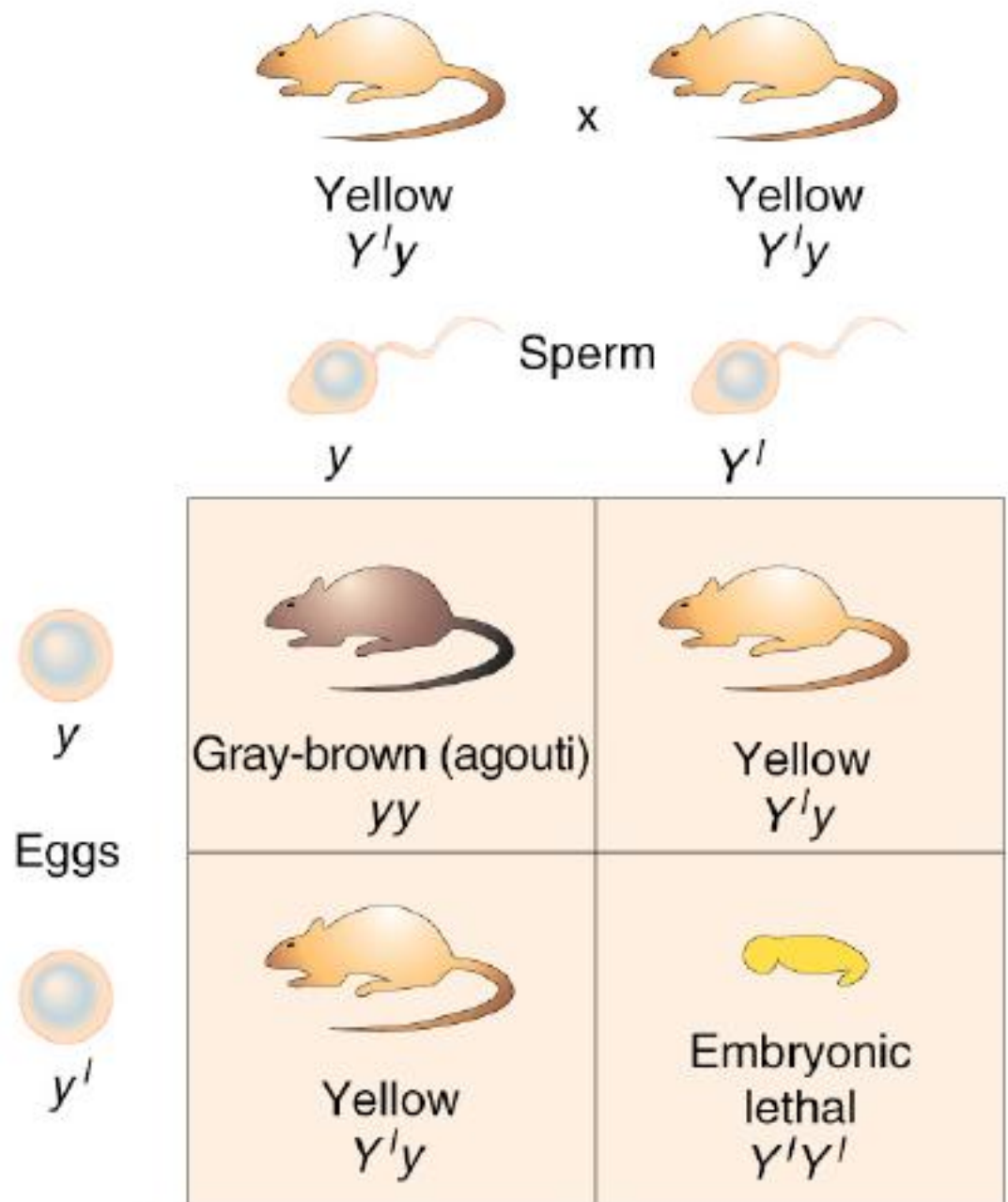
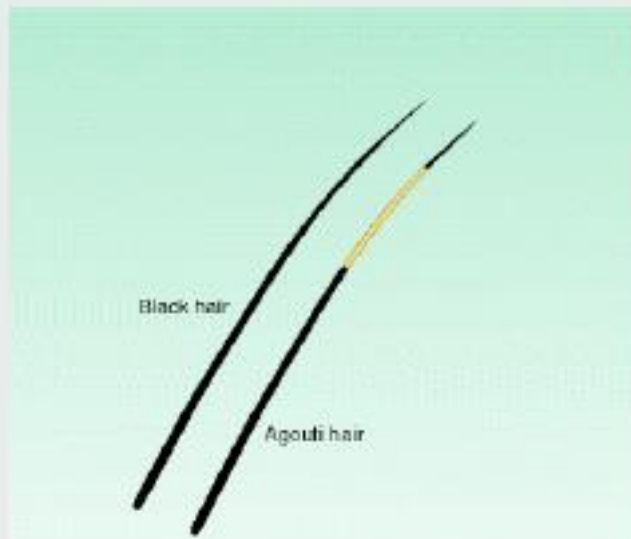


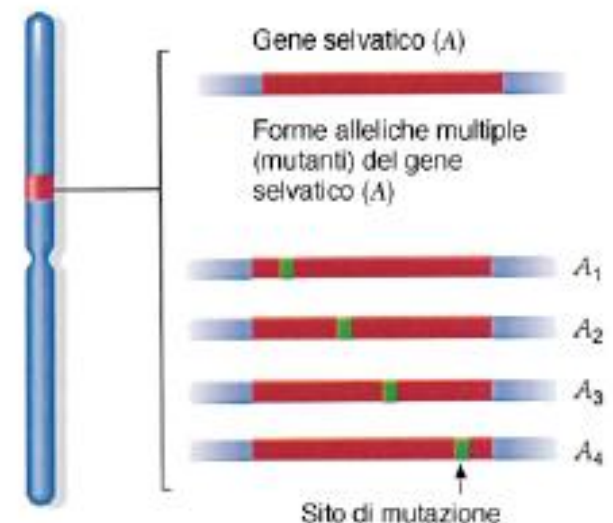
Figure 4.7  $Y^l$ , the yellow-lethal mutation in mice: a dominant visible that is also a recessive lethal. A cross between carriers of this mutation

# Allelia multipla

In ogni individuo diploide sono presenti solo due alleli ad un certo *locus* genetico. Tuttavia nella **popolazione** di individui è possibile che siano presenti **tanti** alleli diversi, che tra loro possono avere rapporti variabili di dominanza, espressività e penetranza.

**Figura 12.1**

Forme alleliche di un gene.



# I gruppi sanguigni ABO

➤ 3 alleli:  $I^A$ ,  $I^B$ ,  $i$  ( $I$  = isoagglutinogeno)

ABO Blood Groups in Humans, Determined by the Alleles  $I^A$ ,  $I^B$ , and  $i$

| PHENOTYPE<br>(BLOOD GROUP) | GENOTYPE             |
|----------------------------|----------------------|
| O                          | $i/i$                |
| A                          | $I^A/I^A$ or $I^A/i$ |
| B                          | $I^B/I^B$ or $I^B/i$ |
| AB                         | $I^A/I^B$            |

➤ Il sistema ABO è un esempio di **codominanza**

# I gruppi sanguigni AB0

**TABLE 4.1**

POTENTIAL PHENOTYPES IN THE OFFSPRING OF PARENTS WITH ALL POSSIBLE ABO BLOOD GROUP COMBINATIONS, ASSUMING HETEROZYGOSITY WHENEVER POSSIBLE

| Phenotypes | Parents                  |  | Potential Offspring |     |     |     |
|------------|--------------------------|--|---------------------|-----|-----|-----|
|            | Genotypes                |  | A                   | B   | AB  | O   |
| A × A      | $I^A I^O \times I^A I^O$ |  | 3/4                 | —   | —   | 1/4 |
| B × B      | $I^B I^O \times I^B I^O$ |  | —                   | 3/4 | —   | 1/4 |
| O × O      | $I^O I^O \times I^O I^O$ |  | —                   | —   | —   | all |
| A × B      | $I^A I^O \times I^B I^O$ |  | 1/4                 | 1/4 | 1/4 | 1/4 |
| A × AB     | $I^A I^O \times I^A I^B$ |  | 1/2                 | 1/4 | 1/4 | —   |
| A × O      | $I^A I^O \times I^O I^O$ |  | 1/2                 | —   | —   | 1/2 |
| B × AB     | $I^B I^O \times I^A I^B$ |  | 1/4                 | 1/2 | 1/4 | —   |
| B × O      | $I^B I^O \times I^O I^O$ |  | —                   | 1/2 | —   | 1/2 |
| AB × O     | $I^A I^B \times I^O I^O$ |  | 1/2                 | 1/2 | —   | —   |
| AB × AB    | $I^A I^B \times I^A I^B$ |  | 1/4                 | 1/2 | 1/2 | —   |

# Spiegazione molecolare gruppi AB0

| Fenotipo<br>(gruppo sanguigno) | Genotipo                       | Tipo di antigene | Anticorpi prodotti dall'organismo | Reazioni indotte nel sangue del ricevente contro i globuli rossi contenuti in quello del donatore |                                                                                       |                                                                                       |                                                                                       |
|--------------------------------|--------------------------------|------------------|-----------------------------------|---------------------------------------------------------------------------------------------------|---------------------------------------------------------------------------------------|---------------------------------------------------------------------------------------|---------------------------------------------------------------------------------------|
|                                |                                |                  |                                   | (anticorpi B)                                                                                     | (anticorpi A)                                                                         | AB<br>(assenza di anticorpi)                                                          | O<br>(anticorpi A e B)                                                                |
| A                              | $I^A I^A$<br>oppure<br>$I^A i$ | A                | anti B                            |                 |    |    |    |
| B                              | $I^B I^B$<br>oppure<br>$I^B i$ | B                | anti A                            |                 |    |    |    |
| AB                             | $I^A I^B$                      | A e B            | Nessuno                           |                |   |   |   |
| O                              | $ii$                           | Nessuno          | anti A e B                        |               |  |  |  |

I globuli rossi che non reagiscono con gli anticorpi del ricevente rimangono disseminati in modo uniforme. Il sangue del donatore e quello del ricevente sono compatibili.

I globuli rossi che reagiscono con gli anticorpi del ricevente si aggregano tra loro. Il sangue del donatore e quello del ricevente non sono compatibili.

**O: donatore universale**

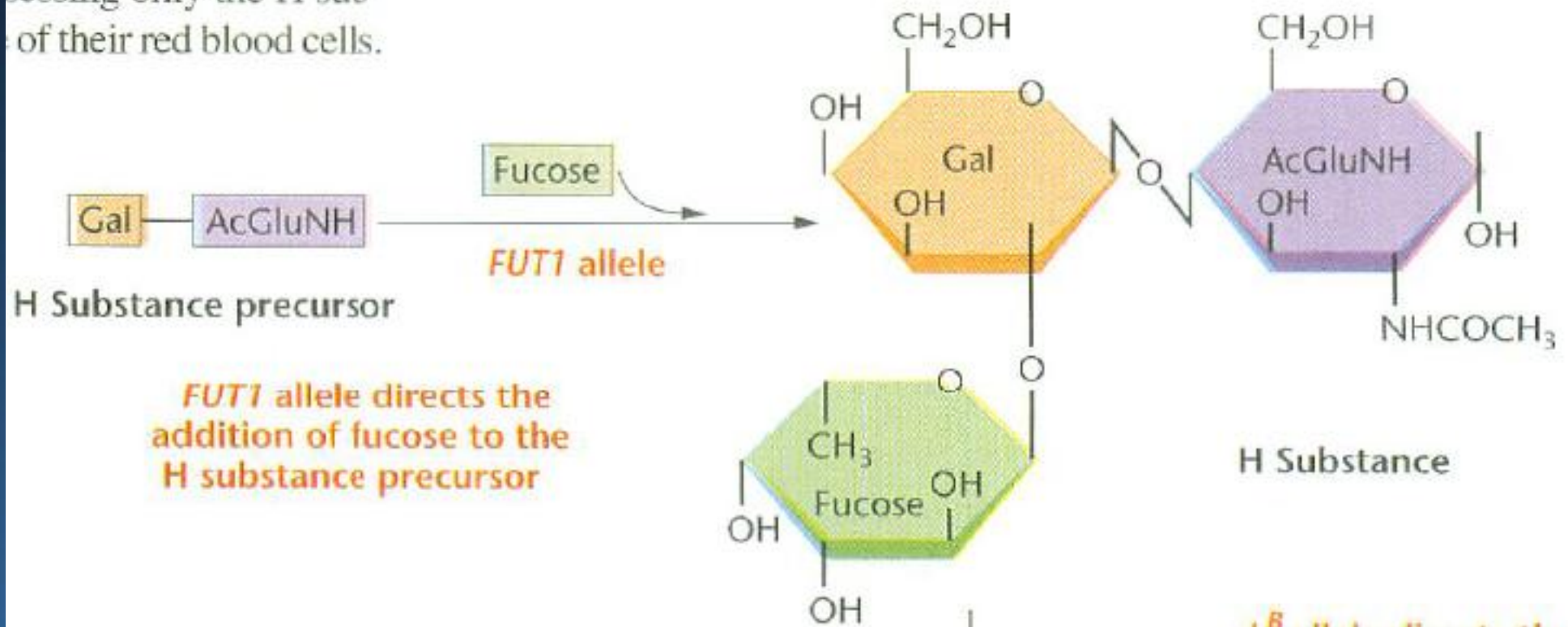
I donatori del gruppo O vanno bene per qualsiasi tipo di ricevente: sono i *donatori universali*.

I riceventi di gruppo AB possono accettare sangue da qualsiasi donatore: sono definiti *riceventi universali*.

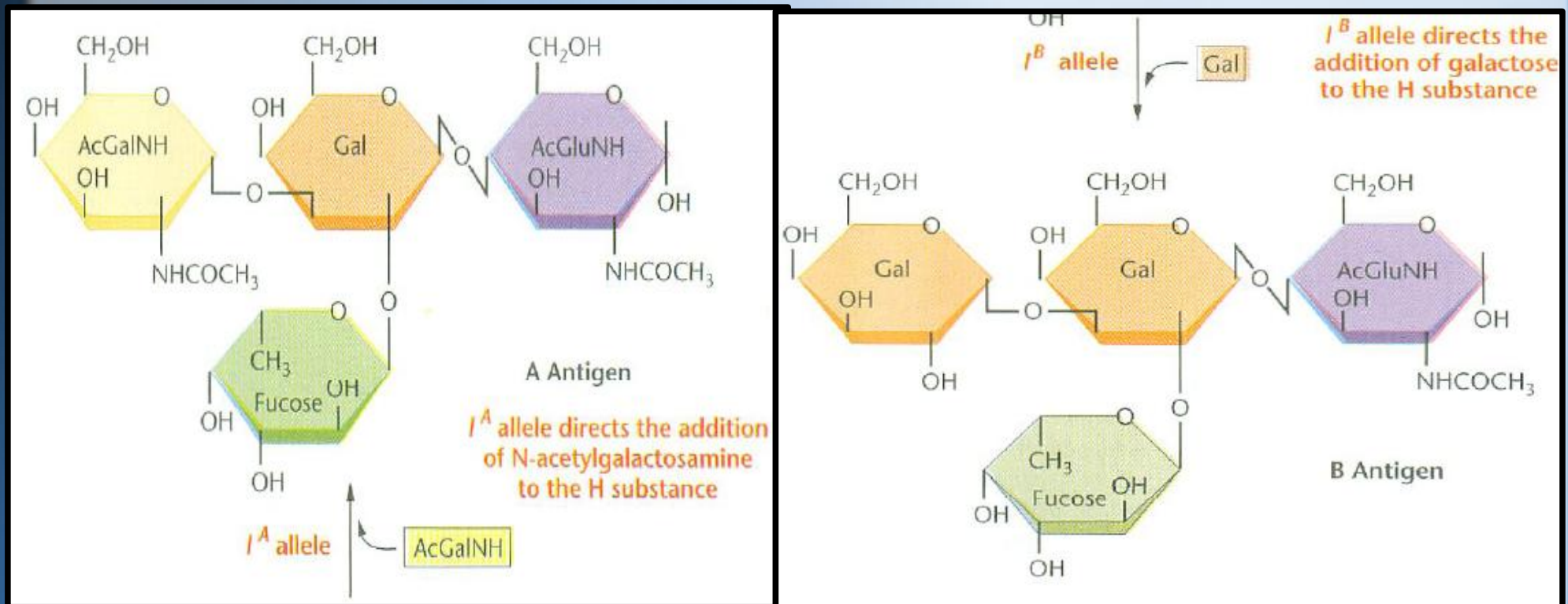
**AB: ricevitore universale**

# Spiegazione molecolare gruppi AB0

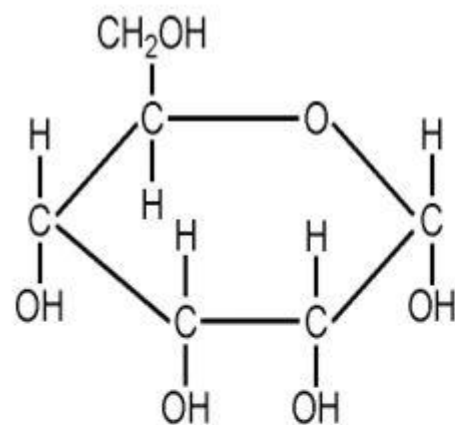
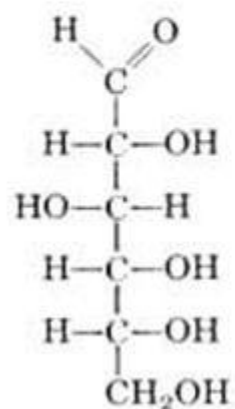
possessing only the H sub-  
stance of their red blood cells.



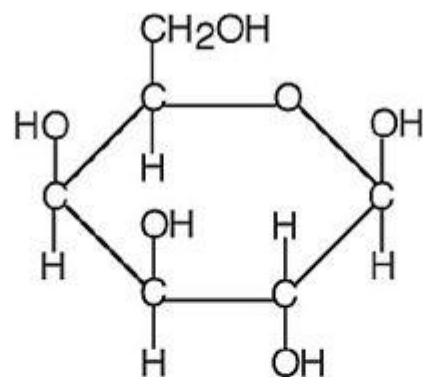
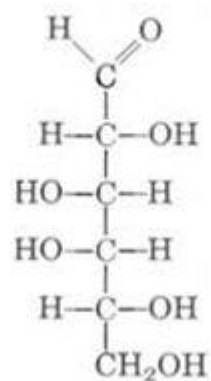
# Spiegazione molecolare gruppi AB0



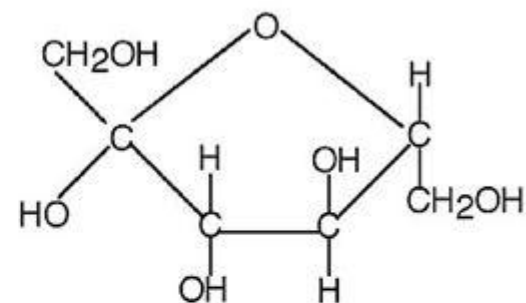
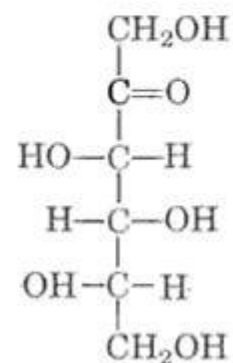
**Glucosio**  
 $C_6H_{12}O_6$



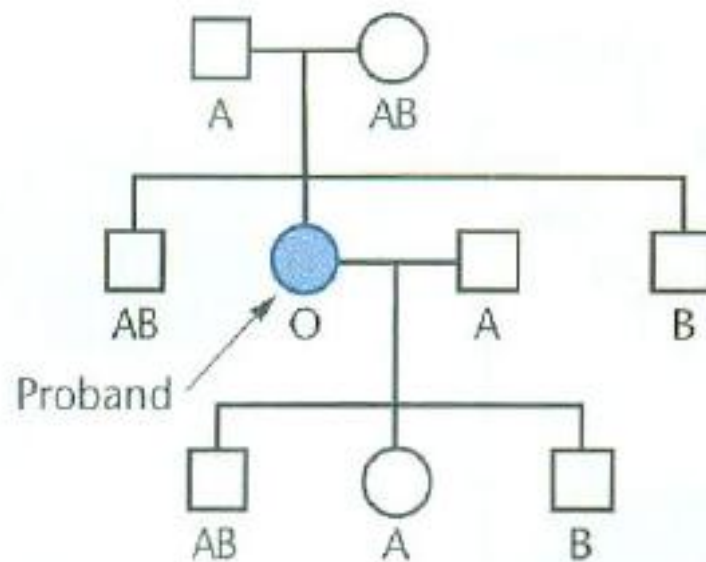
**Galattosio**  
 $C_6H_{12}O_6$



**Fruttosio**  
 $C_6H_{12}O_6$



# Il fenotipo Bombay



**FIGURE 4-3** A partial pedigree of a woman displaying the Bombay phenotype. Functionally, her ABO blood group behaves as type O. Genetically, she is type B.

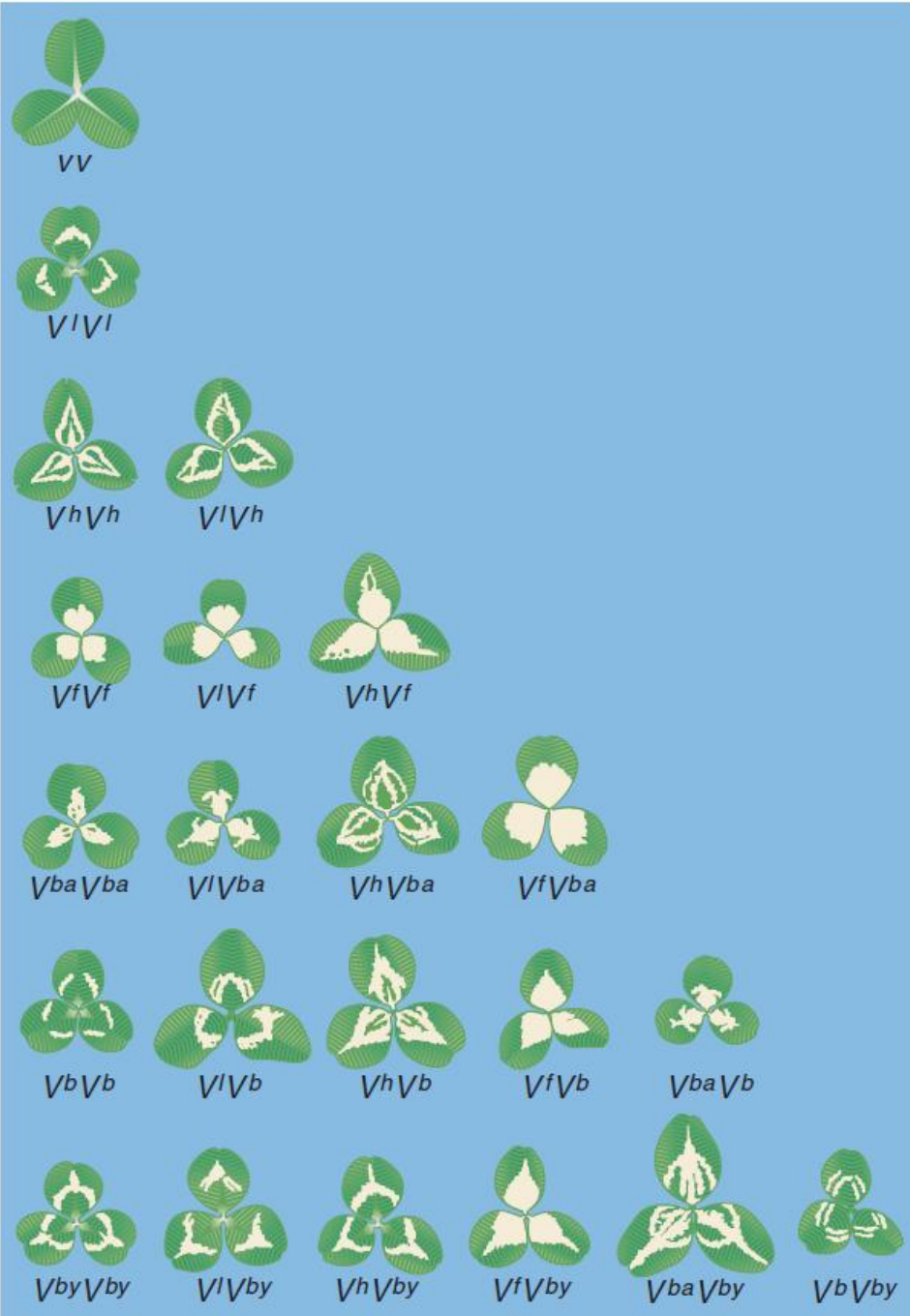
# Il locus *white* in *Drosophila*

Le mutazione dell'occhio sono oltre 100, la prima scoperta da Morgan e Bridges nel 1912 (*white*)

TABLE 4.2

SOME OF THE ALLELES PRESENT AT THE *WHITE* LOCUS OF *DROSOPHILA*

| Allele                 | Name                        | Eye Color                   |
|------------------------|-----------------------------|-----------------------------|
| <i>w</i>               | <i>white</i>                | pure white                  |
| <i>w<sup>a</sup></i>   | <i>white-apricot</i>        | yellowish orange            |
| <i>w<sup>bf</sup></i>  | <i>white-buff</i>           | light buff                  |
| <i>w<sup>bl</sup></i>  | <i>white-blood</i>          | yellowish ruby              |
| <i>w<sup>cf</sup></i>  | <i>white-coffee</i>         | deep ruby                   |
| <i>w<sup>e</sup></i>   | <i>white-eosin</i>          | yellowish pink              |
| <i>w<sup>mo</sup></i>  | <i>white-mottled orange</i> | light mottled orange        |
| <i>w<sup>sat</sup></i> | <i>white-satsuma</i>        | deep ruby                   |
| <i>w<sup>sp</sup></i>  | <i>white spotted</i>        | fine grain, yellow mottling |
| <i>w<sup>t</sup></i>   | <i>white-tinged</i>         | light pink                  |



**Figure 6-12 Multiple alleles determine the chevron pattern on the leaves of white clover.** The genotype of each plant is shown below it. [After photograph by W. Ellis Davies.]

# Serie alleliche

Gli alleli di una serie allelica possono essere completamente dominanti/recessivi oppure possono mostrare varie forme di codominanza o dominanza incompleta



Colore pieno  
 $CC, C-$



Cincillà  
 $c^{ch}c^{ch}, c^{ch}c$



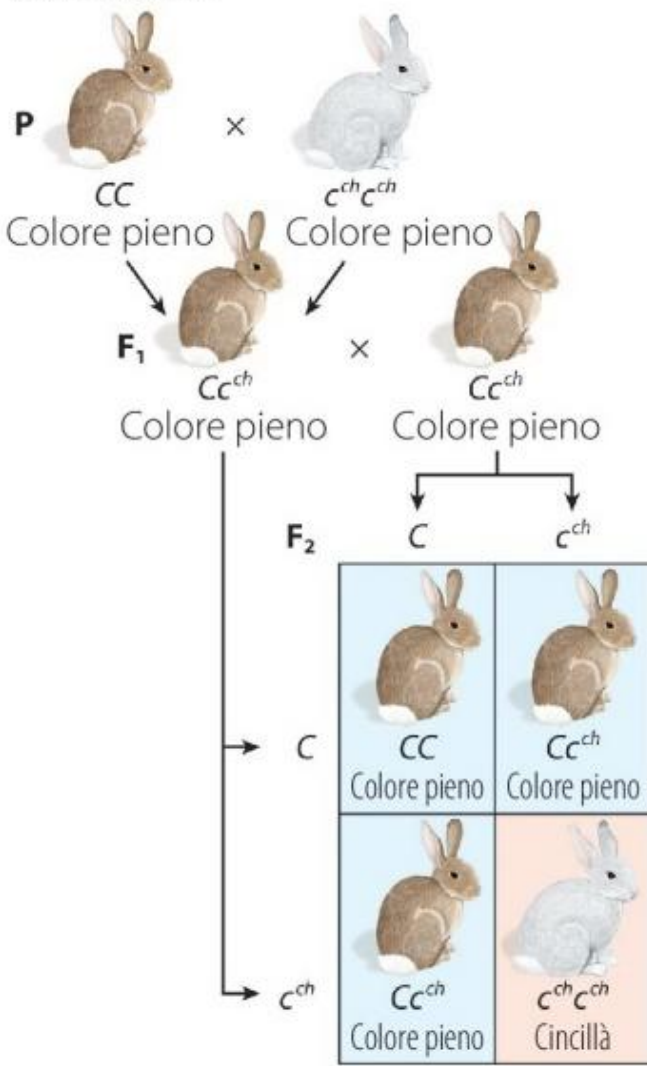
Himalayano  
 $c^Hc^H, c^Hc$



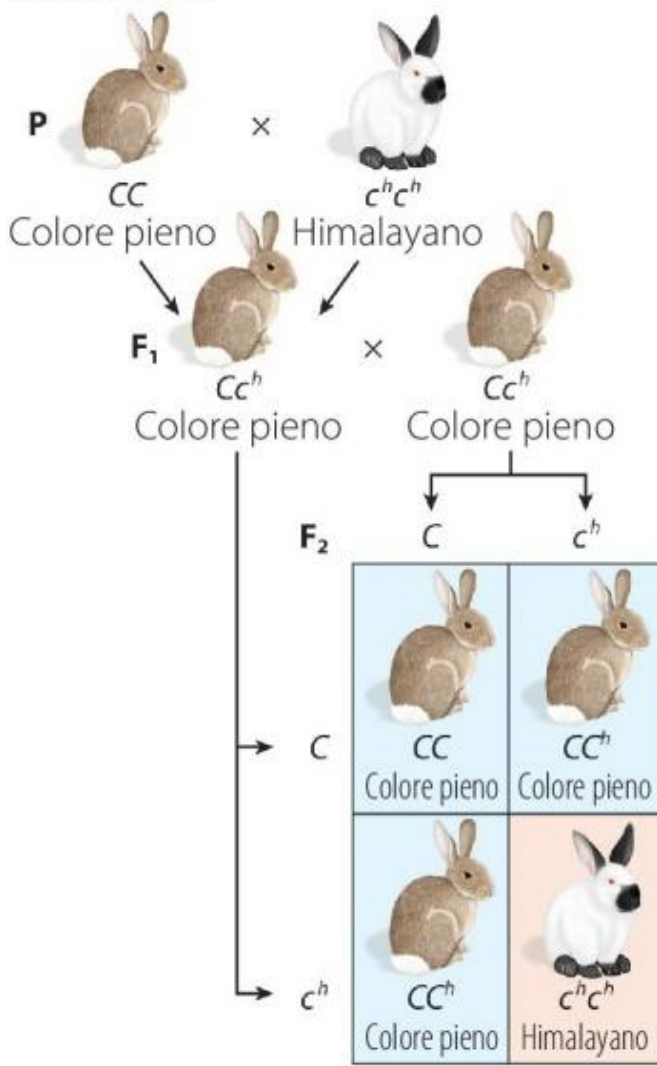
Albino  
 $cc$

**Figura 4.5** Serie allelica per la determinazione del colore del mantello nei mammiferi.

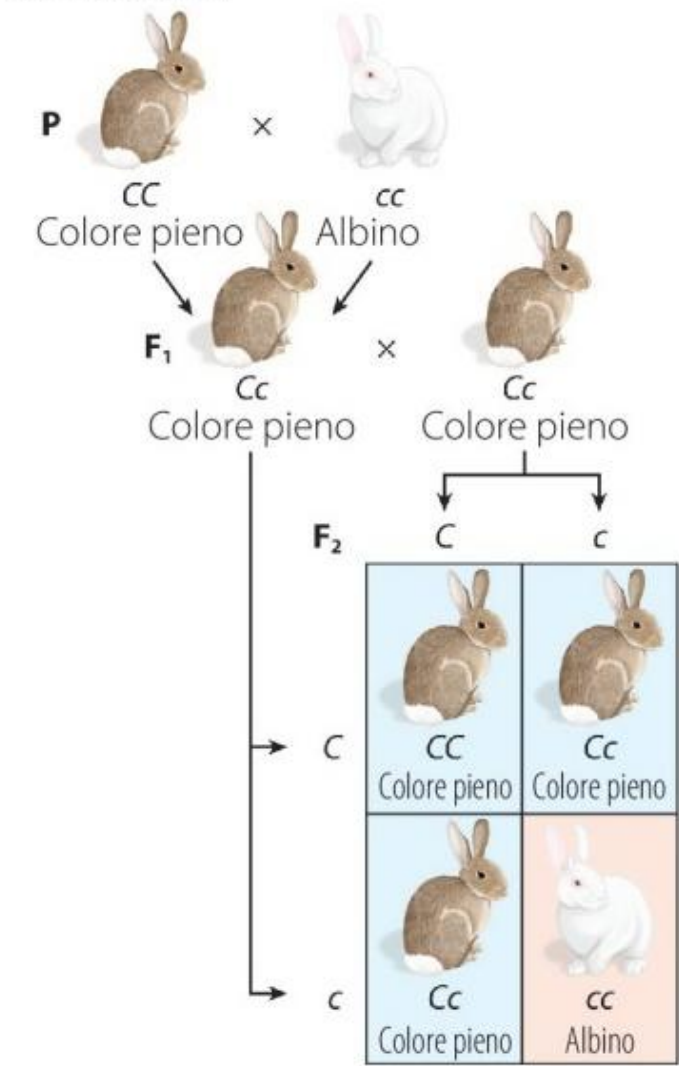
(a) Incrocio A



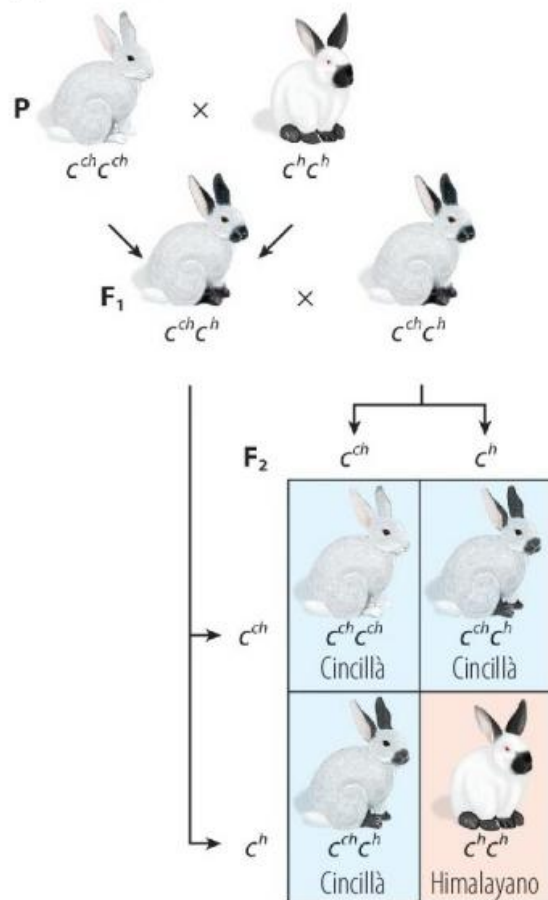
(b) Incrocio B



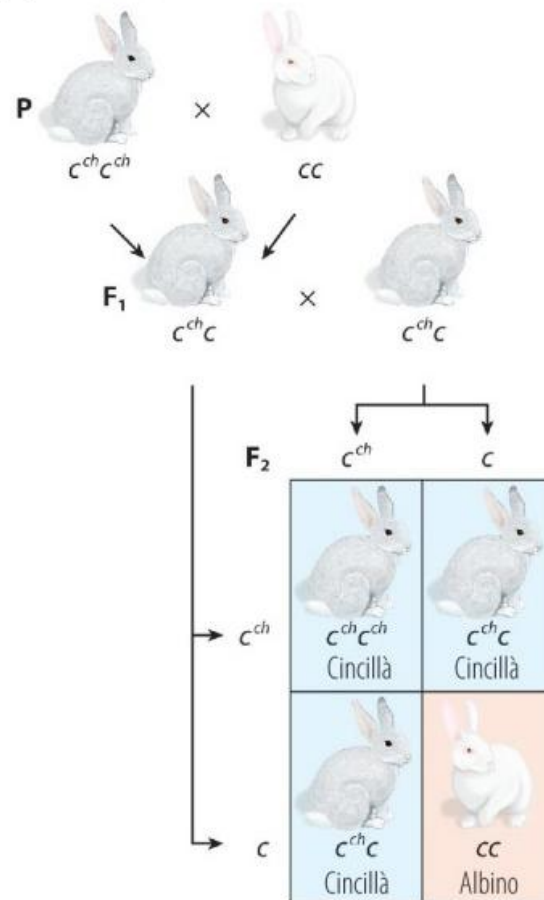
(c) Incrocio C



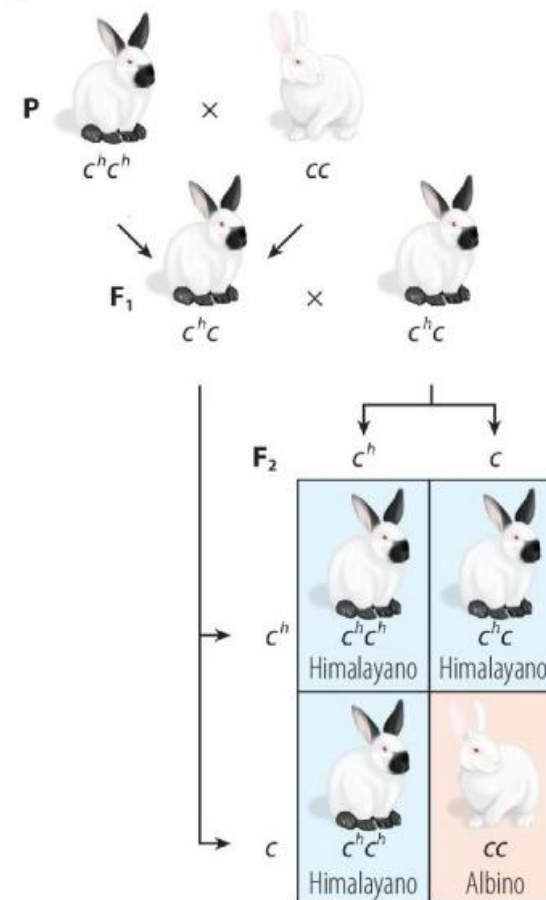
(d) Incrocio D



(e) Incrocio E



(f) Incrocio F

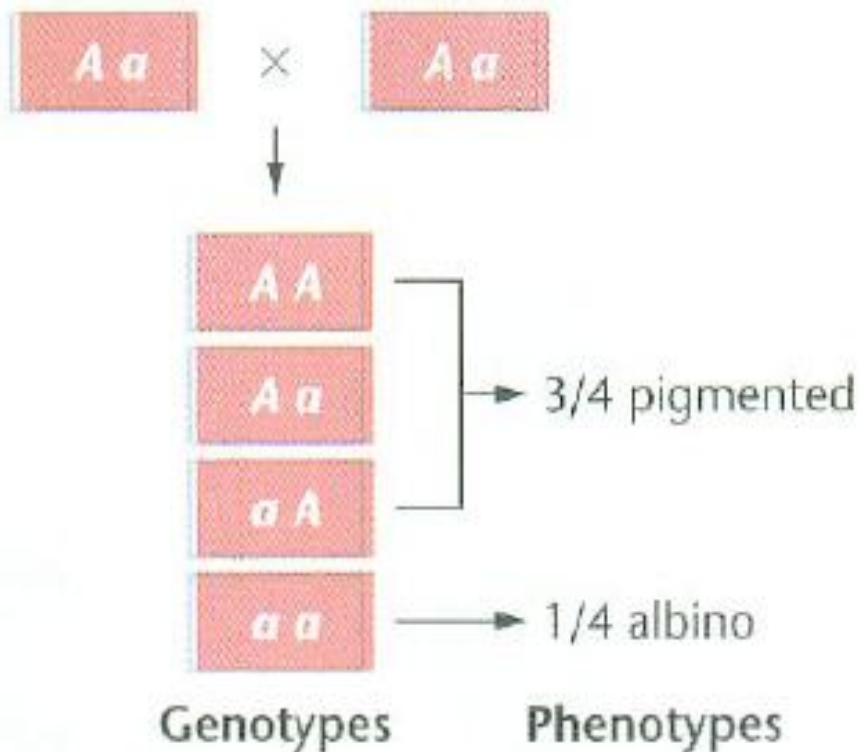


**Figura 4.6 Genetica della dominanza del gene C.** (a)–(f). Gli incroci da A a F illustrano la dominanza completa di C e la recessività completa di c e stabiliscono la serie allelica  $C > c^{ch} > c^h > c$ .

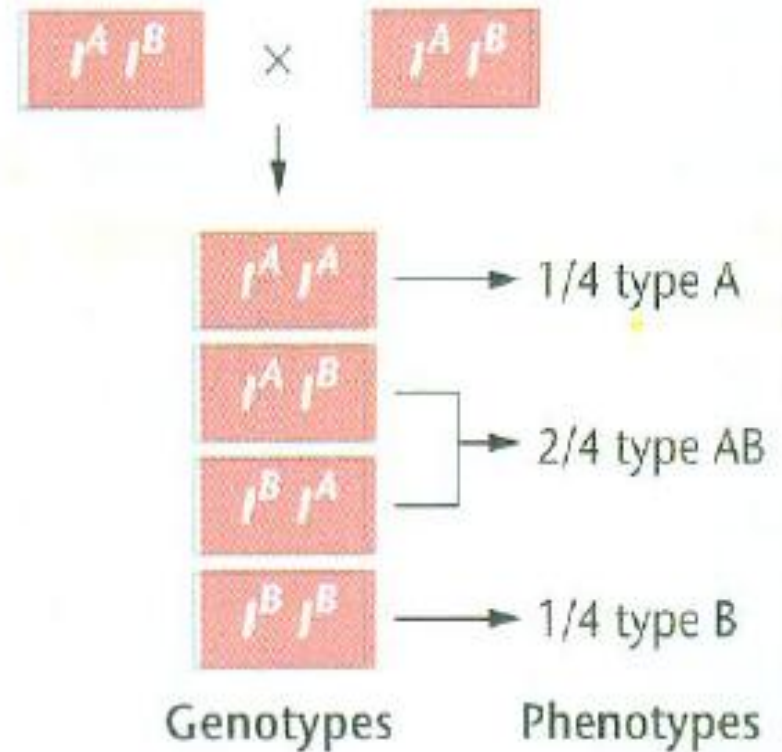
# Calcolo dell'ereditarietà con 2 geni con modelli diversi

$$AaI^A I^B \times AaI^A I^B$$

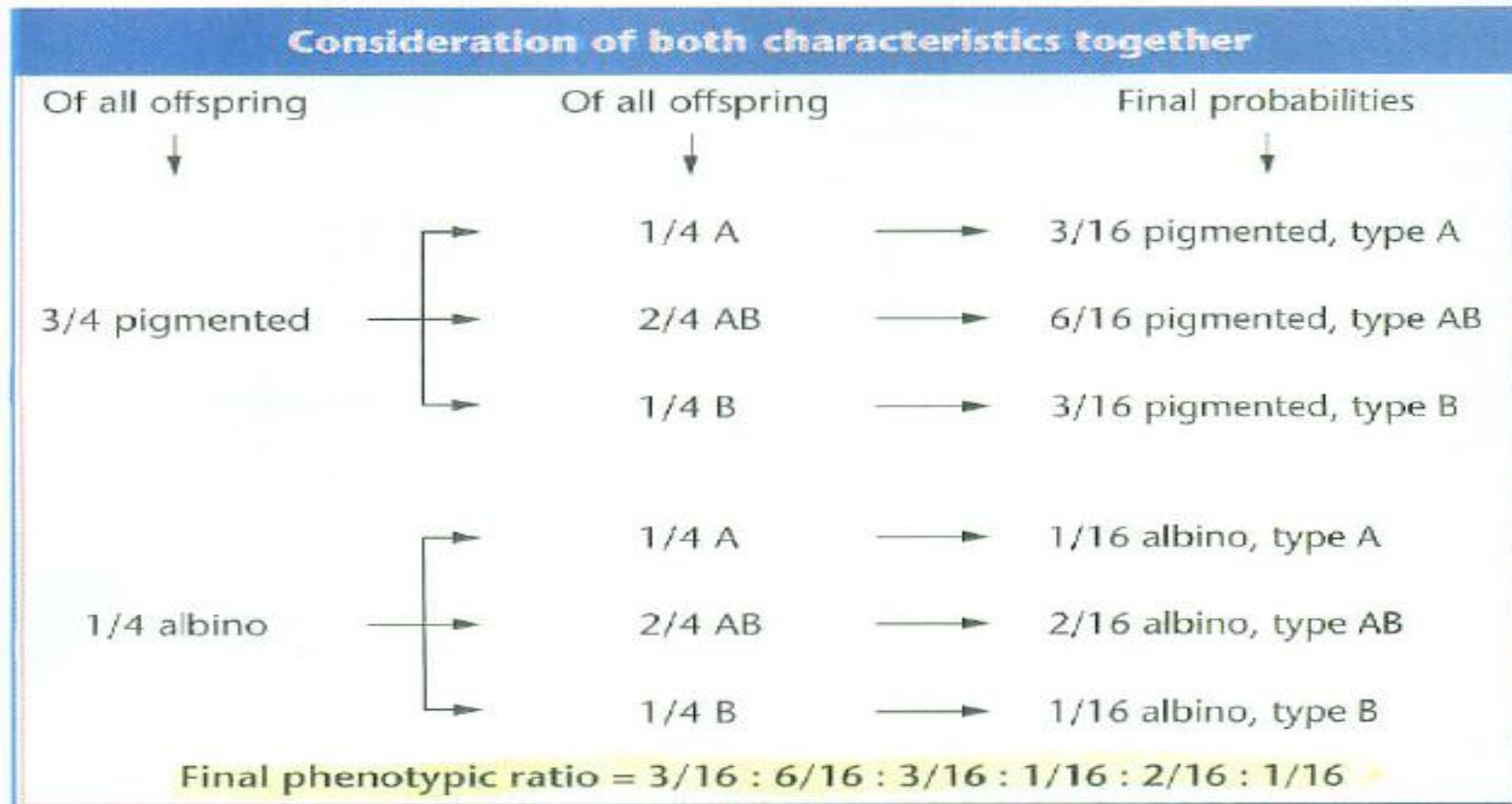
Consideration of pigmentation alone



Consideration of blood types alone



# Calcolo dell'ereditarietà con 2 geni con modelli diversi



**FIGURE 4-5** Calculation of the probabilities in a mating involving the ABO blood type and albinism in humans. The calculation is carried out by using the forked-line method.

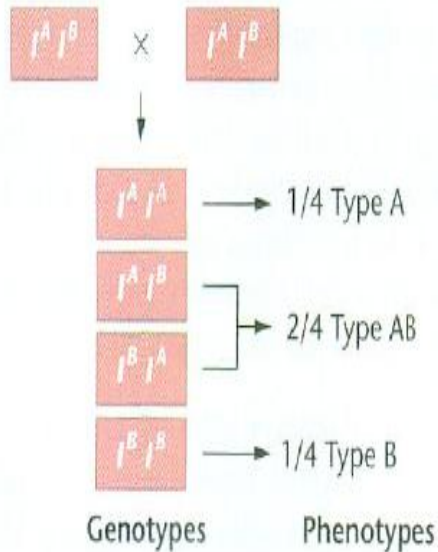
# Interazione genica (epistasi)

- interazione di due o più geni: cooperazione, antagonismo, inibizione, modificazione
- geni diversi influenzano lo stesso fenotipo
- gene epistatico: un gene agisce "su" altri geni (in senso stretto reprime l'azione di altri geni)
- il fenotipo può essere più o meno complesso (colore della pelliccia, sviluppo di un organo)

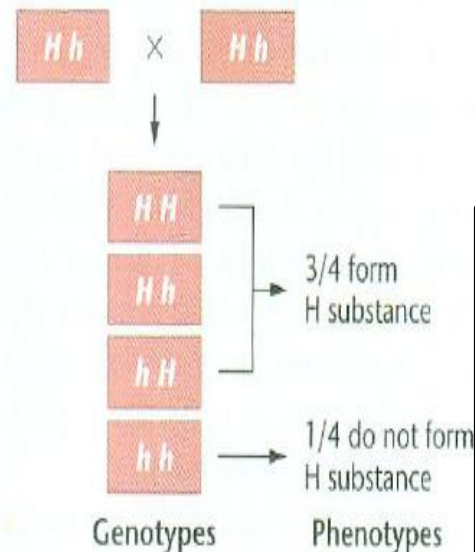
# Il fenotipo Bombay

$I^A I^B \ H h \times I^A I^B \ H h$

Consideration of blood types



Consideration of H substance



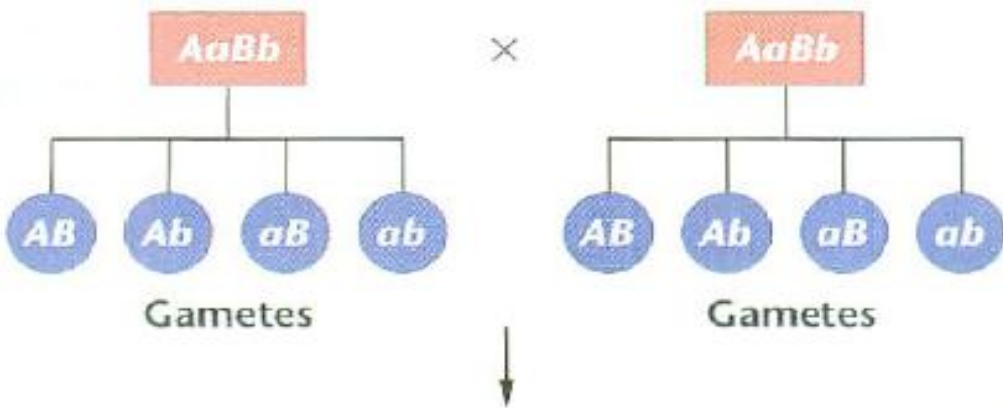
Il gene FUT1 è epistatico sul sistema AB0

Consideration of both gene pairs together

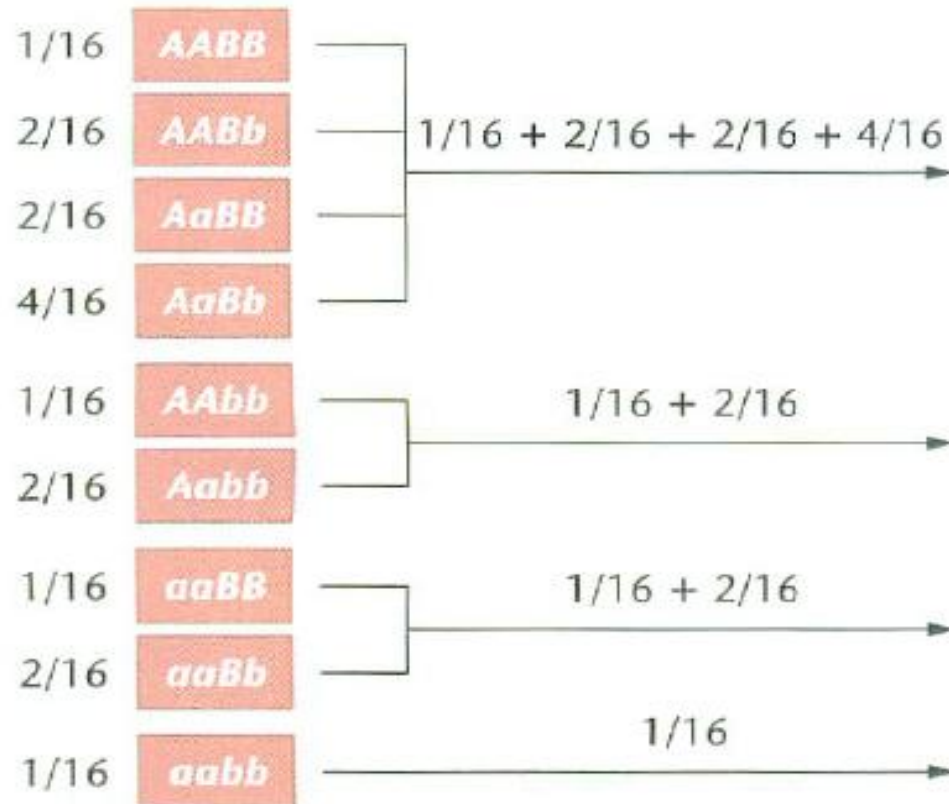
| Of all offspring | Of all offspring            | Final probabilities |
|------------------|-----------------------------|---------------------|
| ↓                | ↓                           | ↓                   |
| 1/4 Type A       | 3/4 form H substance        | 3/16 Type A         |
|                  | 1/4 do not form H substance | 1/16 Type O         |
| 2/4 Type AB      | 3/4 form H substance        | 6/16 Type AB        |
|                  | 1/4 do not form H substance | 2/16 Type O         |
| 1/4 Type B       | 3/4 form H substance        | 3/16 Type B         |
|                  | 1/4 do not form H substance | 1/16 Type O         |

Final phenotypic ratio = 3/16 A: 6/16 AB: 3/16 B: 4/16 O

# Altri modelli di ereditarietà



**FIGURE 4-7** Generation of the various modified dihybrid ratios from the nine unique genotypes produced in a cross between individuals heterozygous at two genes.



| Dihybrid ratio | Modified ratios |         |        |        |         |
|----------------|-----------------|---------|--------|--------|---------|
| $9/16 A - B -$ | $9/16$          |         | $9/16$ | $9/16$ |         |
| $3/16 A - bb$  | $3/16$          | $12/16$ |        |        | $15/16$ |
| $3/16 aaB -$   |                 | $3/16$  | $7/16$ | $6/16$ |         |
| $1/16 aabb$    | $4/16$          | $1/16$  |        | $1/16$ | $1/16$  |

# Epistasi resessiva

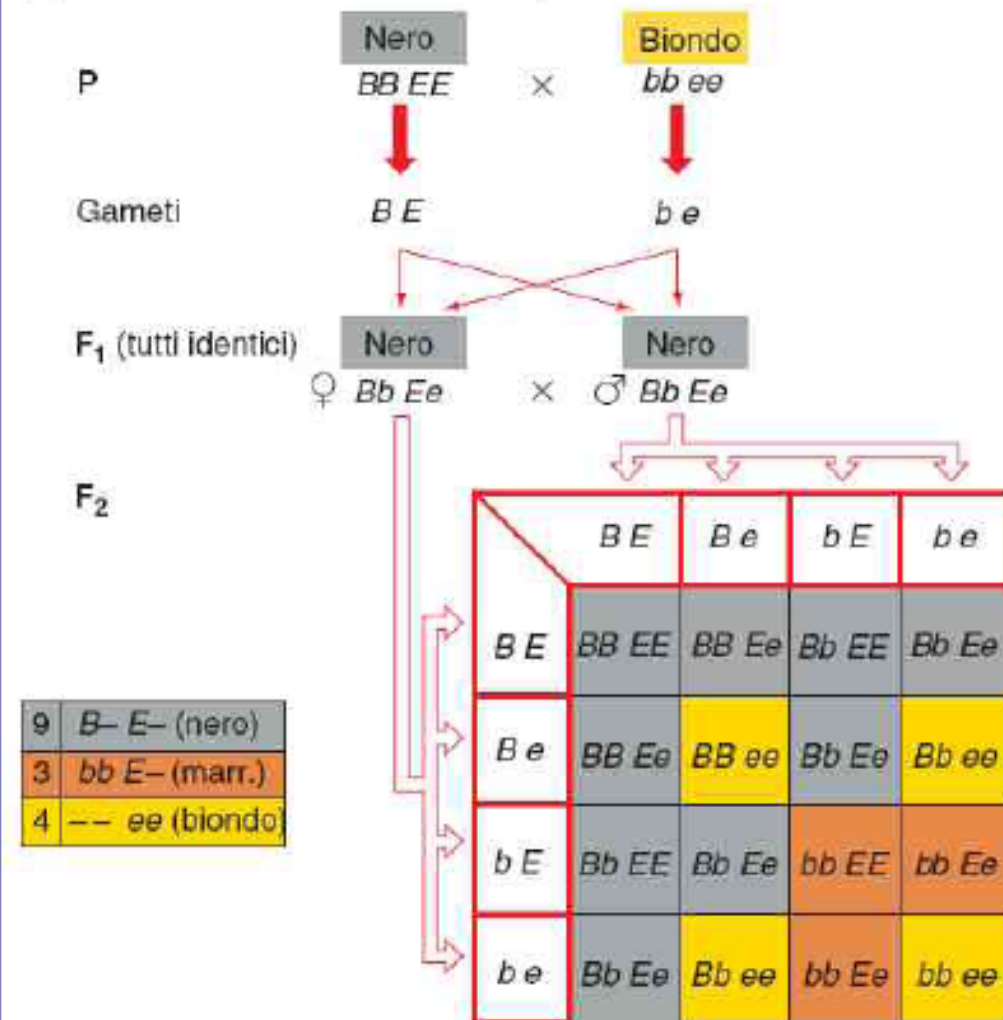
| Case | Organism         | Character    | F <sub>2</sub> Phenotypes |        |         |               | Modified ratio |
|------|------------------|--------------|---------------------------|--------|---------|---------------|----------------|
|      |                  |              | 9/16                      | 3/16   | 3/16    | 1/16          |                |
| 1    | Mouse            | Coat color   | agouti                    | albino | black   | albino        | 9:3:4          |
| 2    | Squash           | Color        | white                     |        | yellow  | green         | 12:3:1         |
| 3    | Pea              | Flower color | purple                    | white  |         |               | 9:7            |
| 4    | Squash           | Fruit shape  | disc                      | sphere |         | long          | 9:6:1          |
| 5    | Chicken          | Color        | white                     |        | colored | white         | 13:3           |
| 6    | Mouse            | Color        | white-spotted             | white  | colored | white-spotted | 10:3:3         |
| 7    | Shepherd's purse | Seed capsule | triangular                |        |         | ovoid         | 15:1           |
| 8    | Flour beetle     | Color        | 6/16 sooty : 3/16 red     | black  | jet     | black         | 6:3:3:4        |

# Epistasi recessiva

➤ l'allele recessivo di un locus prevale (come genotipo omozigote recessivo) sul fenotipo determinato dall'altro locus

Es. colore del pelo dei Labrador  
 B: produzione di melanina  
 B- nero  
 bb: marrone  
 E: deposito del pigmento  
 E- normale  
 ee impedita

(a) Incrocio diibrido che mostra epistasi recessiva



# Epistasi dominante

| Case | Organism         | Character    | F <sub>2</sub> Phenotypes |        |         |               | Modified ratio |
|------|------------------|--------------|---------------------------|--------|---------|---------------|----------------|
|      |                  |              | 9/16                      | 3/16   | 3/16    | 1/16          |                |
| 1    | Mouse            | Coat color   | agouti                    | albino | black   | albino        | 9:3:4          |
| 2    | Squash           | Color        | white                     |        | yellow  | green         | 12:3:1         |
| 3    | Pea              | Flower color | purple                    | white  |         |               | 9:7            |
| 4    | Squash           | Fruit shape  | disc                      | sphere |         | long          | 9:6:1          |
| 5    | Chicken          | Color        | white                     |        | colored | white         | 13:3           |
| 6    | Mouse            | Color        | white-spotted             | white  | colored | white-spotted | 10:3:3         |
| 7    | Shepherd's purse | Seed capsule | triangular                |        |         | ovoid         | 15:1           |
| 8    | Flour beetle     | Color        | 6/16 sooty : 3/16 red     | black  | jet     | black         | 6:3:3:4        |

# Epistasi dominante



$F_1: AaBb \times AaBb$   
↓

| $F_2$ Ratio | Genotype | Phenotype | Final Phenotypic Ratio                   |
|-------------|----------|-----------|------------------------------------------|
| 9/16        | $A- B-$  | white     | 12/16 white<br>3/16 yellow<br>1/16 green |
| 3/16        | $A- bb$  | white     |                                          |
| 3/16        | $aa B-$  | yellow    |                                          |
| 1/16        | $aa bb$  | green     |                                          |

# Epsitasi duplicata recessiva

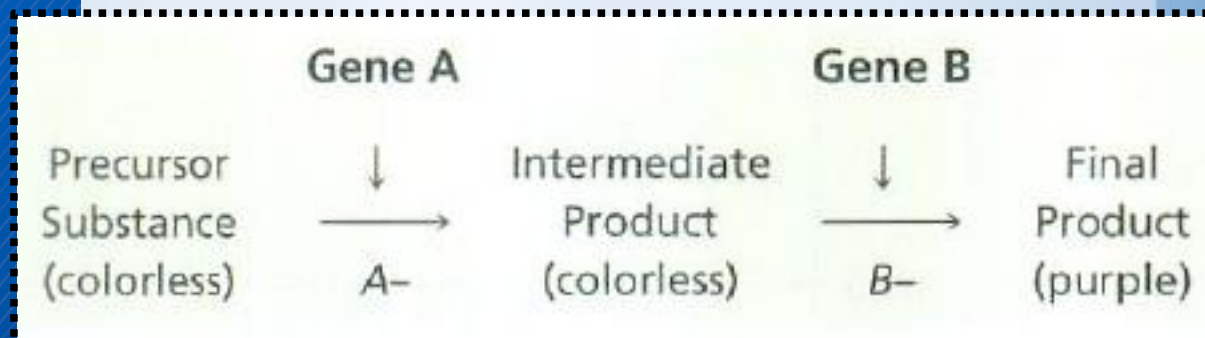
| Case | Organism         | Character    | F <sub>2</sub> Phenotypes |        |         |               | Modified ratio |
|------|------------------|--------------|---------------------------|--------|---------|---------------|----------------|
|      |                  |              | 9/16                      | 3/16   | 3/16    | 1/16          |                |
| 1    | Mouse            | Coat color   | agouti                    | albino | black   | albino        | 9:3:4          |
| 2    | Squash           | Color        | white                     |        | yellow  | green         | 12:3:1         |
| 3    | Pea              | Flower color | purple                    | white  |         |               | 9:7            |
| 4    | Squash           | Fruit shape  | disc                      | sphere |         | long          | 9:6:1          |
| 5    | Chicken          | Color        | white                     |        | colored | white         | 13:3           |
| 6    | Mouse            | Color        | white-spotted             | white  | colored | white-spotted | 10:3:3         |
| 7    | Shepherd's purse | Seed capsule | triangular                |        |         | ovoid         | 15:1           |
| 8    | Flour beetle     | Color        | 6/16 sooty : 3/16 red     | black  | jet     | black         | 6:3:3:4        |

# Epistasi duplicata recessiva

- il fenotipo dominante è determinato dalla presenza di almeno 1 allele dominante in ciascuno di 2 loci genici
- *il fenotipo recessivo è identico*
- *epistasi recessiva doppia*

Es. colore dei fiori della pianta *Lathyrus odoratus*  
 C, P: porpora  
 c, p: bianchi

| $P_1: AAbb \times aaBB$<br>white                      white |          |           |                           |
|-------------------------------------------------------------|----------|-----------|---------------------------|
| ↓                                                           |          |           |                           |
| $F_1: \text{All } AaBb \text{ (purple)}$                    |          |           |                           |
| ↓                                                           |          |           |                           |
| F <sub>2</sub> Ratio                                        | Genotype | Phenotype | Final Phenotypic Ratio    |
| 9/16                                                        | $A- B-$  | purple    | 9/16 purple<br>7/16 white |
| 3/16                                                        | $A- bb$  | white     |                           |
| 3/16                                                        | $aa B-$  | white     |                           |
| 1/16                                                        | $aabb$   | white     |                           |



# Interazione duplicata dominante

| Case | Organism         | Character    | F <sub>2</sub> Phenotypes |        |         |               | Modified ratio |
|------|------------------|--------------|---------------------------|--------|---------|---------------|----------------|
|      |                  |              | 9/16                      | 3/16   | 3/16    | 1/16          |                |
| 1    | Mouse            | Coat color   | agouti                    | albino | black   | albino        | 9:3:4          |
| 2    | Squash           | Color        | white                     |        | yellow  | green         | 12:3:1         |
| 3    | Pea              | Flower color | purple                    | white  |         |               | 9:7            |
| 4    | Squash           | Fruit shape  | disc                      | sphere |         | long          | 9:6:1          |
| 5    | Chicken          | Color        | white                     |        | colored | white         | 13:3           |
| 6    | Mouse            | Color        | white-spotted             | white  | colored | white-spotted | 10:3:3         |
| 7    | Shepherd's purse | Seed capsule | triangular                |        |         | ovoid         | 15:1           |
| 8    | Flour beetle     | Color        | 6/16 sooty : 3/16 red     | black  | jet     | black         | 6:3:3:4        |

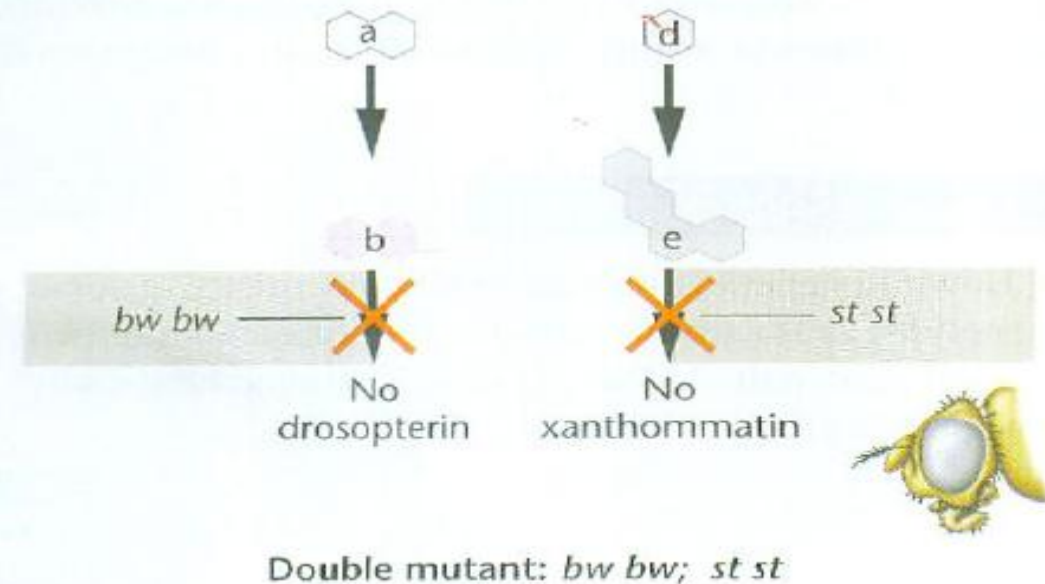
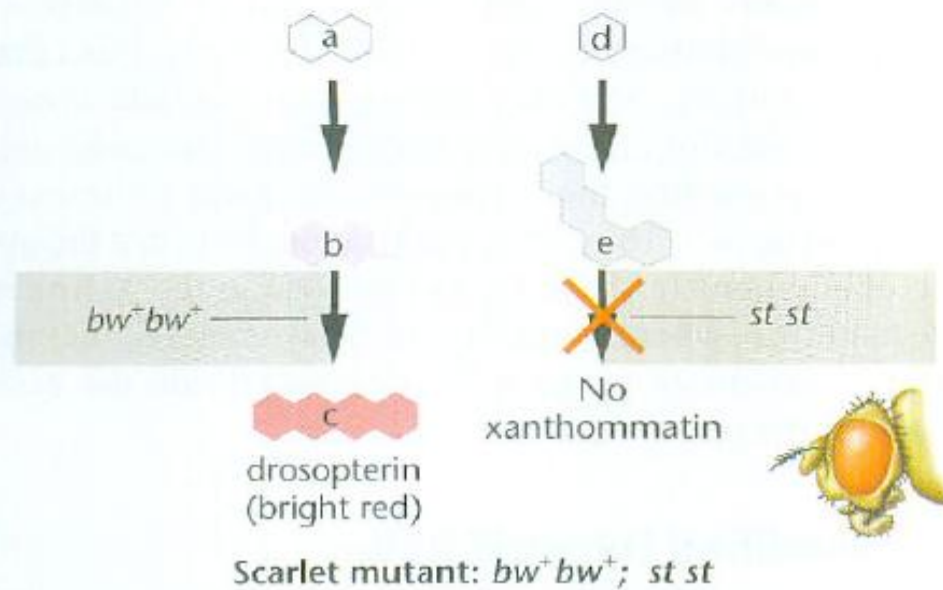
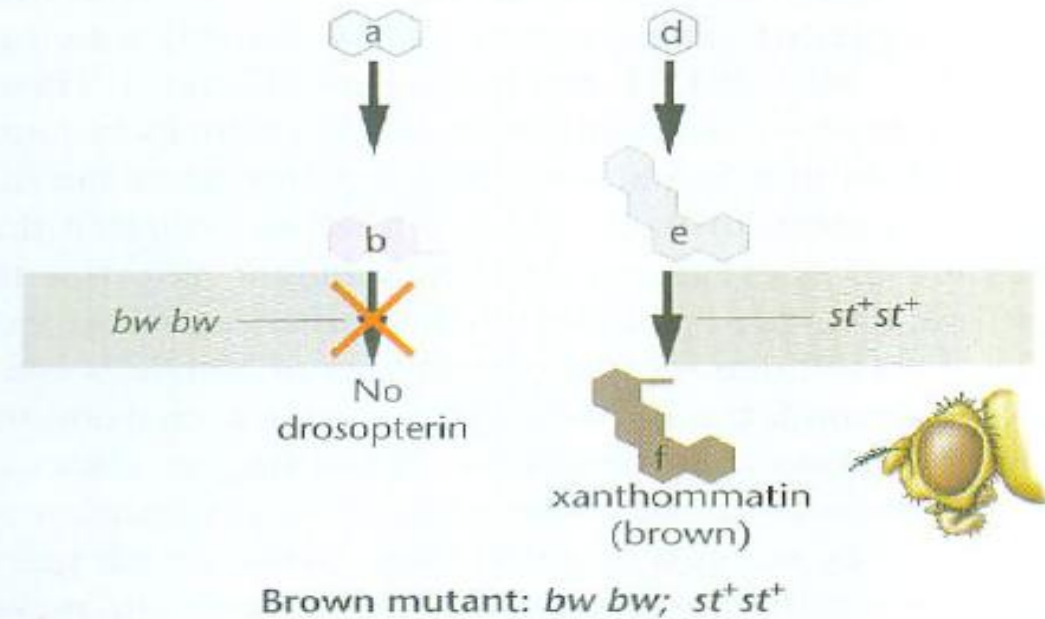
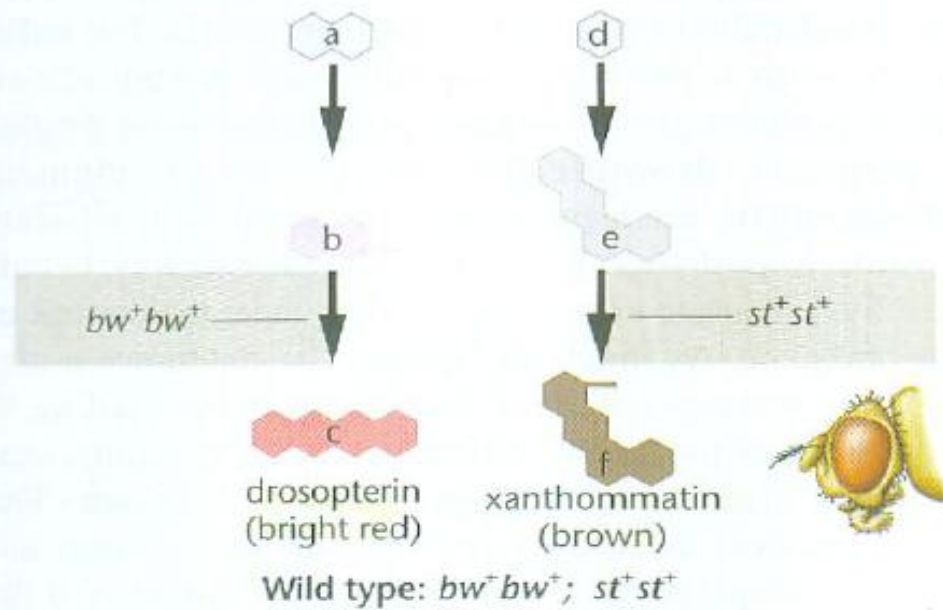


$F_1: AaBb \times AaBb$

disc  $\downarrow$  disc

| $F_2$ Ratio | Genotype | Phenotype | Final Phenotypic Ratio |
|-------------|----------|-----------|------------------------|
| 9/16        | $A- B-$  | disc      | 9/16 disc              |
| 3/16        | $A- bb$  | sphere    | 6/16 sphere            |
| 3/16        | $aa B-$  | sphere    |                        |
| 1/16        | $aa bb$  | long      | 1/16 long              |

# Interazione genica

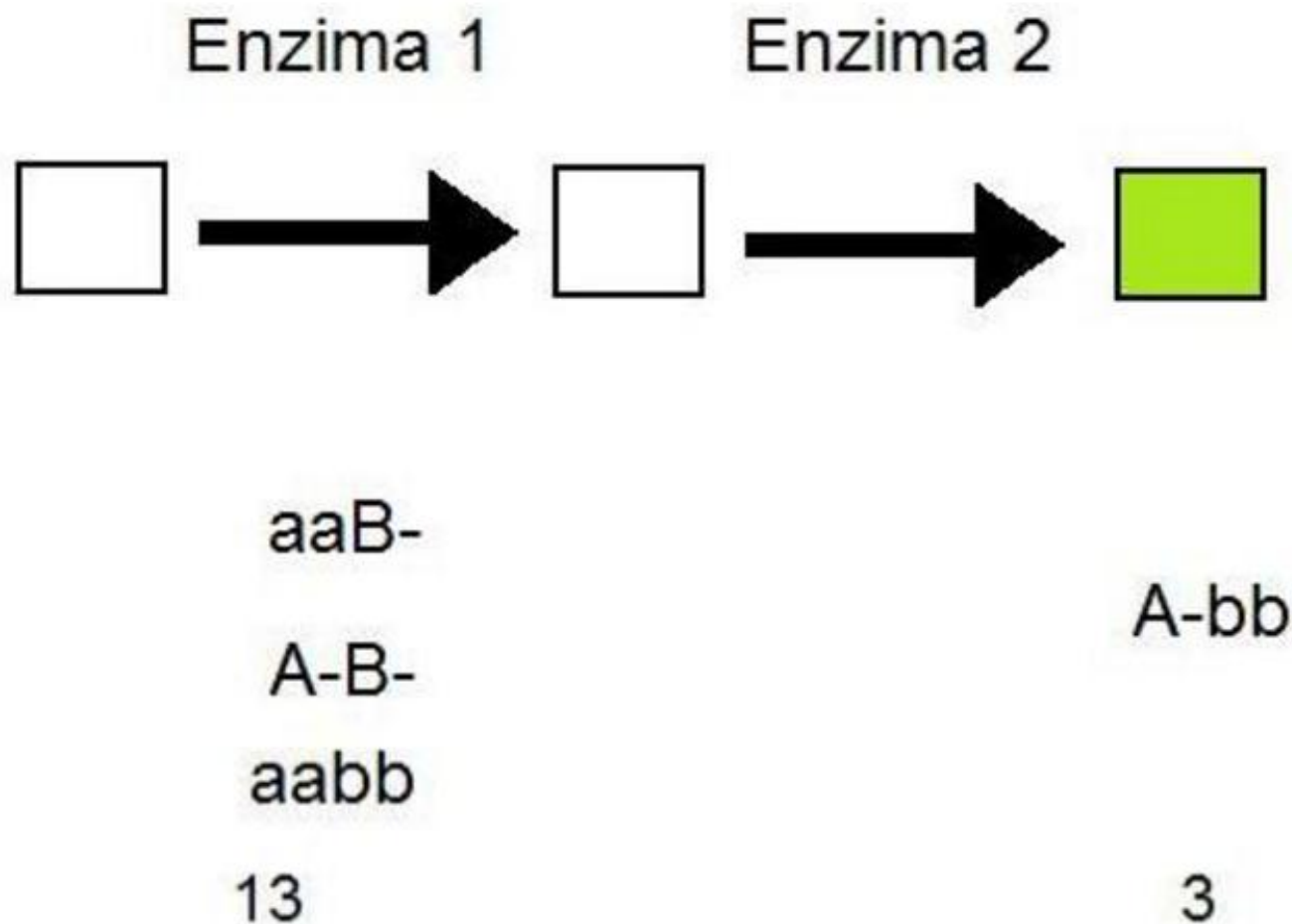


# Epistasi dominante e recessiva

| Case | Organism         | Character    | F <sub>2</sub> Phenotypes |        |         |               | Modified ratio |
|------|------------------|--------------|---------------------------|--------|---------|---------------|----------------|
|      |                  |              | 9/16                      | 3/16   | 3/16    | 1/16          |                |
| 1    | Mouse            | Coat color   | agouti                    | albino | black   | albino        | 9:3:4          |
| 2    | Squash           | Color        | white                     |        | yellow  | green         | 12:3:1         |
| 3    | Pea              | Flower color | purple                    | white  |         |               | 9:7            |
| 4    | Squash           | Fruit shape  | disc                      | sphere |         | long          | 9:6:1          |
| 5    | Chicken          | Color        | white                     |        | colored | white         | 13:3           |
| 6    | Mouse            | Color        | white-spotted             | white  | colored | white-spotted | 10:3:3         |
| 7    | Shepherd's purse | Seed capsule | triangular                |        |         | ovoid         | 15:1           |
| 8    | Flour beetle     | Color        | 6/16 sooty : 3/16 red     | black  | jet     | black         | 6:3:3:4        |

**EPISTASI DOMINANTE E RECESSIVA:** Abbiamo un rapporto di 13:3. Uno dei due geni deve essere omozigote recessivo e l'altro gene necessita di almeno un allele dominante.

## EPISTASI DOMINANTE E RECESSIVA



# Epistasi duplicata codominante

| Case | Organism         | Character    | F <sub>2</sub> Phenotypes |        |         |               | Modified ratio |
|------|------------------|--------------|---------------------------|--------|---------|---------------|----------------|
|      |                  |              | 9/16                      | 3/16   | 3/16    | 1/16          |                |
| 1    | Mouse            | Coat color   | agouti                    | albino | black   | albino        | 9:3:4          |
| 2    | Squash           | Color        | white                     |        | yellow  | green         | 12:3:1         |
| 3    | Pea              | Flower color | purple                    | white  |         |               | 9:7            |
| 4    | Squash           | Fruit shape  | disc                      | sphere |         | long          | 9:6:1          |
| 5    | Chicken          | Color        | white                     |        | colored | white         | 13:3           |
| 6    | Mouse            | Color        | white-spotted             | white  | colored | white-spotted | 10:3:3         |
| 7    | Shepherd's purse | Seed capsule | triangular                |        |         | ovoid         | 15:1           |
| 8    | Flour beetle     | Color        | 6/16 sooty : 3/16 red     | black  | jet     | black         | 6:3:3:4        |

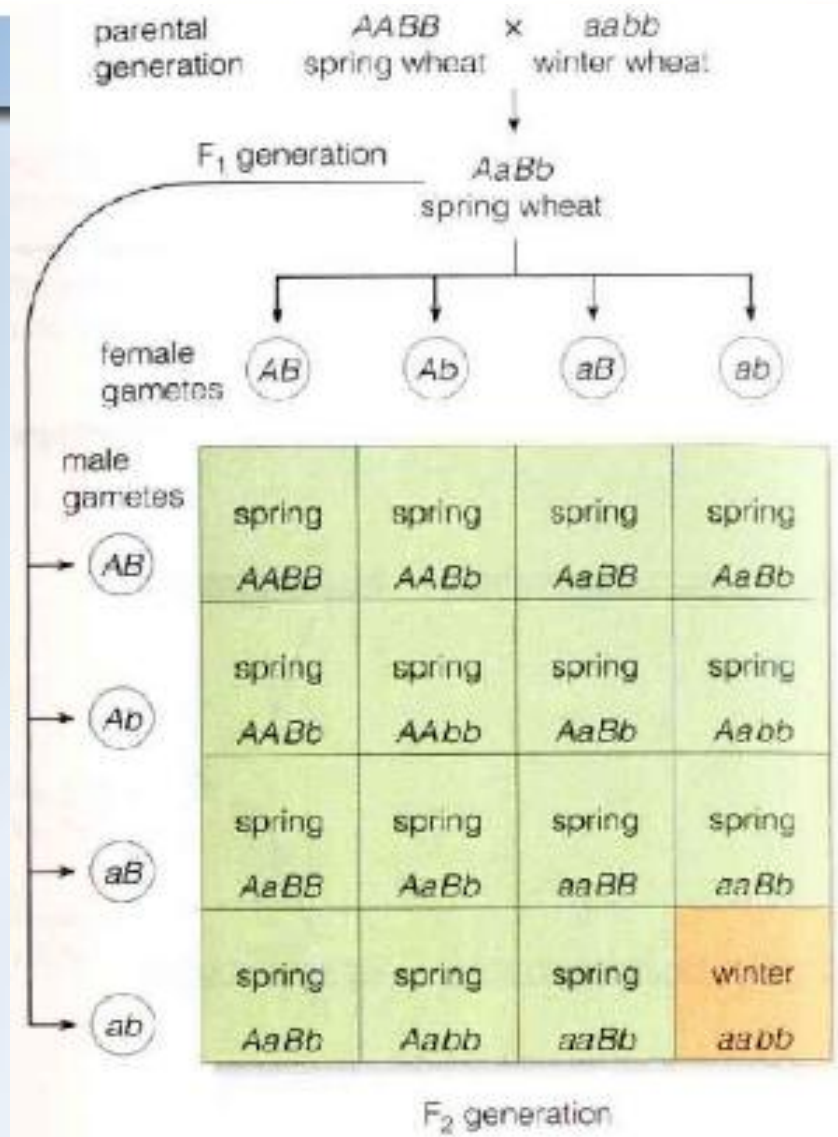
# Duplicazione genica

| Case | Organism         | Character    | F <sub>2</sub> Phenotypes |        |         |               | Modified ratio |
|------|------------------|--------------|---------------------------|--------|---------|---------------|----------------|
|      |                  |              | 9/16                      | 3/16   | 3/16    | 1/16          |                |
| 1    | Mouse            | Coat color   | agouti                    | albino | black   | albino        | 9:3:4          |
| 2    | Squash           | Color        | white                     |        | yellow  | green         | 12:3:1         |
| 3    | Pea              | Flower color | purple                    | white  |         |               | 9:7            |
| 4    | Squash           | Fruit shape  | disc                      | sphere |         | long          | 9:6:1          |
| 5    | Chicken          | Color        | white                     |        | colored | white         | 13:3           |
| 6    | Mouse            | Color        | white-spotted             | white  | colored | white-spotted | 10:3:3         |
| 7    | Shepherd's purse | Seed capsule | triangular                |        |         | ovoid         | 15:1           |
| 8    | Flour beetle     | Color        | 6/16 sooty : 3/16 red     | black  | jet     | black         | 6:3:3:4        |

# Epistasi duplicata dominante

- il fenotipo dominante è determinato dalla presenza di almeno 1 allele dominante in uno di 2 loci genici
- solo il doppio omozigote recessivo esprime il fenotipo recessivo

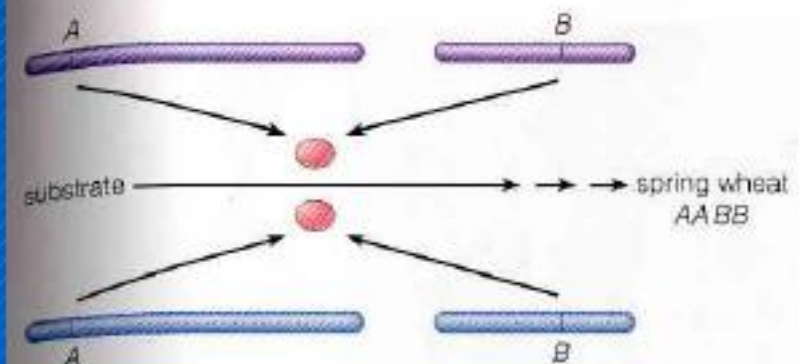
Es. crescita del grano  
 A, B: tipo estivo  
 aabb: tipo invernale



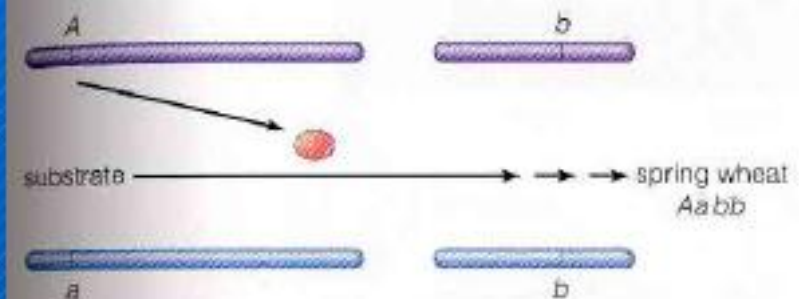
15  
16  $A\_B\_$   $aaB\_$   $A\_bb$ : spring wheat  
 1  
16  $aa\ bb$ : winter wheat

# Poliploidia

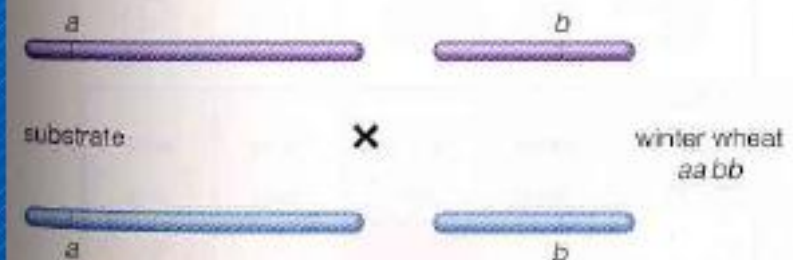
- i due geni codificano per lo stesso prodotto, sufficiente ad esprimere il fenotipo anche in singola dose



**a** Dominant alleles at loci *A* and *B* encode the same product, which causes wheat to follow the spring pattern of growth.



**b** A single dominant allele at either locus produces enough product for the dominant phenotype.



**c** Plants that are doubly homozygous recessive do not produce the product and instead follow the winter pattern of growth.

# Epistasi e codominanza

| Case | Organism         | Character    | F <sub>2</sub> Phenotypes |        |         |               | Modified ratio |
|------|------------------|--------------|---------------------------|--------|---------|---------------|----------------|
|      |                  |              | 9/16                      | 3/16   | 3/16    | 1/16          |                |
| 1    | Mouse            | Coat color   | agouti                    | albino | black   | albino        | 9:3:4          |
| 2    | Squash           | Color        | white                     |        | yellow  | green         | 12:3:1         |
| 3    | Pea              | Flower color | purple                    | white  |         |               | 9:7            |
| 4    | Squash           | Fruit shape  | disc                      | sphere |         | long          | 9:6:1          |
| 5    | Chicken          | Color        | white                     |        | colored | white         | 13:3           |
| 6    | Mouse            | Color        | white-spotted             | white  | colored | white-spotted | 10:3:3         |
| 7    | Shepherd's purse | Seed capsule | triangular                |        |         | ovoid         | 15:1           |
| 8    | Flour beetle     | Color        | 6/16 sooty : 3/16 red     | black  | jet     | black         | 6:3:3:4        |

# Bombay (epistasi)

$$I^A I^B Hh \times I^A I^B Hh$$

Svolgete questo incrocio e calcolate i fenotipi

Troverete un rapporto fenotipico 6:3:3:4

Due ceppi puri di zucche che producono frutto giallo,  $Y_1$  e  $Y_2$ , sono incrociati con un ceppo puro di zucche che produce frutti verdi,  $G_1$ , e tra loro. Si ottengono i seguenti risultati:

| Incrocio | P                                  | $F_1$        | $F_2$                                         |
|----------|------------------------------------|--------------|-----------------------------------------------|
| I        | $Y_1$ (giallo)<br>x $G_1$ (verde)  | Tutti gialli | $\frac{3}{4}$ giallo : $\frac{1}{4}$ verde    |
| II       | $Y_2$ (giallo)<br>x $G_1$ (verde)  | Tutti verdi  | $\frac{3}{4}$ verde : $\frac{1}{4}$ giallo    |
| III      | $Y_1$ (giallo)<br>x $Y_2$ (giallo) | Tutti gialli | $\frac{13}{16}$ giallo : $\frac{3}{16}$ verde |

- Esamine i risultati di ogni incrocio e prevedete quanti geni sono responsabili per la determinazione del colore del frutto nelle zucche. Giustificate la risposta.
- Usando simboli chiaramente definiti di vostra scelta, fornite i genotipi delle piante parentali,  $F_1$ ,  $F_2$  in ogni incrocio.
- Se le  $F_1$  degli incroci I e II sono incrociate, prevedete il rapporto fenotipico della  $F_2$ .

I lieviti sono organismi eucarioti unicellulari che crescono in coltura sia come aploidi sia come diploidi. Lieviti diploidi sono generati quando due ceppi aploidi si fondono. Sette ceppi aploidi mostrano tipi di crescita simili: a 25°C, tutti i ceppi crescono normalmente, ma a 37°C, i ceppi mostrano capacità di crescita diverse. La tabella sottostante mostra il pattern di crescita.

| Crescita del ceppo |  | A                  | B | C | D | E | F | G |
|--------------------|--|--------------------|---|---|---|---|---|---|
| 25°C               |  | ●                  | ● | ● | ● | ● | ● | ● |
| 37°C               |  | ○                  | ● | ○ | ○ | ○ | ○ | ● |
|                    |  | ● Crescita normale |   |   |   |   |   |   |
|                    |  | ● Crescita lenta   |   |   |   |   |   |   |
|                    |  | ○ Nessuna crescita |   |   |   |   |   |   |

A) Descrivete la natura della mutazione che colpisce ciascuno di questi ceppi mutanti di lievito. Spiegate perché i ceppi B e G mostrano differenti abilità di crescita a 37°C rispetto agli altri ceppi.

Ciascuno dei lieviti mutanti aploidi è fuso con uno degli altri e i diploidi risultanti sono testati per la loro capacità di crescere a 37°C. I risultati dell'esperimento sono mostrati di seguito.

Ceppo

|   | A | B | C | D | E | F | G |
|---|---|---|---|---|---|---|---|
| A | ○ |   |   |   |   |   |   |
| B | ● | ● |   |   |   |   |   |
| C | ● | ● | ○ |   |   |   |   |
| D | ○ | ● | ● | ○ |   |   |   |
| E | ● | ● | ○ | ● | ○ |   |   |
| F | ○ | ● | ● | ○ | ● | ○ |   |
| G | ● | ● | ● | ● | ● | ● | ● |

# VERIFICA 1

## In the ABO blood types, which offspring is *not* possible?

a. An individual with type O blood from a mother with type A and a father with type AB.

Correct. To have a child with type O blood, both parents must have an  $I^O$  allele to donate. The mother can be  $I^A/I^O$ , but the father is  $I^A/I^B$  and has no  $I^O$  allele to donate.

b. An individual with type AB blood from a mother with type B and a father with type AB.

Incorrect. The father is  $I^A/I^B$ , and the mother will have at least one  $I^B$  allele to donate. The father must donate the  $I^A$  allele.

c. An individual with type A blood from a mother with type O and a father with type AB.

Incorrect. The mother is  $I^O/I^O$  and can donate only an  $I^O$  allele, and the father is  $I^A/I^B$  and can donate an  $I^A$  allele.

d. An individual with type B blood from a mother with type A and a father with type AB.

Incorrect. The father is  $I^A/I^B$  and can donate an  $I^B$ , and the mother can be  $I^A/I^O$  and donate the  $I^O$  allele.

# VERIFICA 2

**Which of the following statements is always true when mutations occur in genes whose products are essential to an organism's survival?**

a. The homozygous recessive is a lethal combination.

Correct. If the gene product is essential for survival, having two mutant alleles means that the organism will not produce the gene product at all.

b. Reversion of the mutation must occur for the organism to survive.

Incorrect. Typically, when an individual is heterozygous, enough of the essential product is produced for survival. However, sometimes the mutation behaves as a dominant lethal allele.

c. The heterozygote always has a normal phenotype.

Incorrect. This may or may not be true. Even in cases where the heterozygote individual appears normal, there may be a decreased level of gene product.

d. One normal copy of the gene will allow survival.

Incorrect. There are some cases in which the mutation behaves as a dominant lethal allele.

# VERIFICA 3

The following interactions are found in mice. The *albino* allele (*a*) eliminates all color in homozygotes and is recessive to the normal pigment allele (*A*). The *black* allele (*b*) causes the normal gray-brown agouti coat color to be much darker and is recessive to the normal agouti allele (*B*). A mouse of genotype *Aabb* will:

a. Have a black coat

Correct. Because the *albino* allele is recessive to normal coloring and this mouse has an *A* allele, it will have coloring. Because the mouse is *bb*, it will be black.

b. Show an epistatic interaction

Incorrect. If this mouse were *aa*, it would be an albino regardless of the combination of *B* and *b* alleles, and the *aa* alleles would be epistatic to the *bb* alleles. However, the mouse is not *aa*.

c. Be an albino

Incorrect. Only an *aa* combination will produce an albino mouse.

d. Have an agouti coat

Incorrect. Although the *Aa* genotype will provide normal coloration, the *bb* genotype will produce a darker than normal mouse.

# VERIFICA 3

**A ratio of 12:3:1 is most characteristic of a cross involving:**

a. 1 gene pair

Incorrect. If there were only one pair of genes involved in the expression of a trait, the ratio would add up to 4, such as 1:2:1.

b. 2 gene pairs

Correct. A single trait that is expressed in 16 parts (12:3:1) suggests that two gene pairs are interacting.

c. 3 gene pairs

Incorrect. If there were three gene pairs involved (a trihybrid cross), the ratio would add up to 64.

d. 4 gene pairs

Incorrect. If there were four gene pairs involved, the ratio would add up to 256.

# VERIFICA 5

**What is the expected phenotypic ratio of a cross between a disc-shaped squash that is heterozygous at both loci and a long squash?**

a. 9/16 disc, 6/16 sphere, 1/16 long

Incorrect. This is the expected ratio of two disc-shaped squash that are heterozygous at both loci.

b. 9/16 disc, 6/16 long, 1/16 sphere

Incorrect. Long squash have all recessive alleles at both loci and sphere-shaped squash have a dominant allele at only one locus.

c. 4/16 disc, 8/16 sphere, 4/16 long

Correct. The genotypic ratio for this cross is 4/16 *AB*, 4/16 *Ab*, 4/16 *aB*, and 4/16 *ab*, making the phenotypic ratio 4/16 disc, 8/16 sphere, 4/16 long.

d. 4/16 disc, 8/16 long, 4/16 sphere

Incorrect. Long squash have all recessive alleles at both loci, and sphere-shaped squash have a dominant allele at only one locus.