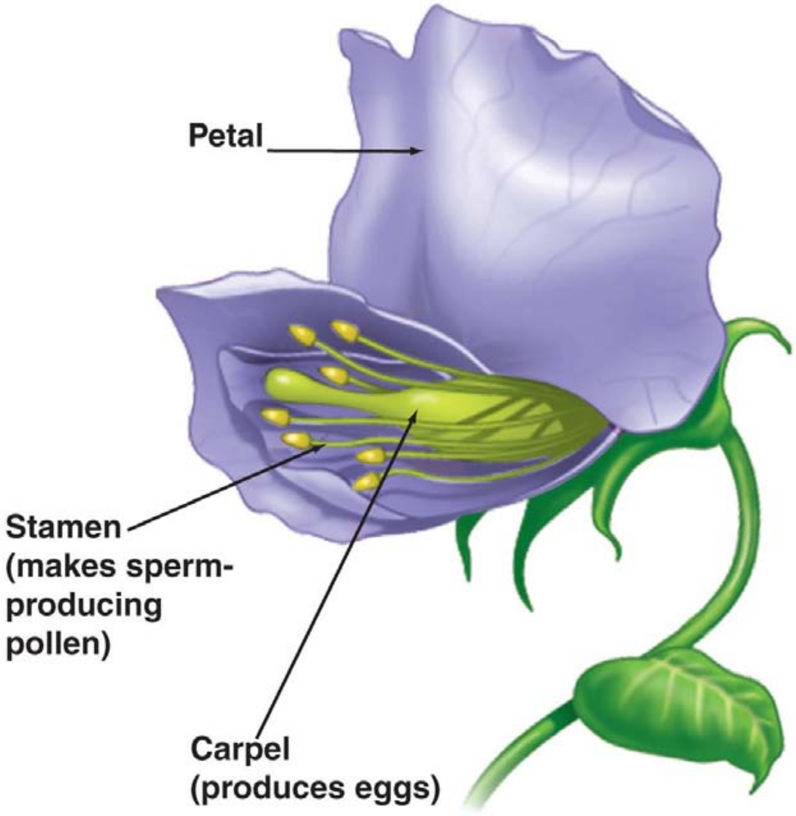
Genetics

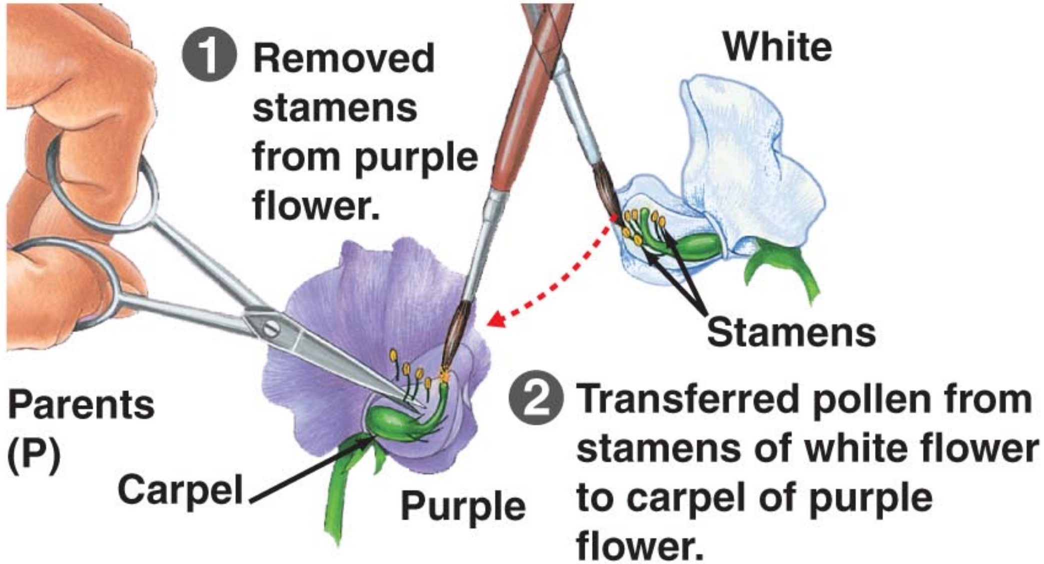
Bio 11

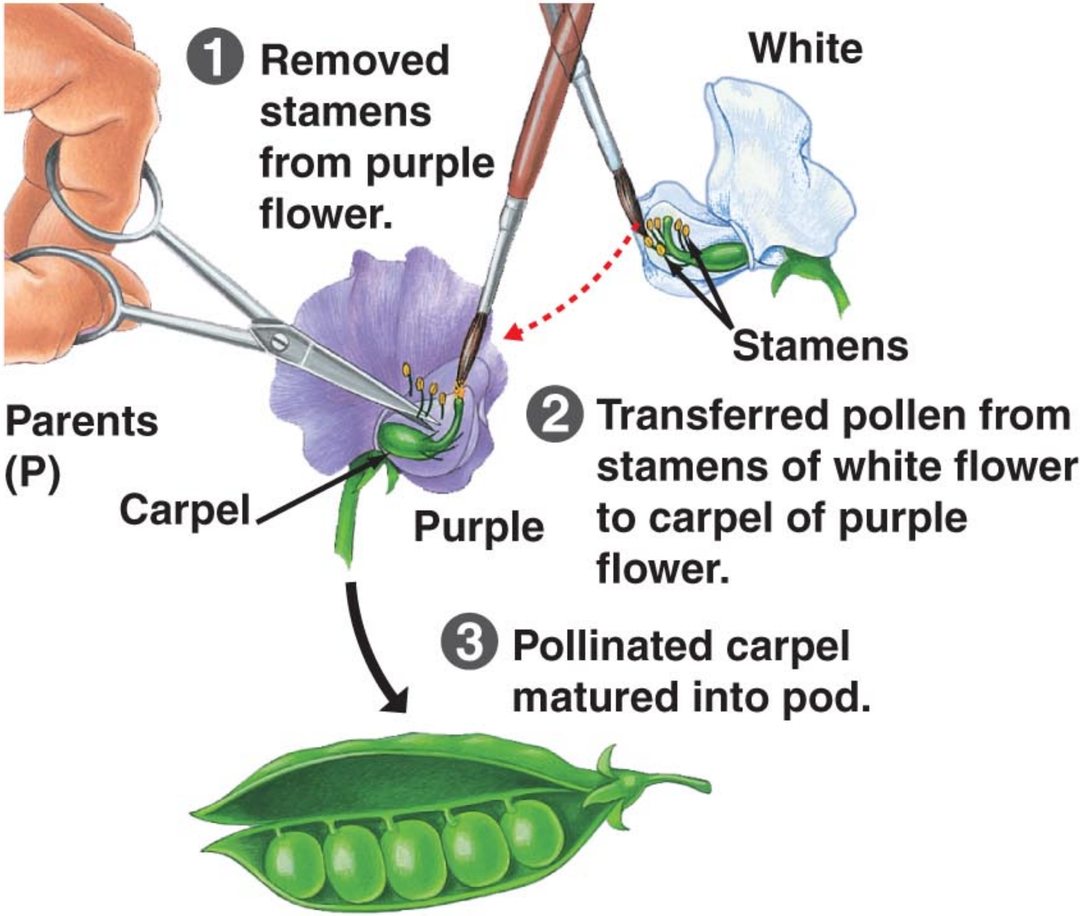
Mendel
Father of Genetics
- A little history

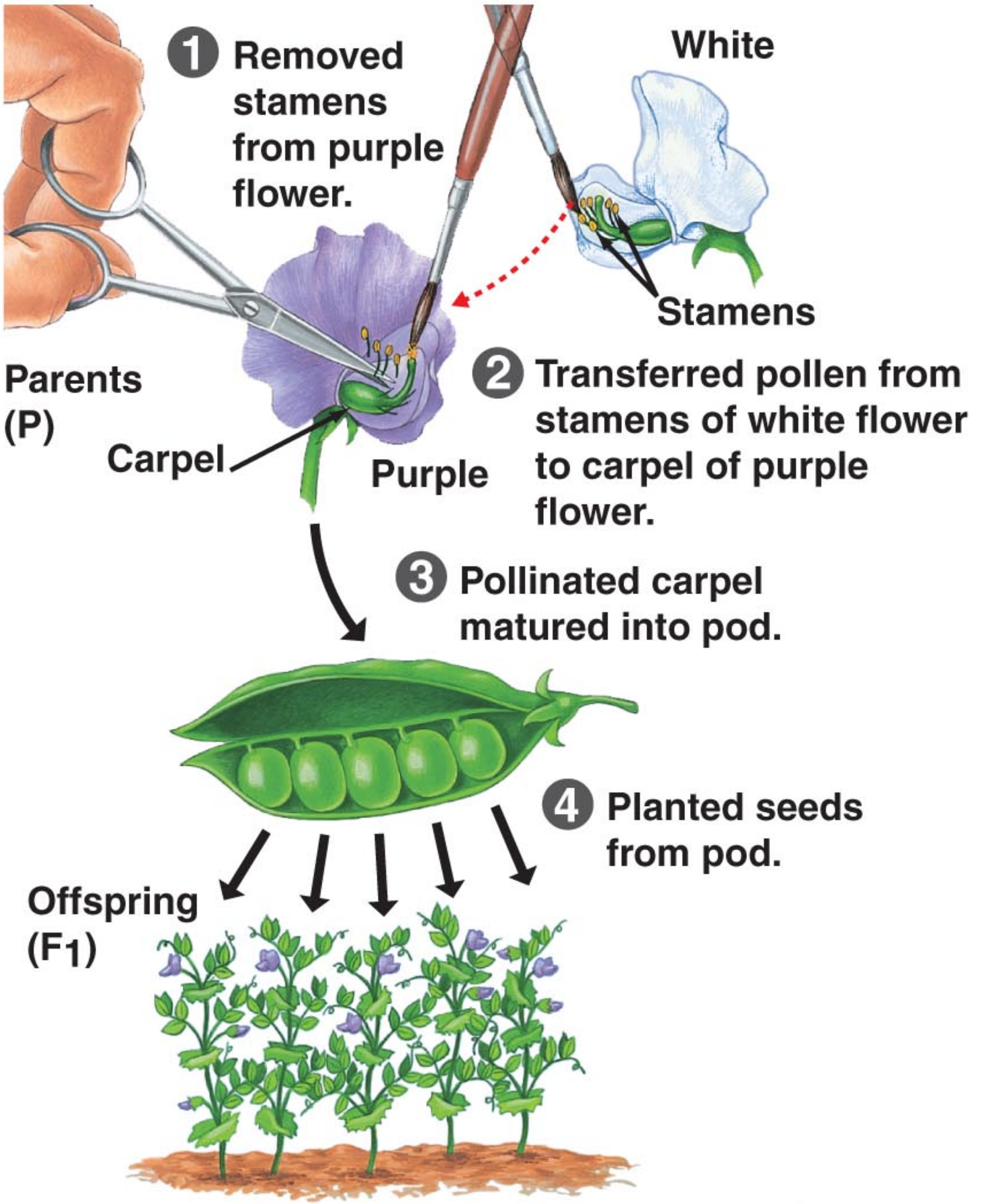
Definitions

- **Homologous Chromosomes**: Chromosomes that are the same length and have the same genes, but not necessarily the same alleles.
- **Genes**: DNA that encodes for a function such as eye color.
- **Alleles**: versions of genes.
- **Genotype**:The allelic makeup (which alleles organisms has)**.**
- **Phenotype**: The appearance of an organism**.**


Petal
Stamen
(makes sperm-
producing
pollen)
Carpel
(produces eggs)


1 Removed stamens from purple flower.
White
Stamens
2 Transferred pollen from stamens of white flower to carpel of purple flower.
Parents (P)
Carpel
Purple


1 Removed stamens from purple flower.
White
Stamens
Parents (P)
Carpel
Purple
2 Transferred pollen from stamens of white flower to carpel of purple flower.
3 Pollinated carpel matured into pod.



1 Removed stamens from purple flower.
White
Stamens
Parents (P)
Carpel
Purple
2 Transferred pollen from stamens of white flower to carpel of purple flower.
3 Pollinated carpel matured into pod.
4 Planted seeds from pod.
Offspring (F_1)

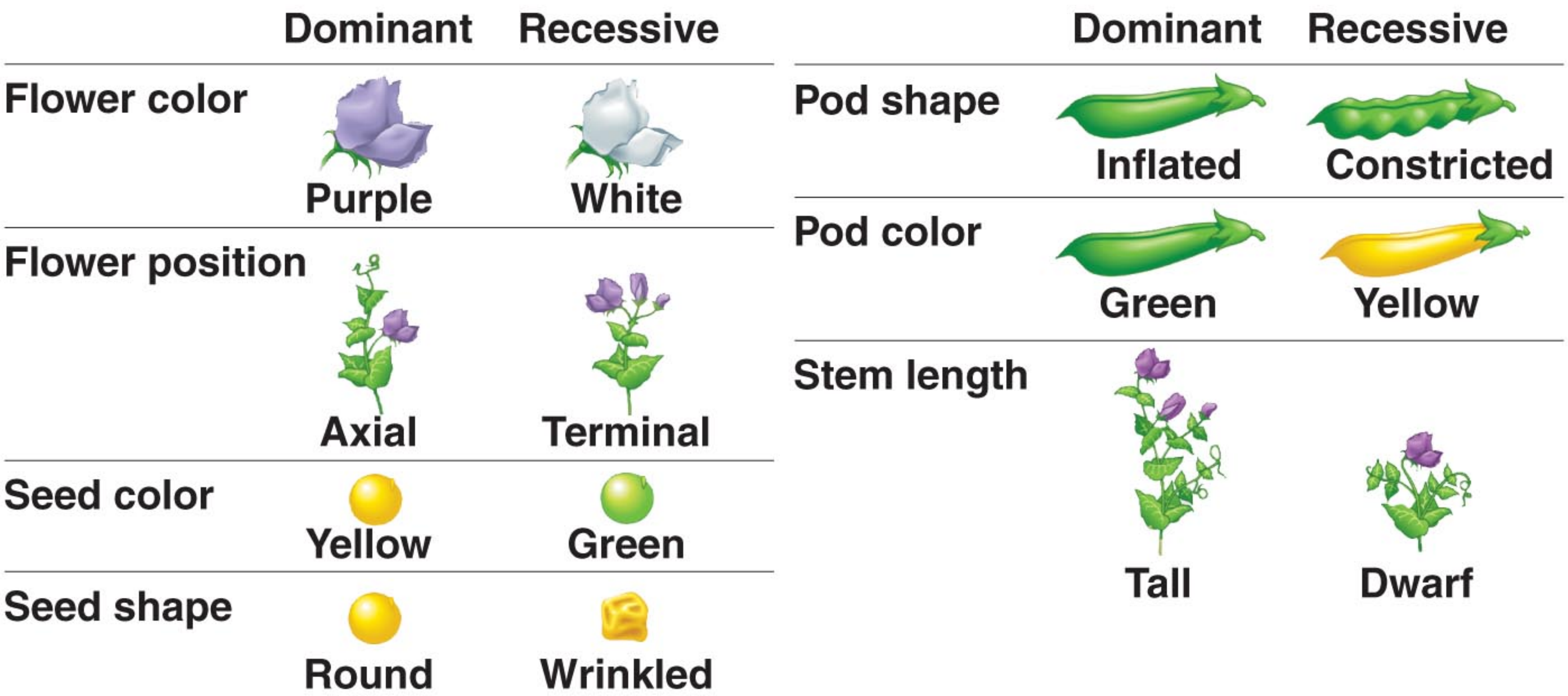
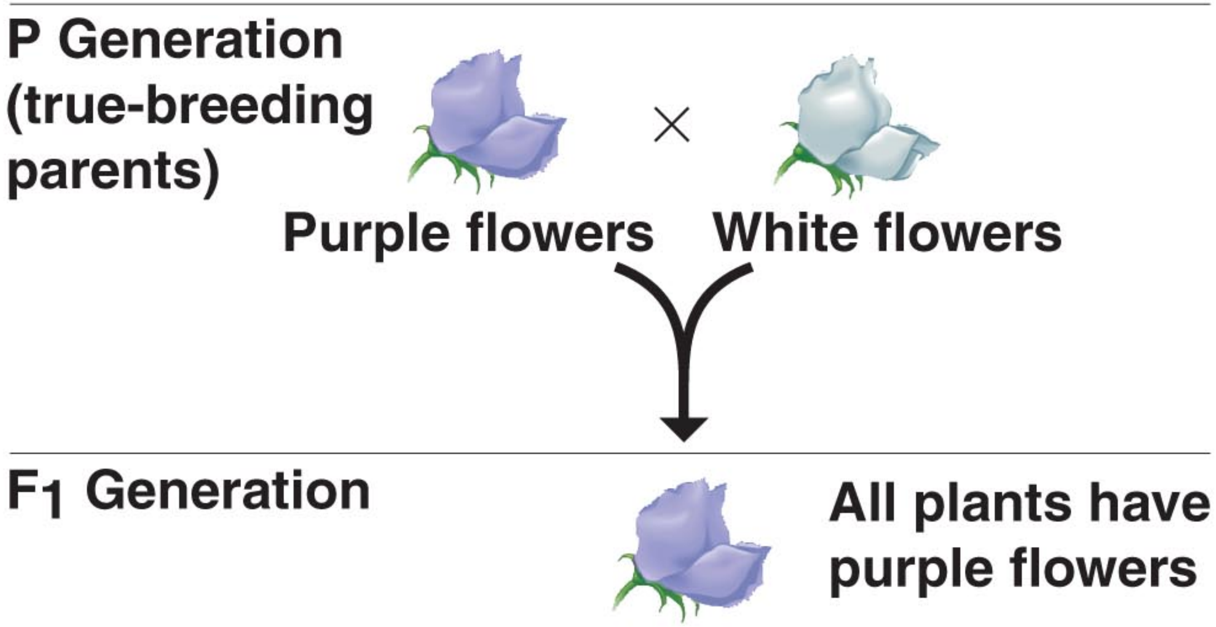

Dominant
Recessive
Flower color
Purple
White
Flower position
Axial
Terminal
Seed color
Yellow
Green
Seed shape
Round
Wrinkled
Dominant
Recessive
Pod shape
Inflated
Constricted
Pod color
Green
Yellow
Stem length
Tall
Dwarf

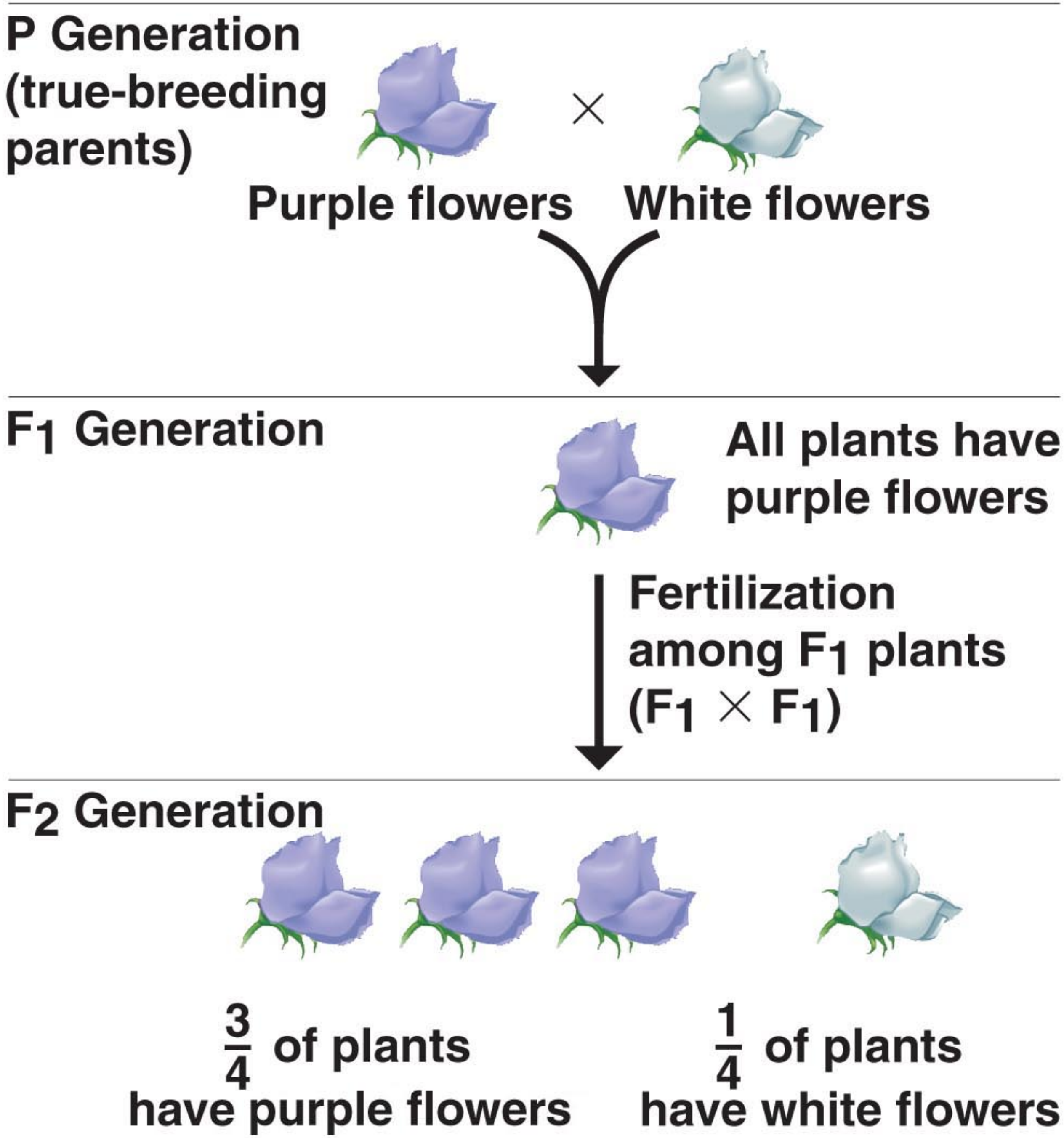
Table 11.2 Ratios of Dominant to Recessive in Mendel's Plants

Dominant trait	**Recessive trait**	**Ratio of dominant to recessive in F_2 generation**
Smooth seed	Wrinkled seed	2.96:1 (5,474 smooth, 1,850 wrinkled)
Yellow seed	Green seed	3.01:1 (6,022 yellow, 2,001 green)
Inflated pod	Wrinkled pod	2.95:1 (882 inflated, 299 wrinkled)
Green pod	Yellow pod	2.82:1 (428 green, 152 yellow)
Purple flower	White flower	3.14:1 (705 purple, 224 white)
Flower on stem	Flower at tip	3.14:1 (651 along stem, 207 at tip)
Tall stem	Dwarf stem	2.84:1 (787 tall plants, 277 dwarfs)
	Average ratio, all traits:	**3:1**

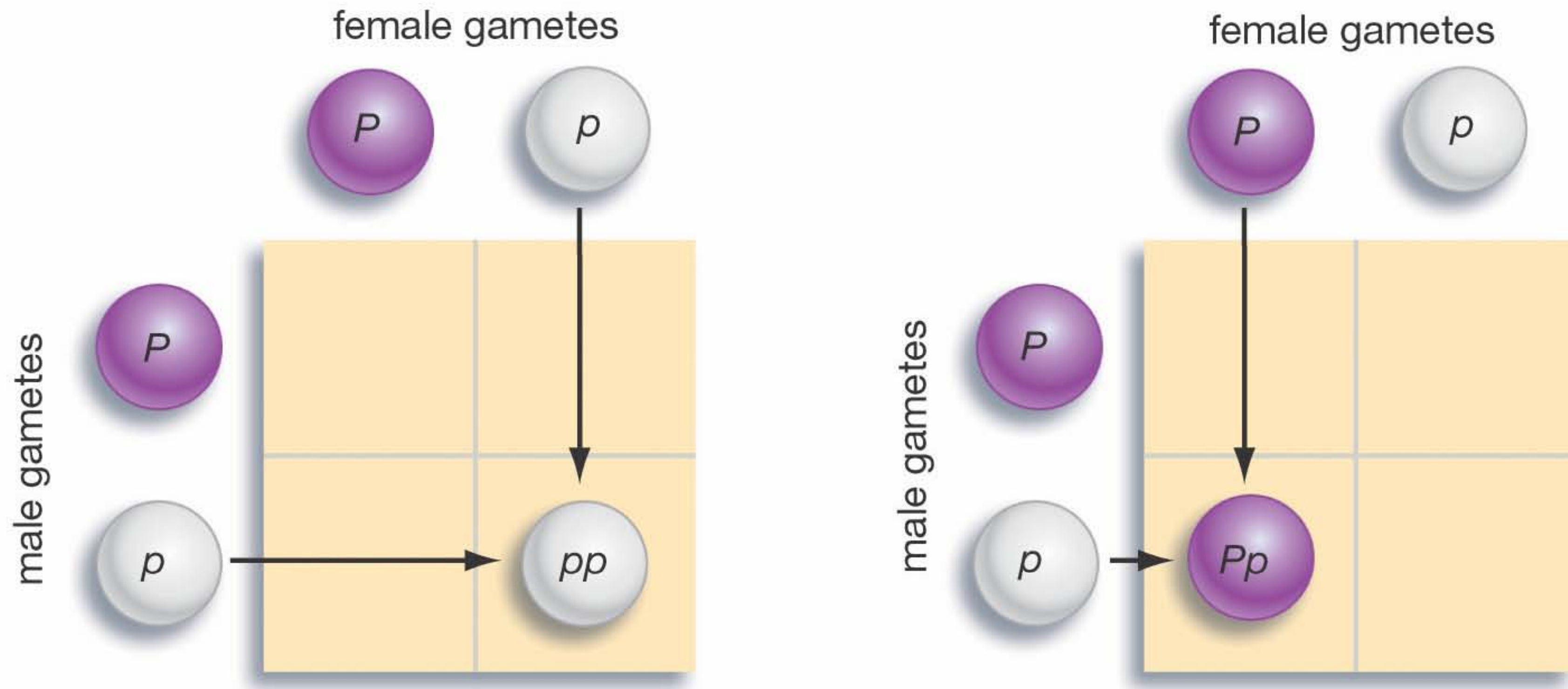
P Generation
(true-breeding
parents)

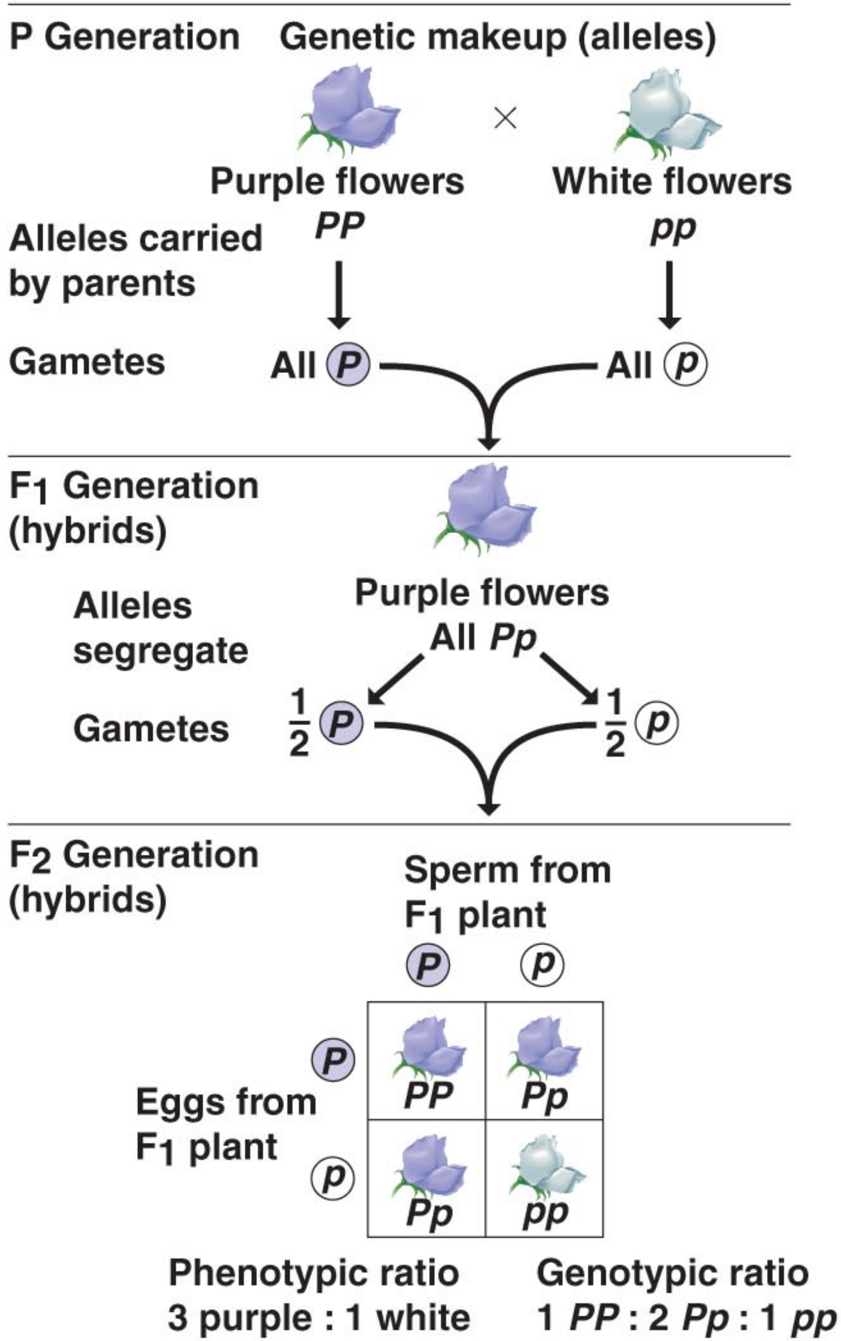


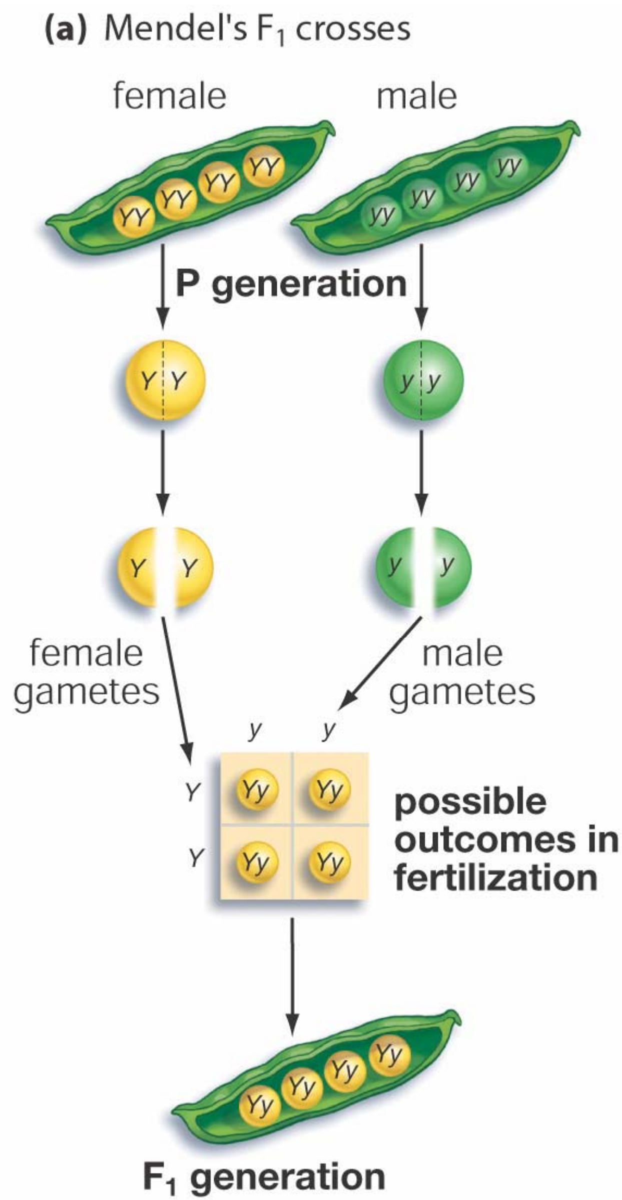
P Generation
(true-breeding
parents)
×
Purple flowers
White flowers
F1 Generation
All plants have
purple flowers

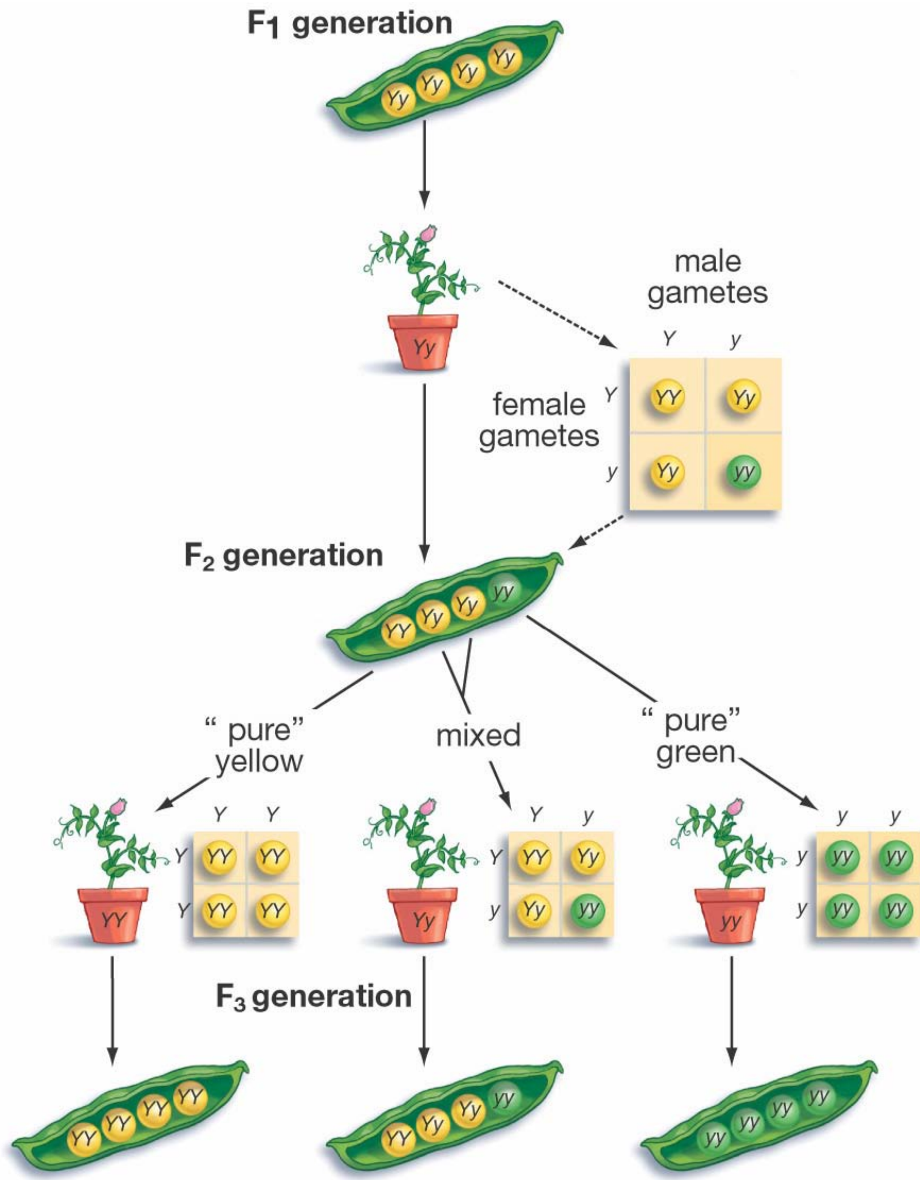

P Generation
(true-breeding
parents)
×
Purple flowers
White flowers
F_1 Generation
All plants have
purple flowers
Fertilization
among F_1 plants
($F_1 \times F_1$)
F_2 Generation
$\frac{3}{4}$ of plants
have purple flowers
$\frac{1}{4}$ of plants
have white flowers

(b) How to read a Punnett square

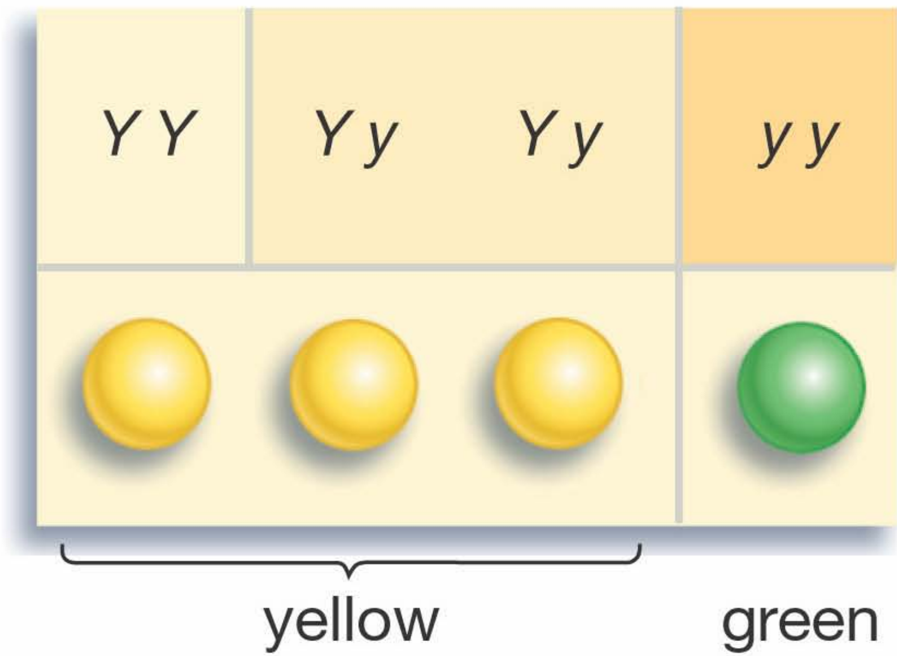



P Generation
Genetic makeup (alleles)
×
Purple flowers
PP
White flowers
pp
Alleles carried by parents
Gametes
All P
All p
F1 Generation (hybrids)
Purple flowers
All Pp
Alleles segregate
Gametes
1/2 P
1/2 p
F2 Generation (hybrids)
Sperm from F1 plant
P
p
Eggs from F1 plant
P
p
PP
Pp
Pp
pp
Phenotypic ratio
3 purple : 1 white
Genotypic ratio
1 PP : 2 Pp : 1 pp


(a) Mendel's F1 crosses
female
male
YY
yy
P generation
Y Y
y y
Y Y
y y
female gametes
male gametes
y
y
Y
Y
Yy
possible outcomes in fertilization
F1 generation



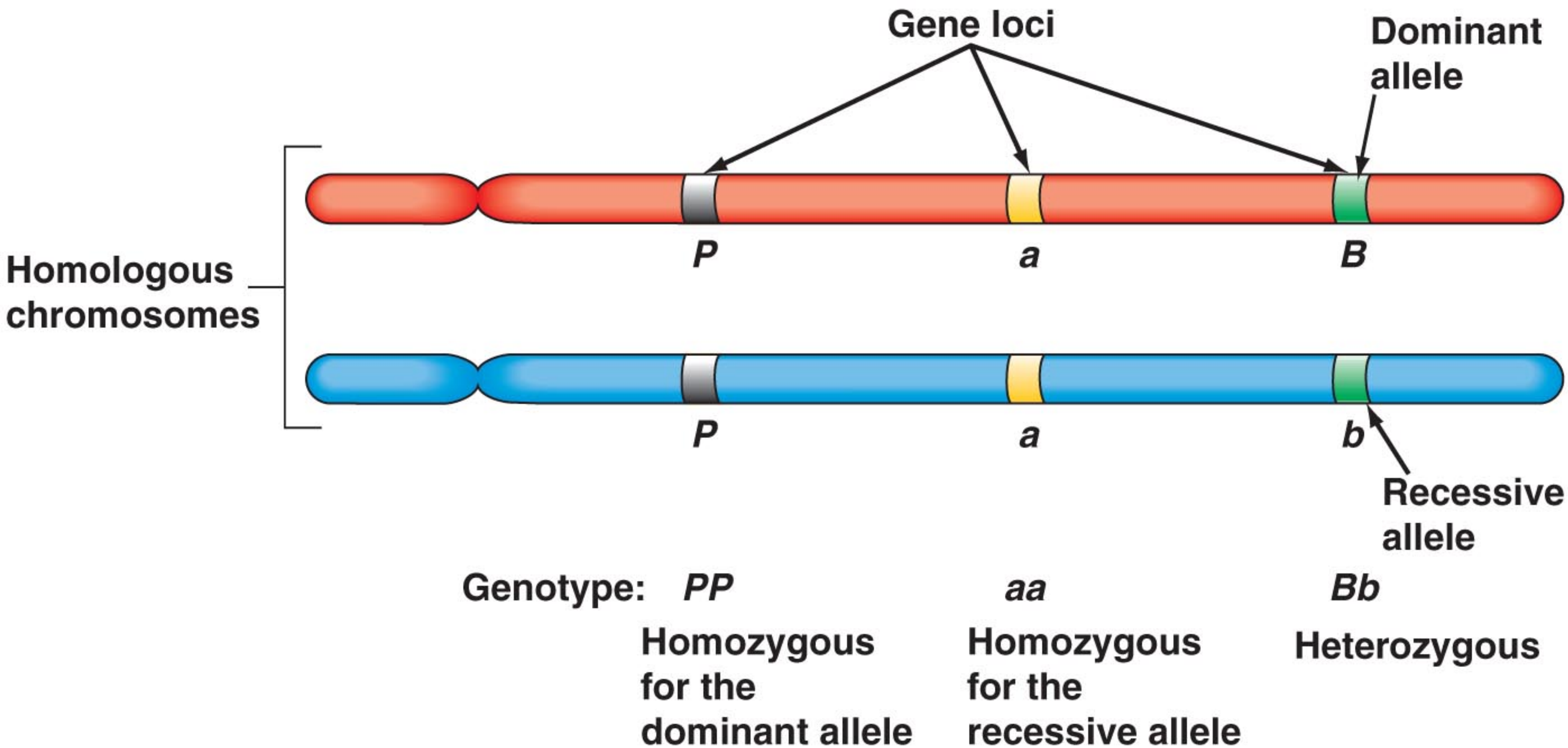
F1 generation
Yy
male gametes
female gametes
Y
y
YY
Yy
yy
F2 generation
"pure" yellow
mixed
"pure" green
F3 generation

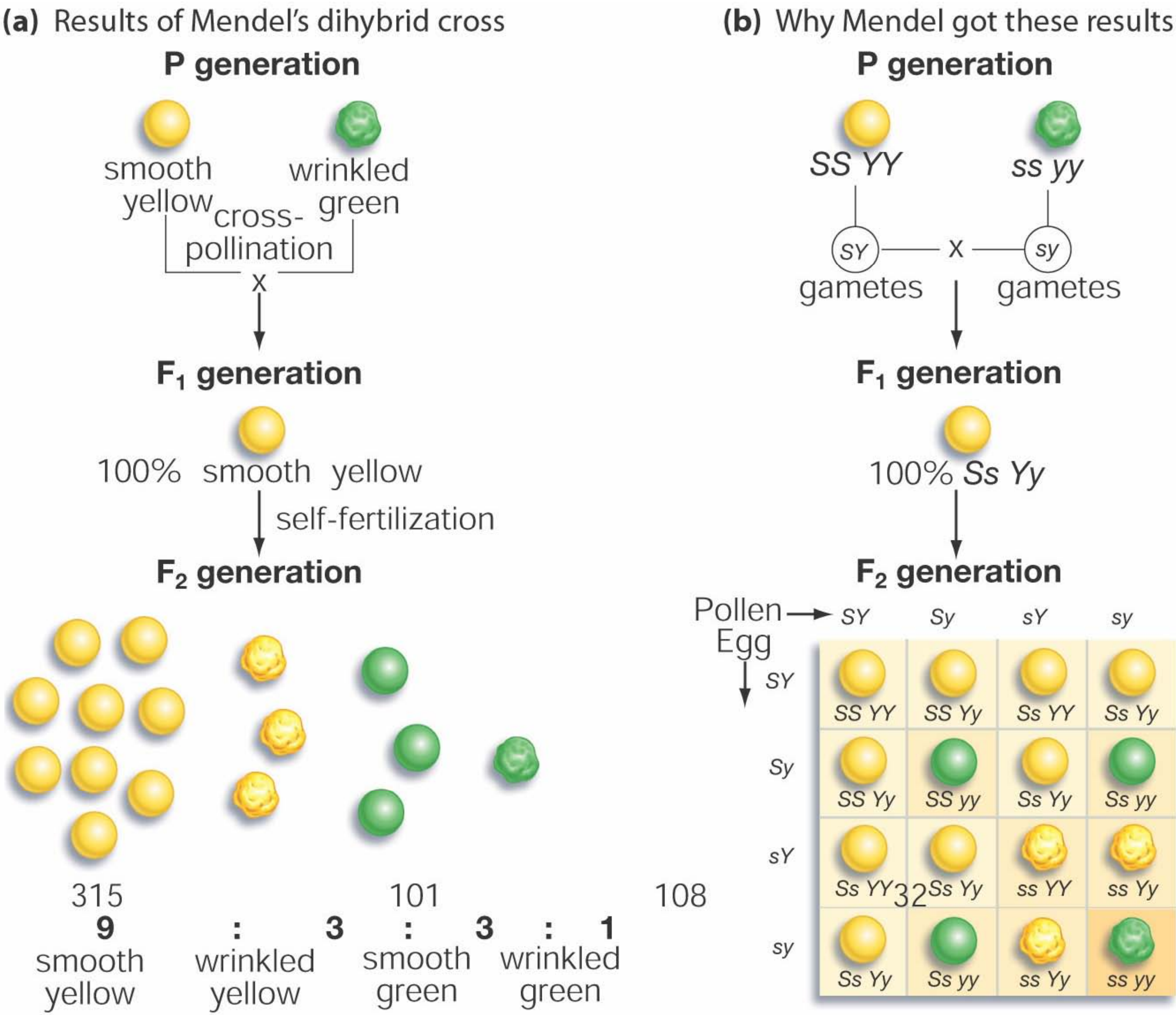


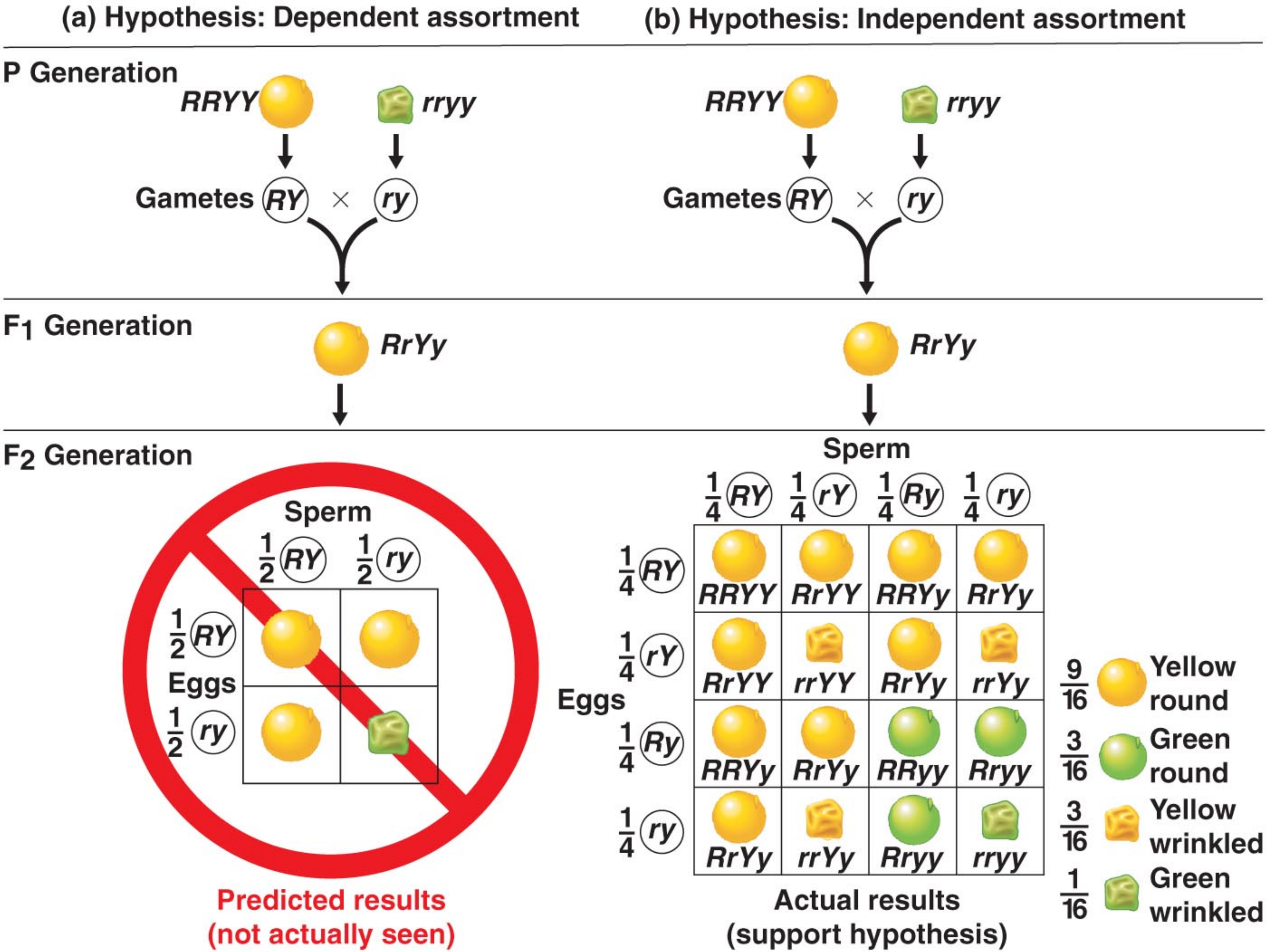
Three genotypes yield . . .

two phenotypes.

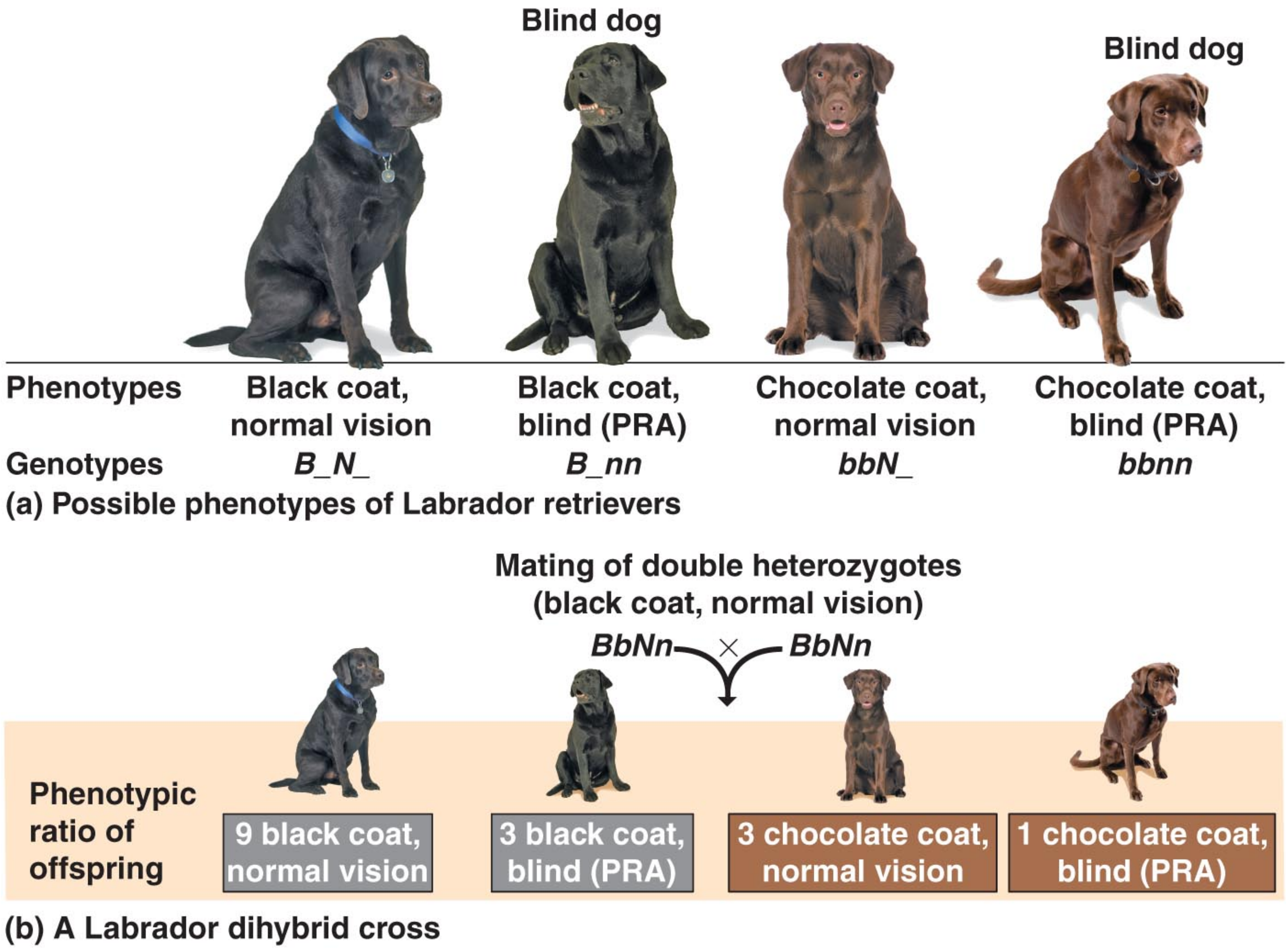

Some of these peas have a smooth texture, while others are wrinkled.

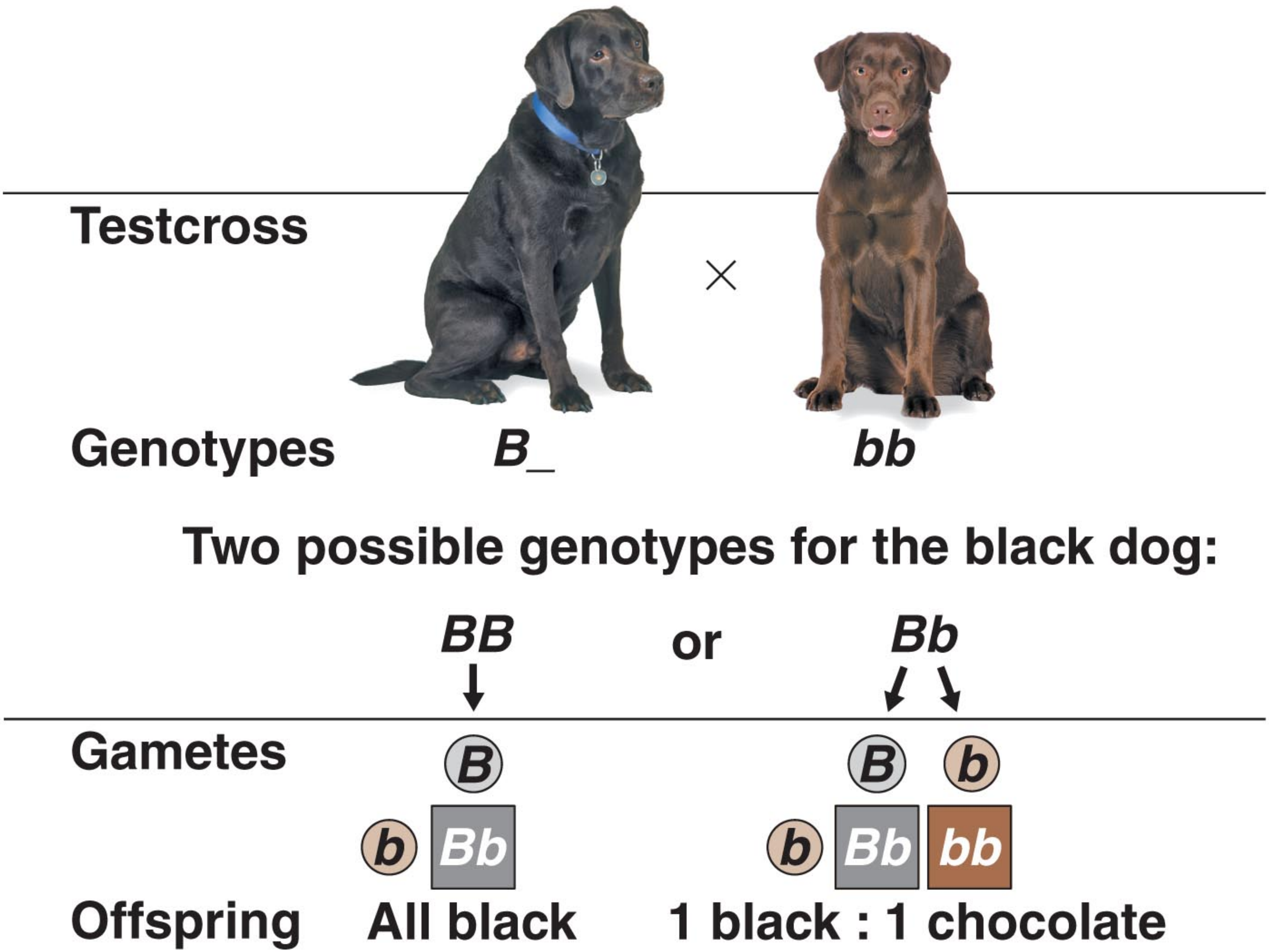

Gene loci
Dominant allele
Homologous chromosomes
P
a
B
P
a
b
Recessive allele
Genotype: PP
aa
Bb
Homozygous for the dominant allele
Homozygous for the recessive allele
Heterozygous

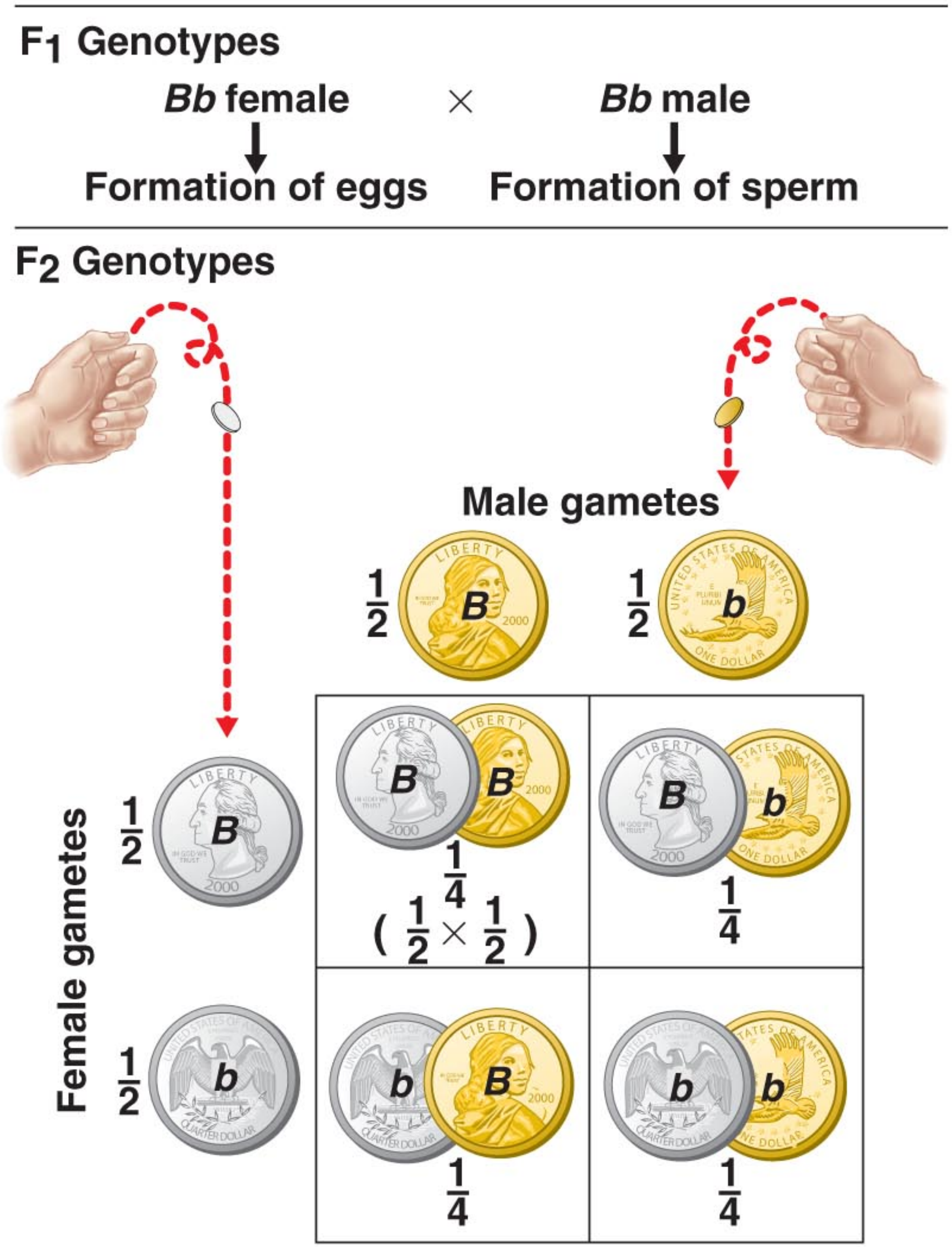

(a) Results of Mendel's dihybrid cross
P generation
smooth yellow
wrinkled green
cross-pollination
x
F1 generation
100% smooth yellow
self-fertilization
F2 generation
315
101
108
9 : 3 : 3 : 1
smooth yellow
wrinkled yellow
smooth green
wrinkled green
(b) Why Mendel got these results
P generation
SS YY
ss yy
SY
sy
x
gametes
gametes
F1 generation
100% Ss Yy
F2 generation
Pollen
Egg
SY
Sy
sY
sy
SS YY
SS Yy
Ss YY
Ss Yy
SS Yy
SS yy
Ss Yy
Ss yy
Ss YY
Ss Yy
ss YY
ss Yy
Ss Yy
Ss yy
ss Yy
ss yy

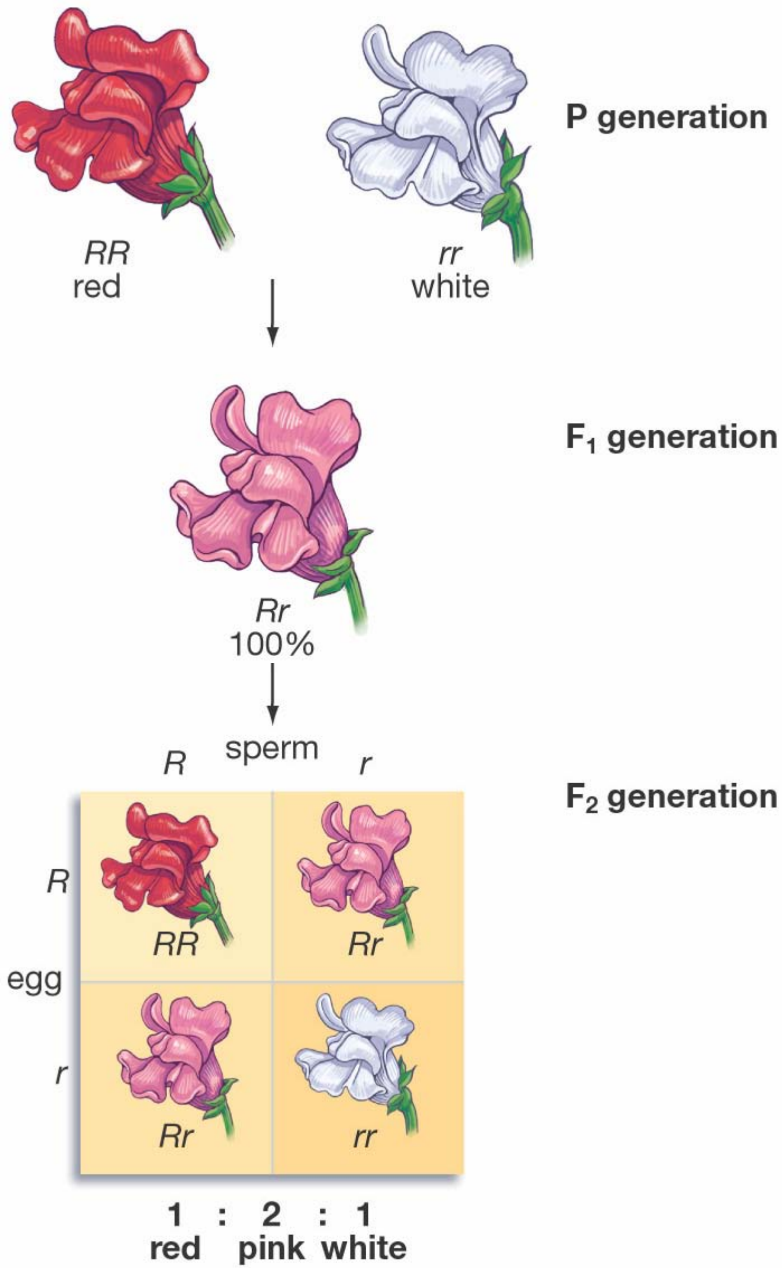


(a) Hypothesis: Dependent assortment
(b) Hypothesis: Independent assortment
P Generation
RRYY
rryy
Gametes
RY
×
ry
F1 Generation
RrYy
F2 Generation
Sperm
1/2 RY
1/2 ry
Eggs
1/2 RY
1/2 ry
Predicted results
(not actually seen)
Sperm
1/4 RY
1/4 rY
1/4 Ry
1/4 ry
Eggs
1/4 RY
1/4 rY
1/4 Ry
1/4 ry
RRYY
RrYY
RRYy
RrYy
RrYY
rrYY
RrYy
rrYy
RRYy
RrYy
RRyy
Rryy
RrYy
rrYy
Rryy
rryy
Actual results
(support hypothesis)
9/16 Yellow round
3/16 Green round
3/16 Yellow wrinkled
1/16 Green wrinkled

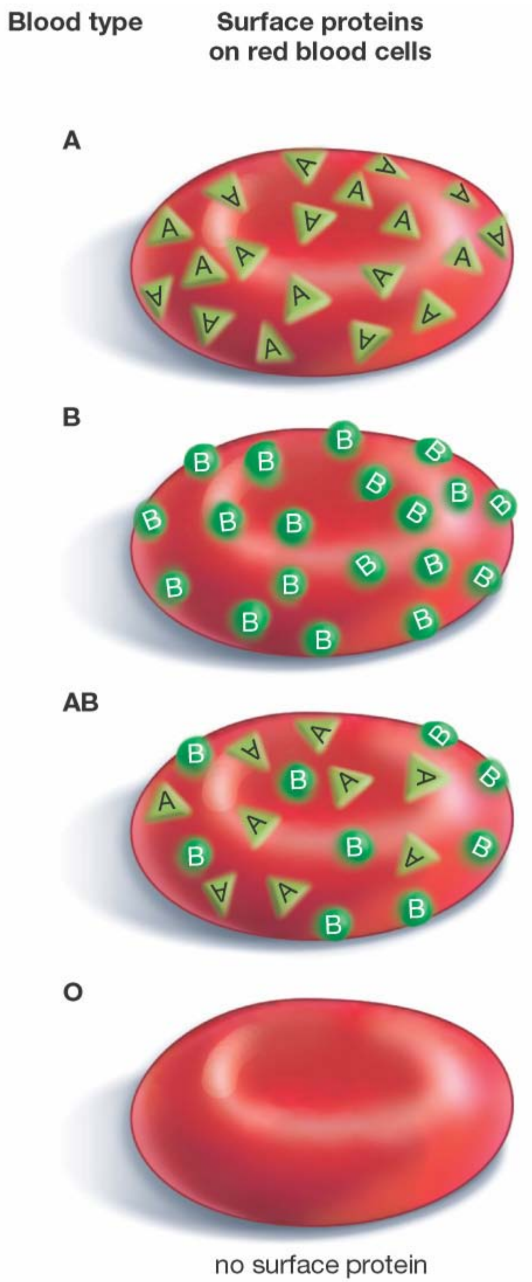

Blind dog
Blind dog
Phenotypes
Black coat, normal vision
Black coat, blind (PRA)
Chocolate coat, normal vision
Chocolate coat, blind (PRA)
Genotypes
B_N_
B_nn
bbN_
bbnn
(a) Possible phenotypes of Labrador retrievers
Mating of double heterozygotes (black coat, normal vision)
BbNn × BbNn
Phenotypic ratio of offspring
9 black coat, normal vision
3 black coat, blind (PRA)
3 chocolate coat, normal vision
1 chocolate coat, blind (PRA)
(b) A Labrador dihybrid cross


Testcross
×
Genotypes
B_
bb
Two possible genotypes for the black dog:
BB
or
Bb
Gametes
B
B
b
b
Bb
b
Bb
bb
Offspring
All black
1 black : 1 chocolate

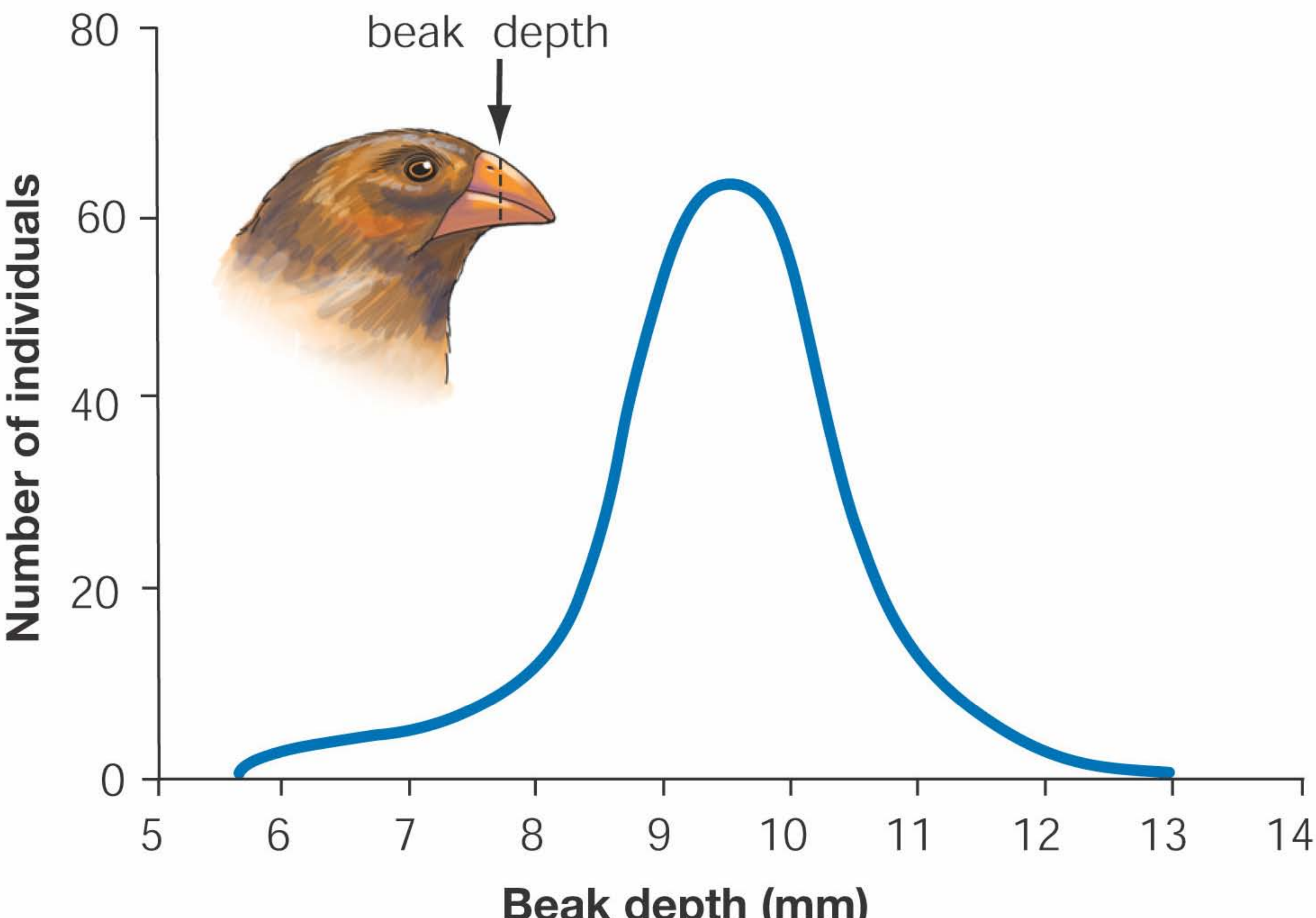

F1 Genotypes
Bb female
×
Bb male
Formation of eggs
Formation of sperm
F2 Genotypes
Male gametes
1/2
B
1/2
b
Female gametes
1/2
B
1/2
b
B B
1/4
(1/2 × 1/2)
B b
1/4
b B
1/4
b b
1/4

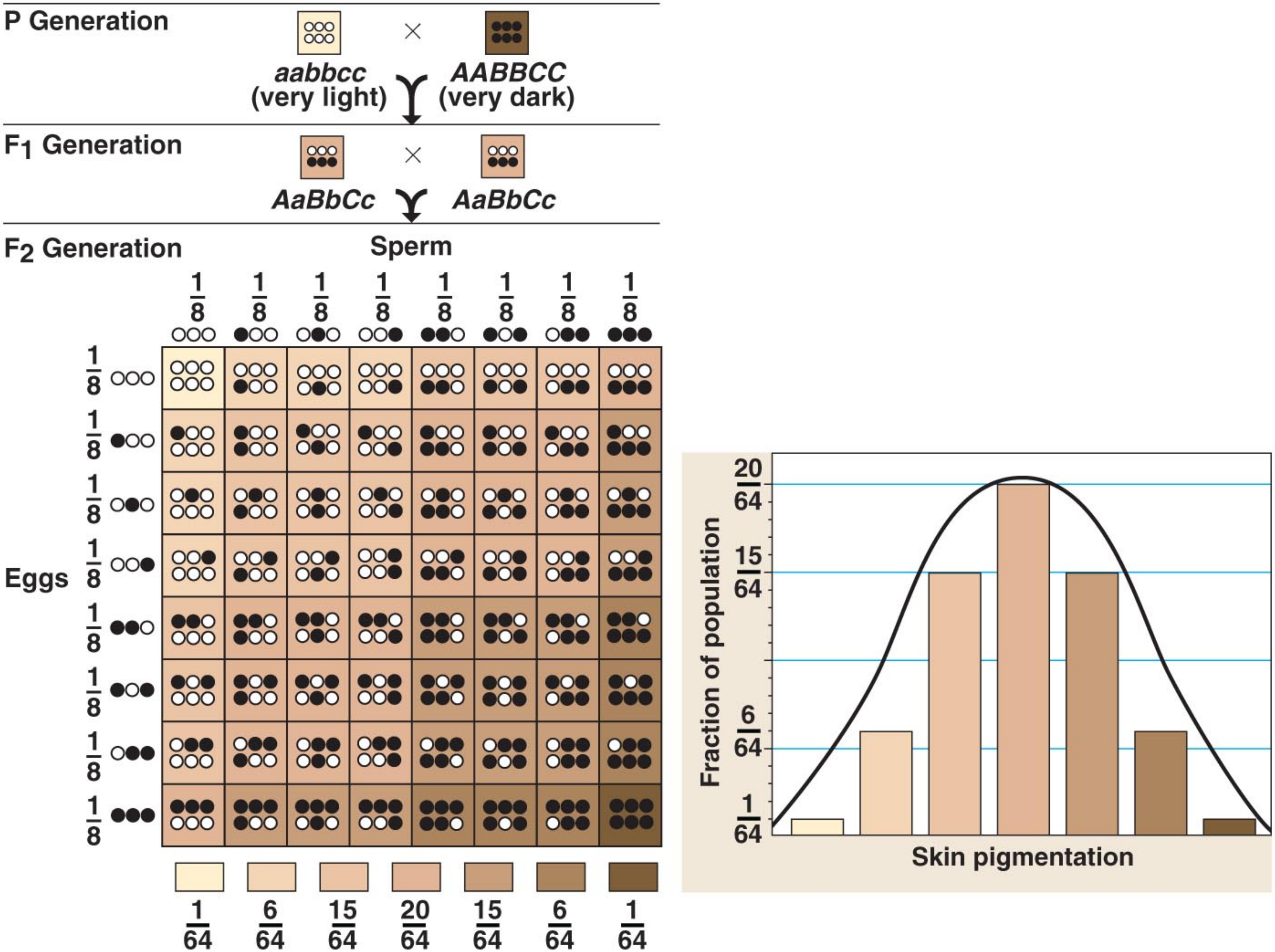


RR
red
rr
white
P generation
Rr
100%
F1 generation
sperm
R
r
F2 generation
R
egg
r
RR
Rr
Rr
rr
1 : 2 : 1
red pink white

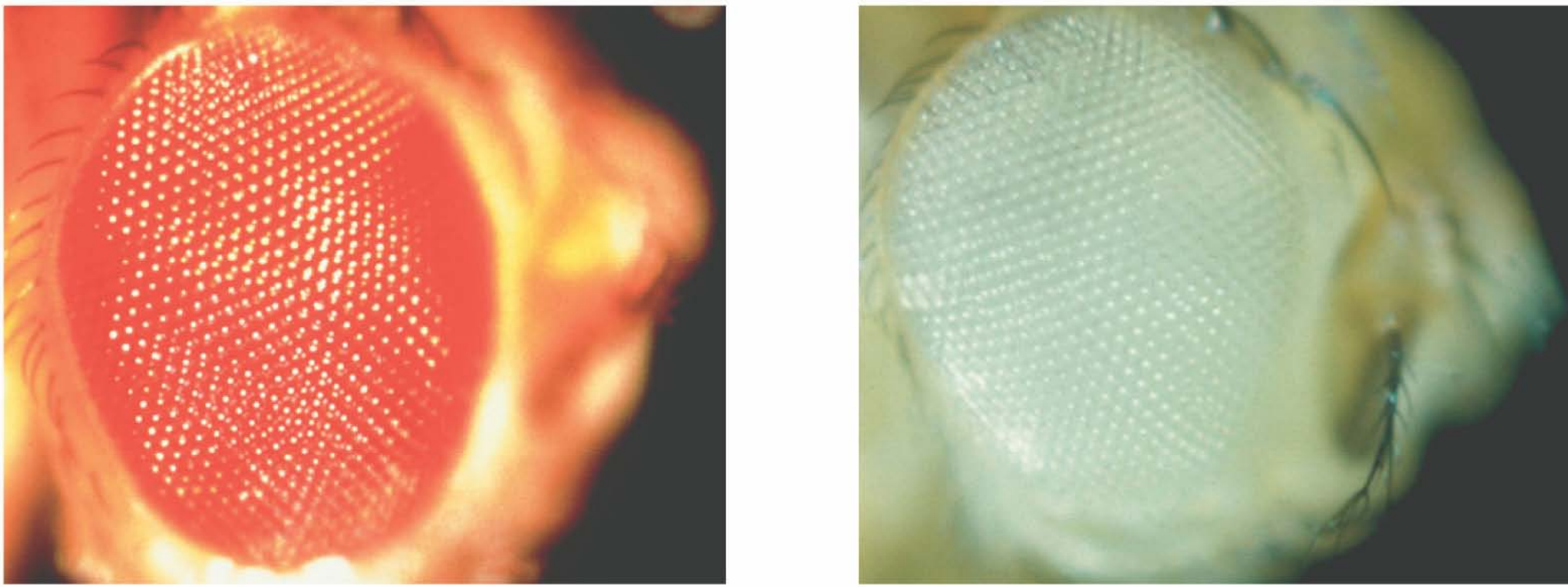

Blood type
Surface proteins on red blood cells
A
B
AB
O
no surface protein

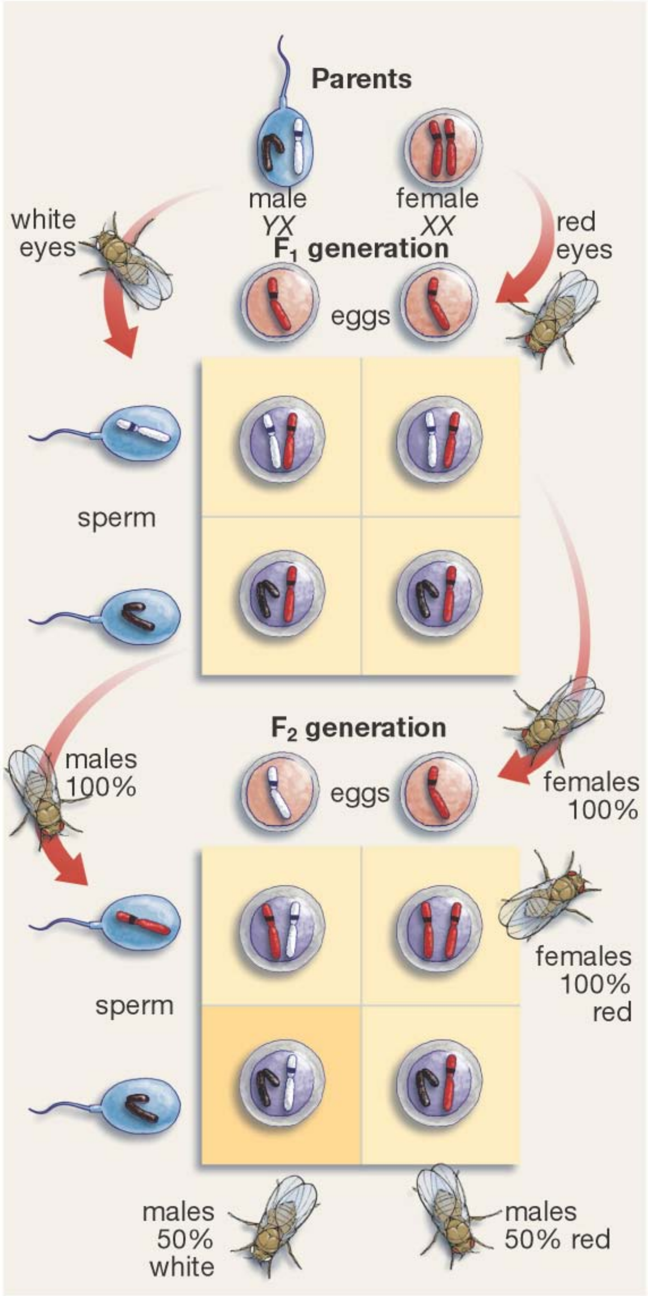
(b) The bell curve

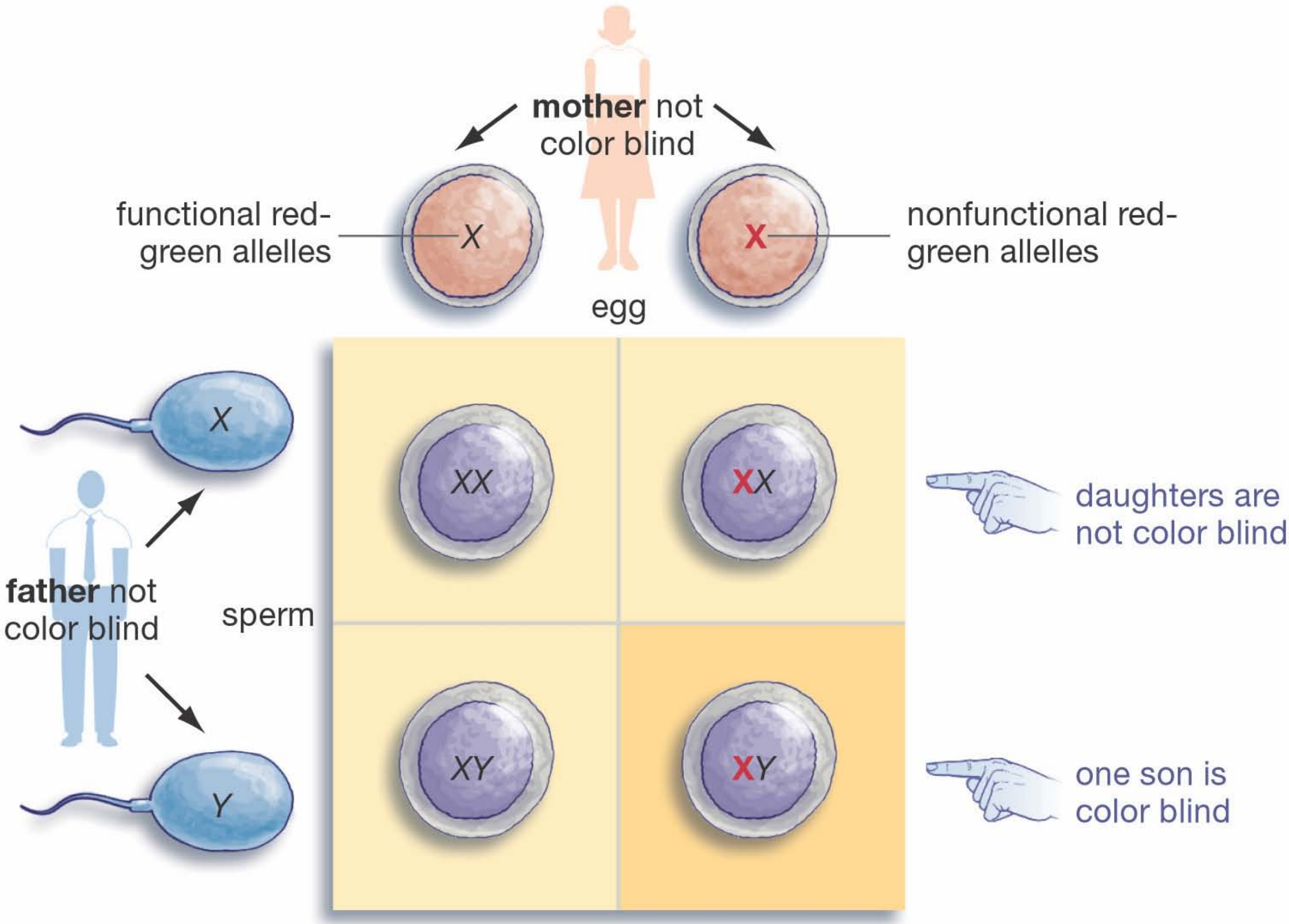


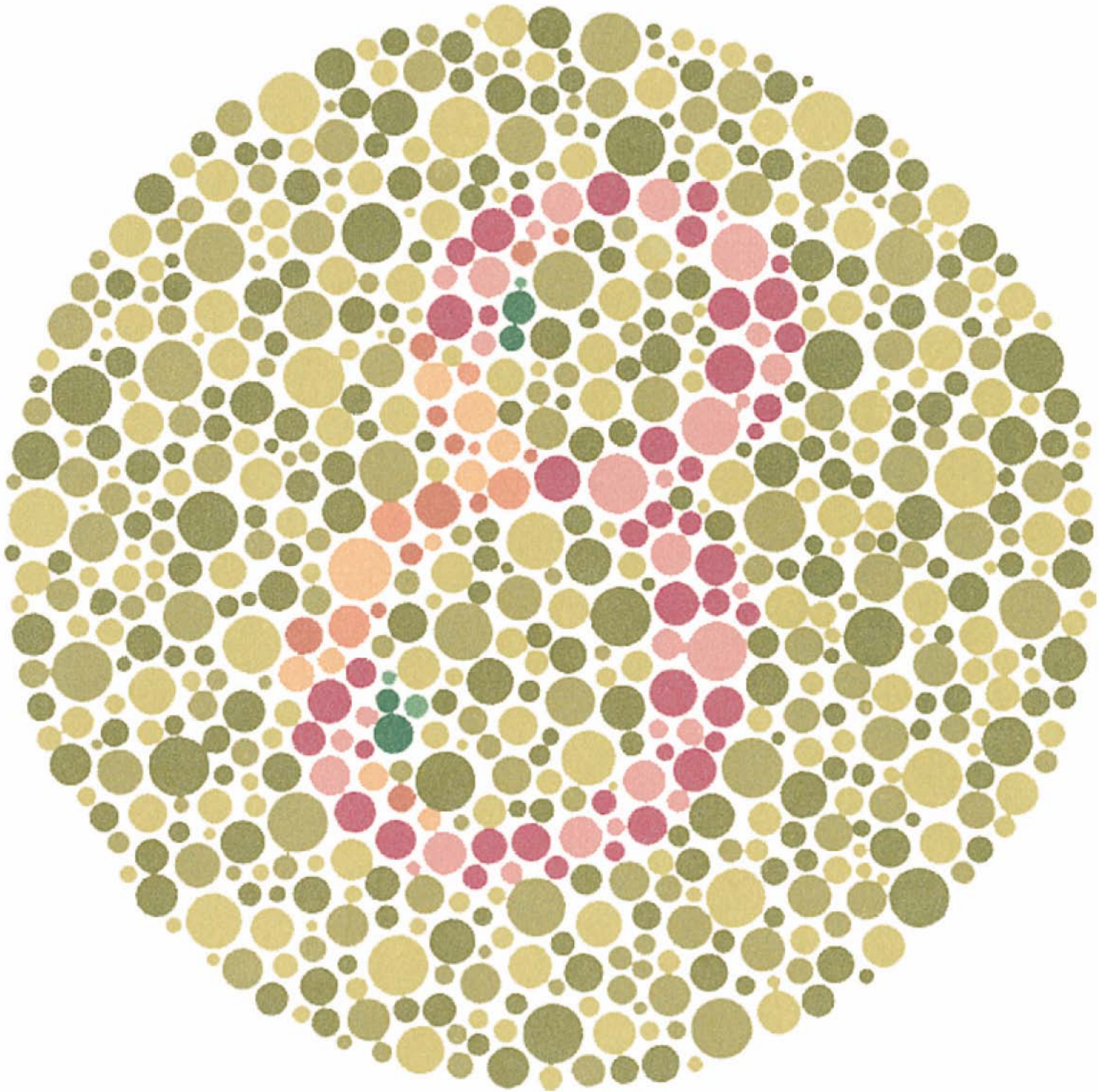

P Generation
aabbcc
(very light)
AABBCC
(very dark)
F1 Generation
AaBbCc
AaBbCc
F2 Generation
Sperm
1/8
Eggs
1/8
1/64
6/64
15/64
20/64
15/64
6/64
1/64
Fraction of population
20/64
15/64
6/64
1/64
Skin pigmentation

(a) Eye of red-eyed *Drosophila* **(b)** Eye of white-eyed *Drosophila*

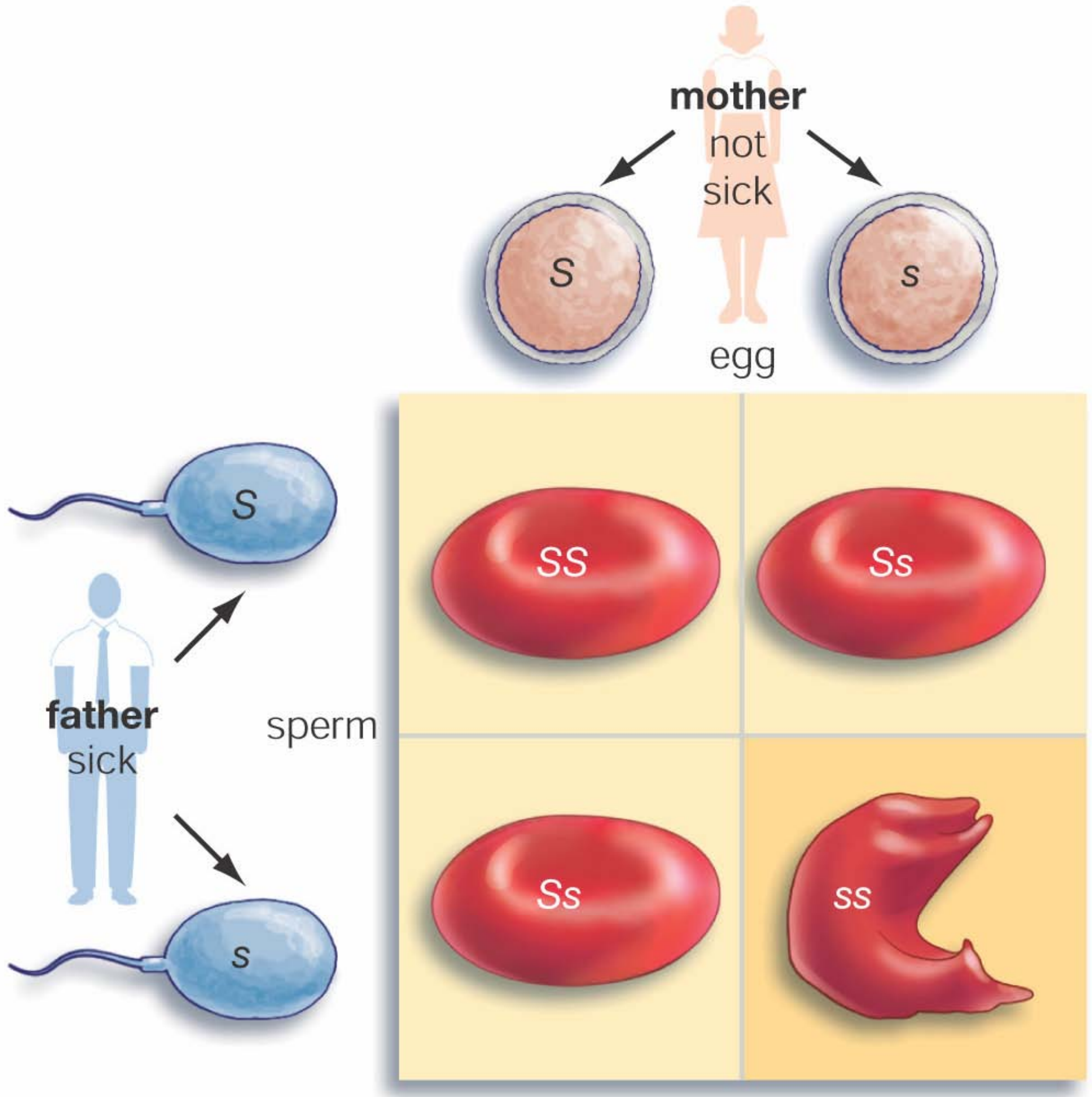



Parents
male
YX
female
XX
white
eyes
red
eyes
F1 generation
eggs
sperm
F2 generation
males
100%
eggs
females
100%
sperm
females
100%
red
males
50%
white
males
50% red

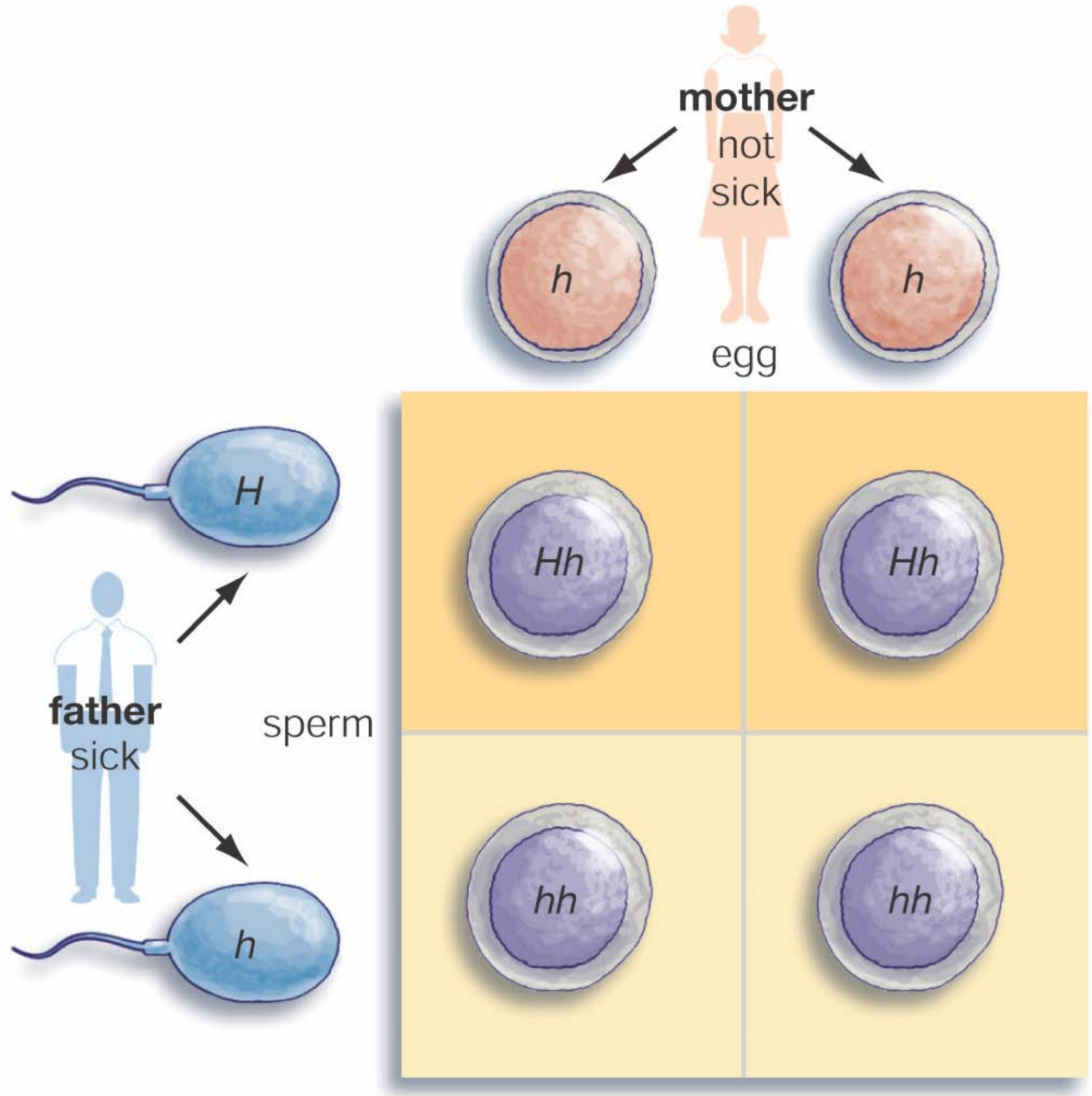

mother not color blind
functional red-green alleles
X
X
nonfunctional red-green alleles
egg
X
Y
father not color blind
sperm
XX
XX
XY
XY
daughters are not color blind
one son is color blind

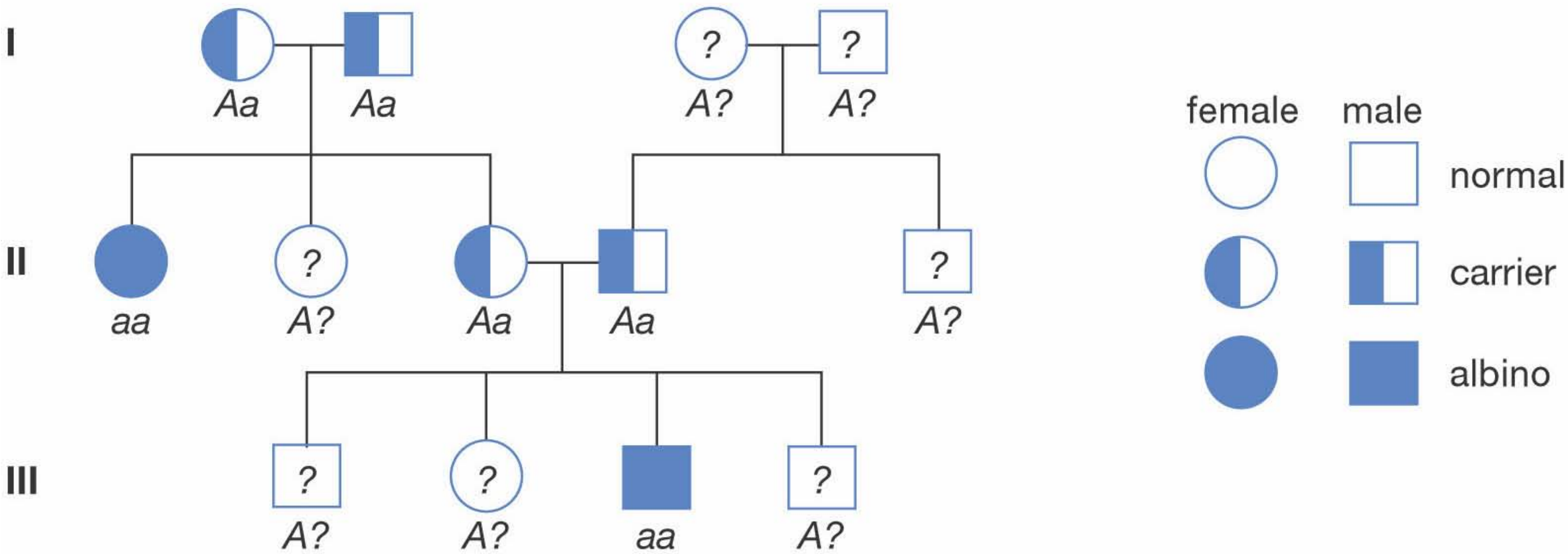


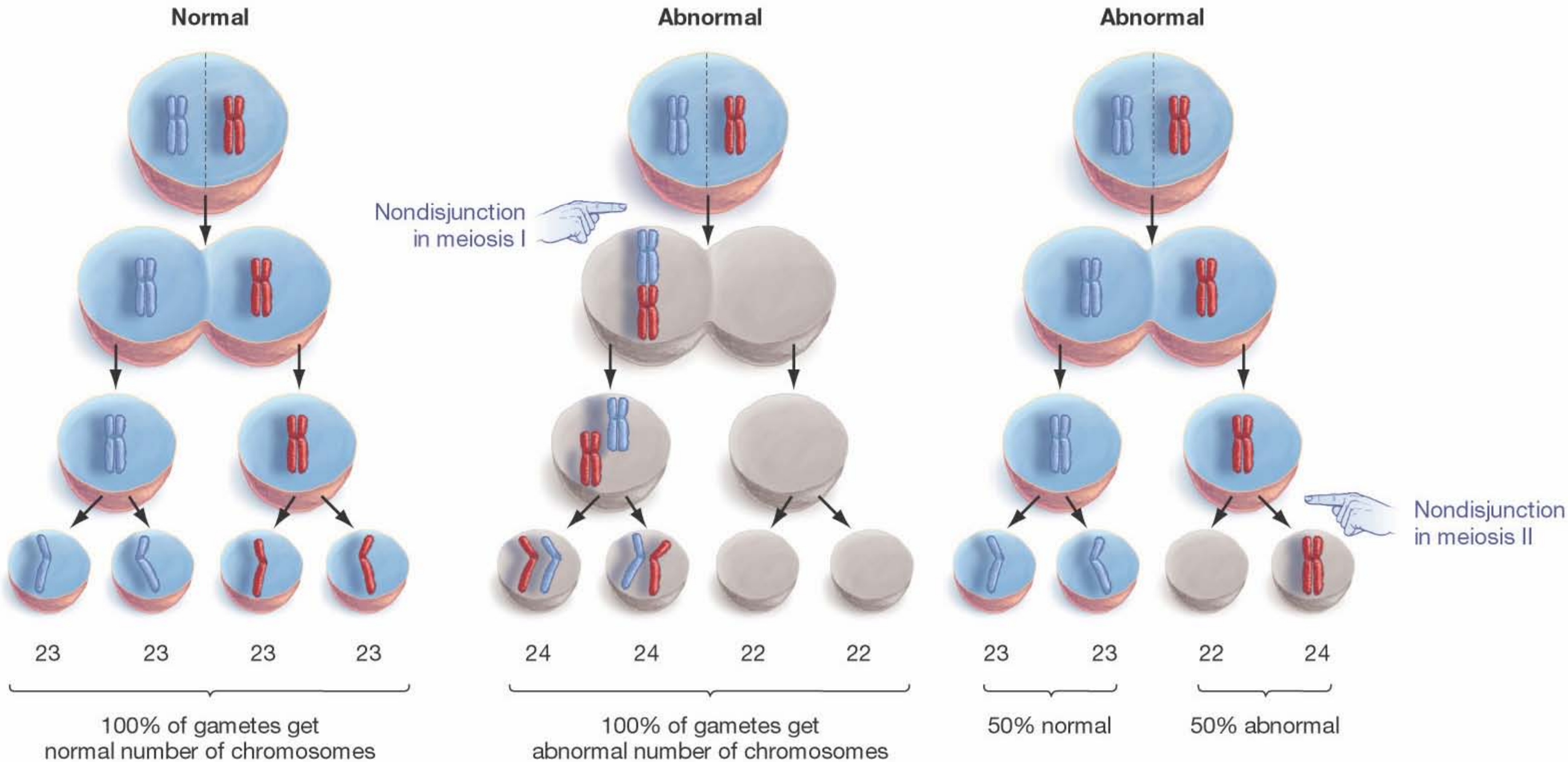
(a) Sickle-cell anemia: transmission of a recessive disorder.



(b) Huntington disease: transmission of a dominant disorder.

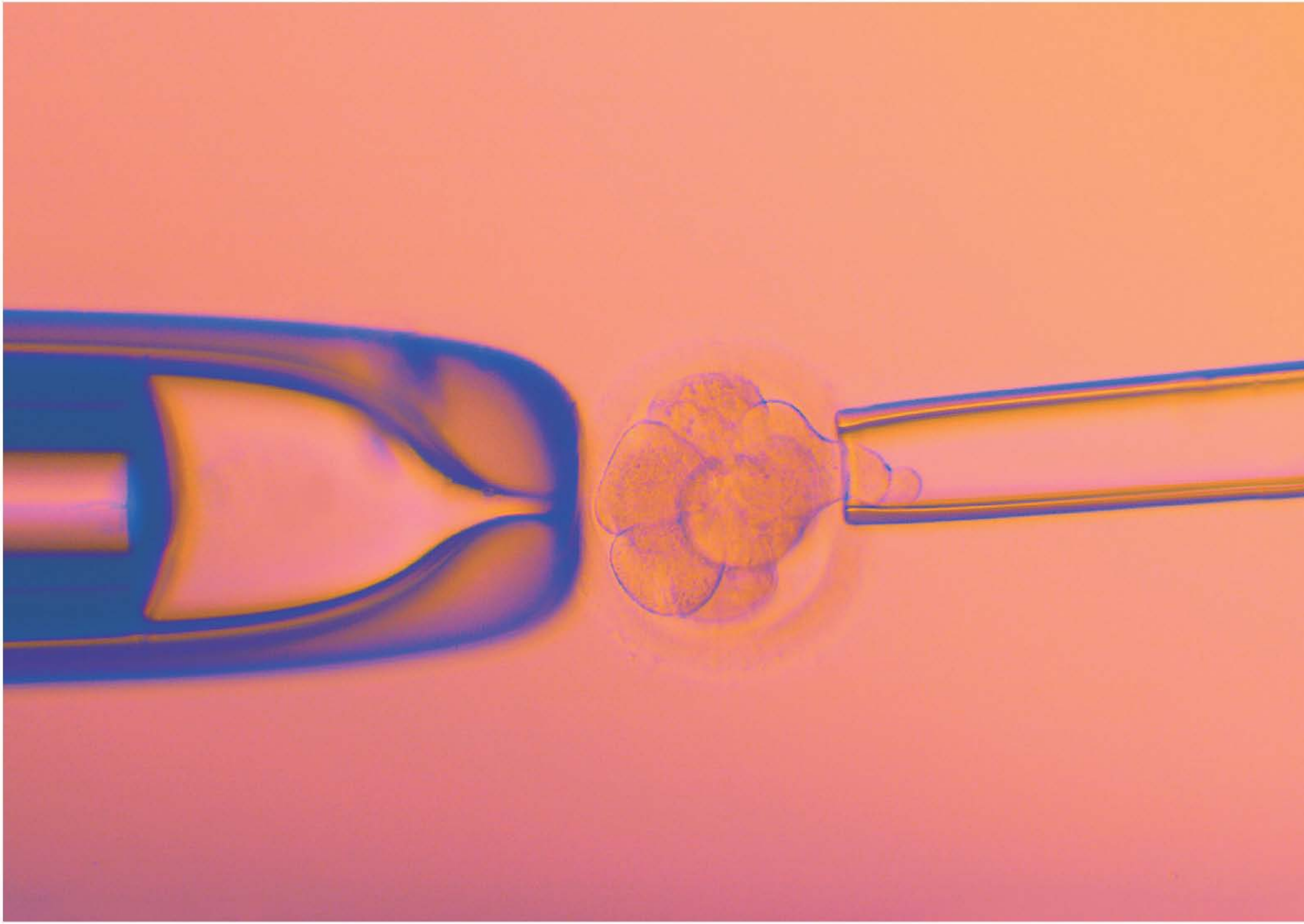


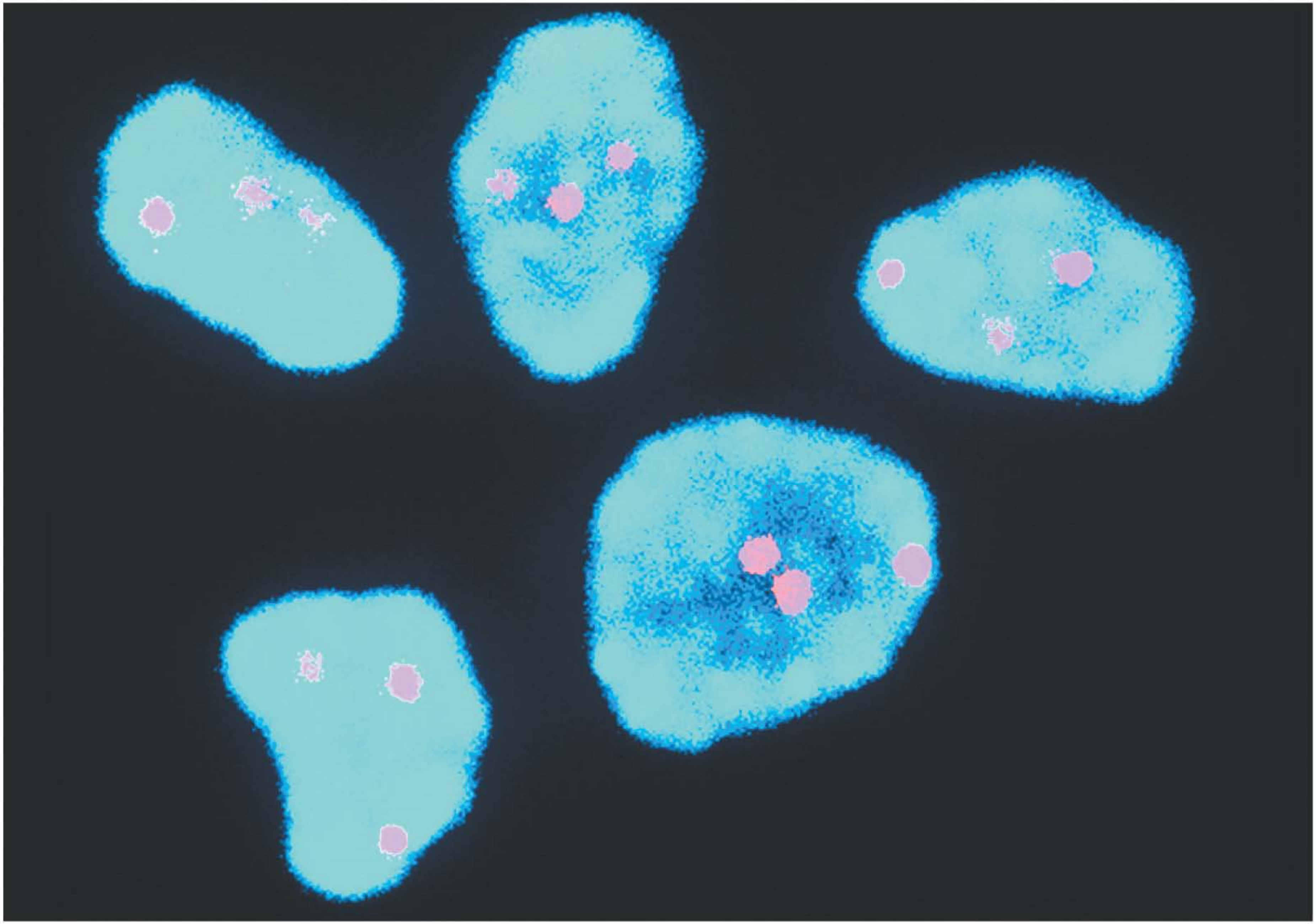

I
II
III
?
Aa
Aa
A?
A?
aa
A?
Aa
Aa
A?
A?
A?
aa
A?
female
male
normal
carrier
albino


Normal
Abnormal
Abnormal
Nondisjunction
in meiosis I
Nondisjunction
in meiosis II
23
23
23
23
24
24
22
22
23
23
22
24
100% of gametes get
normal number of chromosomes
100% of gametes get
abnormal number of chromosomes
50% normal
50% abnormal

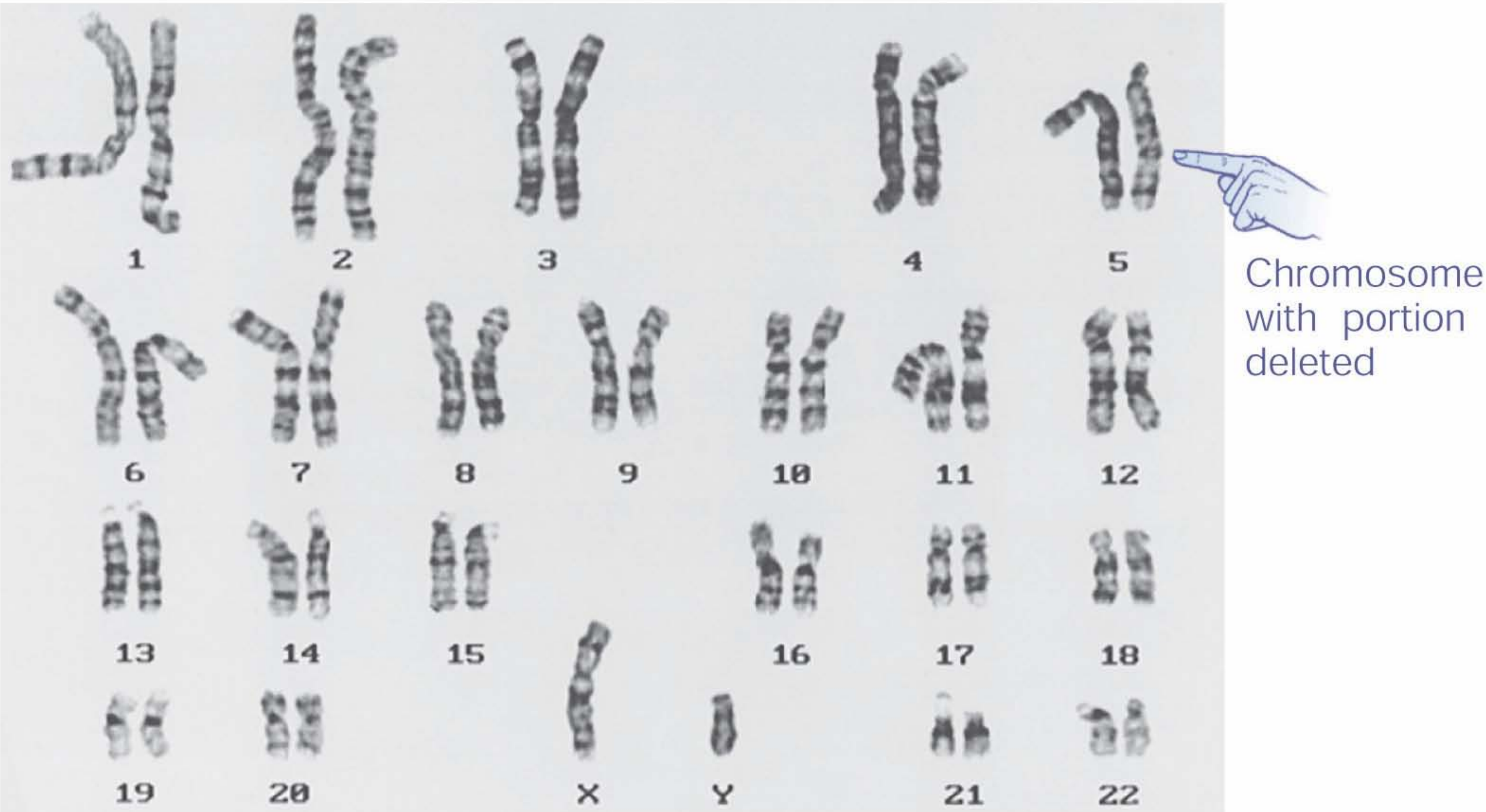
(b) Maternal age and Down syndrome risk

Mother's age	Chances of giving birth to a child with Down syndrome
20	1 in 1925
25	1 in 1205
30	1 in 885
35	1 in 365
40	1 in 110
45	1 in 32





(b) Cri-du-chat karyotype



(a) Cri-du-chat syndrome

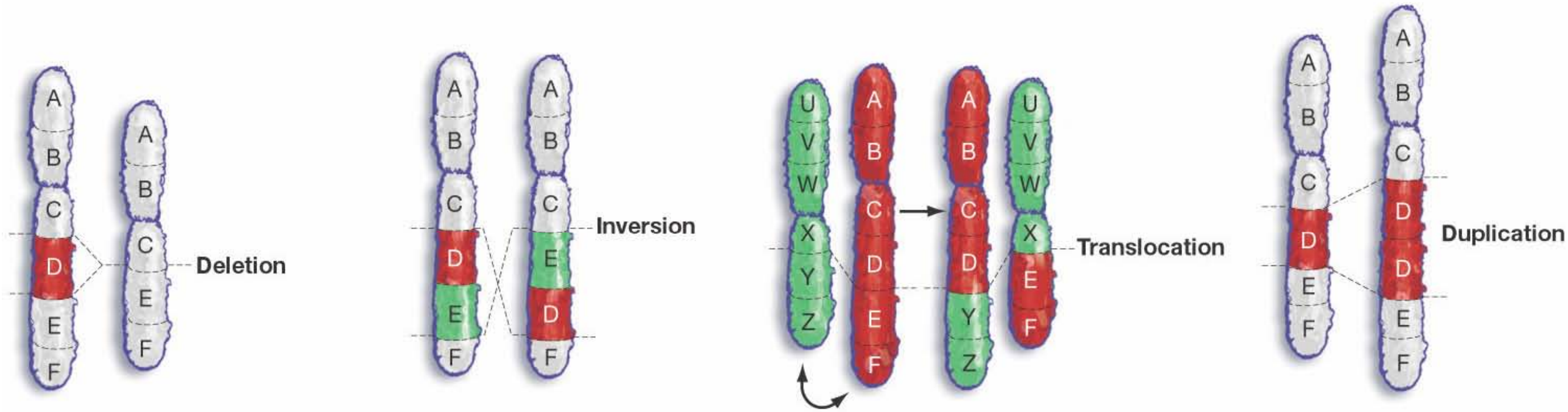

A
B
C
D
E
F
A
B
C
E
F
Deletion
A
B
C
D
E
F
A
B
C
E
D
F
Inversion
U
V
W
X
Y
Z
A
B
C
D
E
F
A
B
C
D
Y
Z
U
V
W
X
E
F
Translocation
A
B
C
D
E
F
A
B
C
D
D
E
F
Duplication

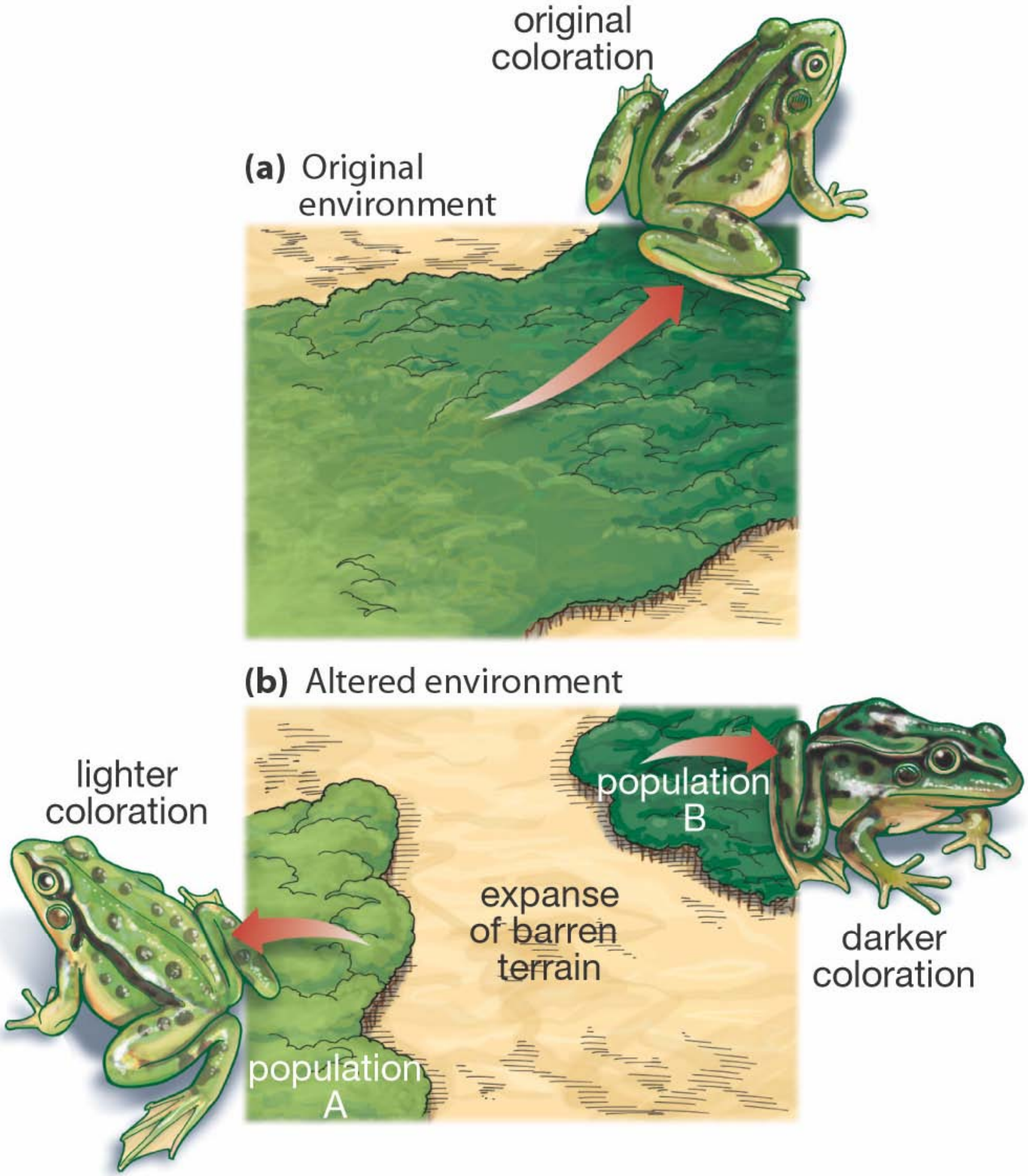
Table 12.1 Selected Examples of Human Genetic Disorders

Type	Name of condition	Effects
X-linked recessive disorders	Hemophilia	Faulty blood clotting
	Duchenne muscular dystrophy	Wasting of muscles
	Red-green color blindness	Inability to distinguish shades of red from green
Autosomal recessive disorders	Albinism	No pigmentation in skin
	Sickle-cell anemia	Decreased oxygen to brain and muscles
	Cystic fibrosis	Impaired lung function, lung infections
	Phenylketonuria	Mental retardation
	Tay-Sachs disease	Nervous system degeneration in infants
	Werner syndrome	Premature aging
Autosomal dominant disorders	Polydactyly	Extra fingers or toes
	Campodactyly	Inability to straighten little finger
	Huntington disease	Brain tissue degeneration
Aberrations in chromosome number	Down syndrome	Mental retardation, shortened life span
	Turner syndrome	Sterility, short stature
	Klinefelter syndrome	Dysfunctional testicles, feminized features
Aberrations in chromosome structure	Cri-du-chat syndrome	Mental retardation, malformed larynx
	Fragile-X syndrome	Mental retardation, facial deformities

Population Genetics

concepts

- Population evolve
- Gene pool
- Microevolution: change in allele frequency


original
coloration
(a) Original
environment
(b) Altered environment
lighter
coloration
population
B
expanse
of barren
terrain
darker
coloration
population
A

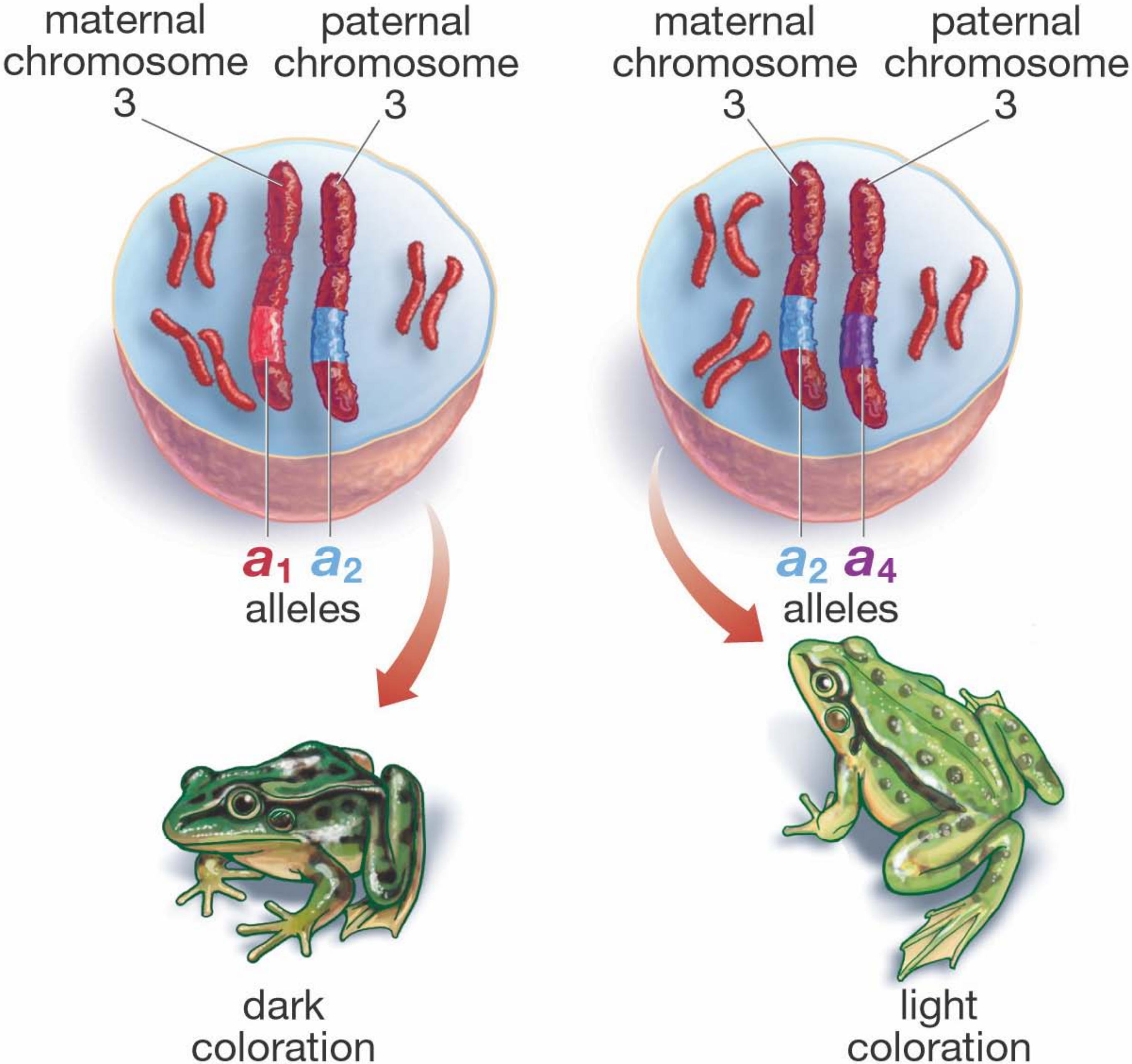
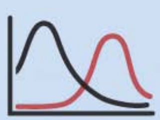
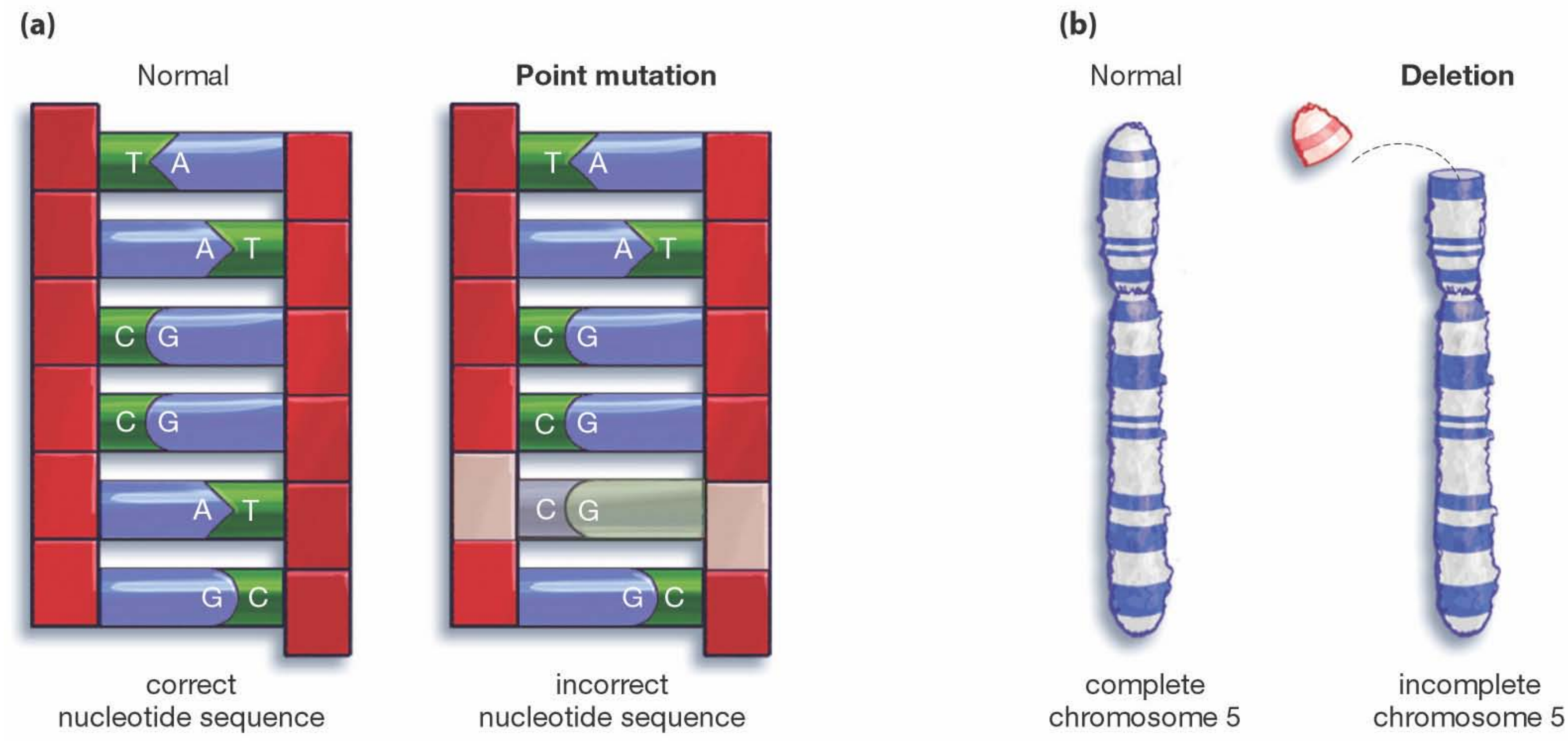

maternal chromosome 3
paternal chromosome 3
maternal chromosome 3
paternal chromosome 3
a_1 a_2
alleles
a_2 a_4
alleles
dark coloration
light coloration

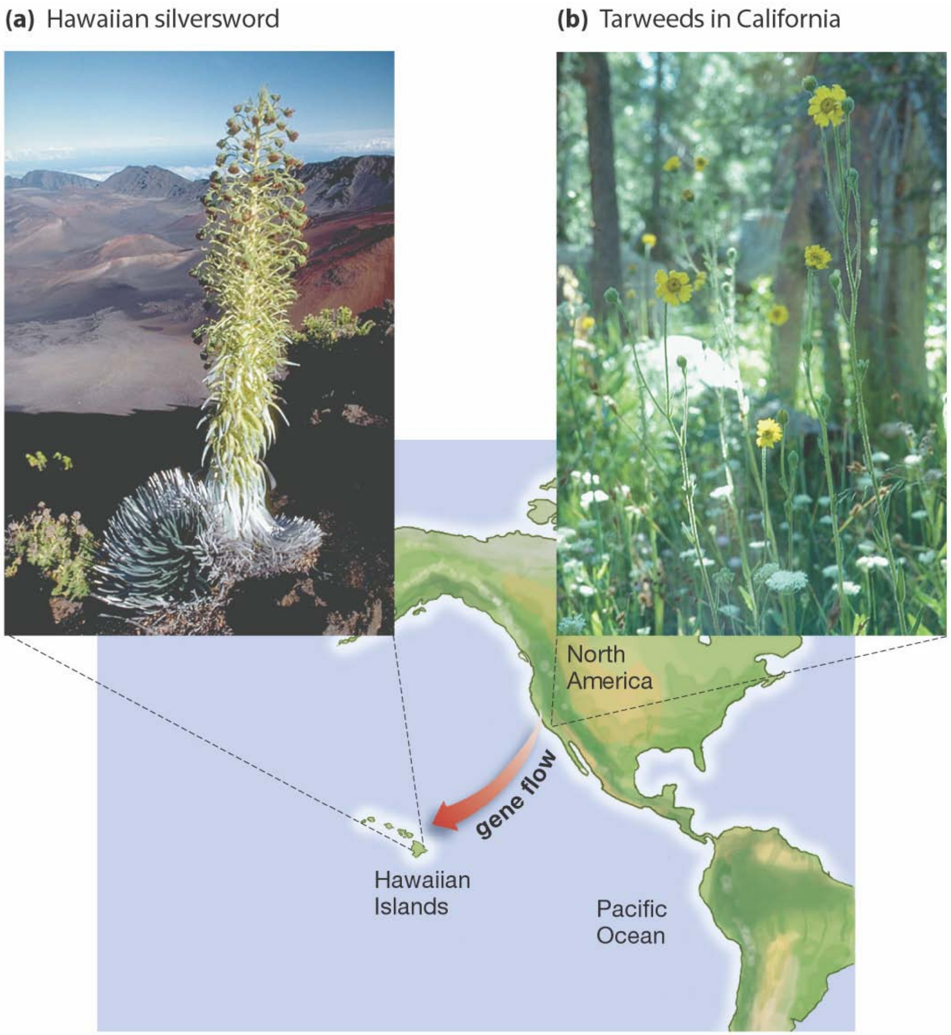
Table 17.1 Agents of Change: Five Forces That Can Bring about Change in Allele Frequencies in a Population

Agent	Description
Mutation 	Alteration in an organism's DNA; generally has no effect or a harmful effect. But beneficial or "adaptive" mutations are indispensable to evolution.
Gene flow 	The movement of alleles from one population to another. Occurs when individuals move between populations or when one population of a species joins another, assuming the second population has different allele frequencies than the first.
Genetic drift 	Chance alteration of gene frequencies in a population. Most strongly affects small populations. Can occur when populations are reduced to small numbers (the bottleneck effect) or when a few individuals from a population migrate to a new, isolated location and start a new population (the founder effect).
Nonrandom mating 	Occurs when one member of a population is not equally likely to mate with any other member. Includes sexual selection, in which members of a population choose mates based on the traits the mates exhibit.
Natural selection 	Some individuals will be more successful than others in surviving and hence reproducing, owing to traits that give them a better "fit" with their environment. The alleles of those who reproduce more will increase in frequency in a population.

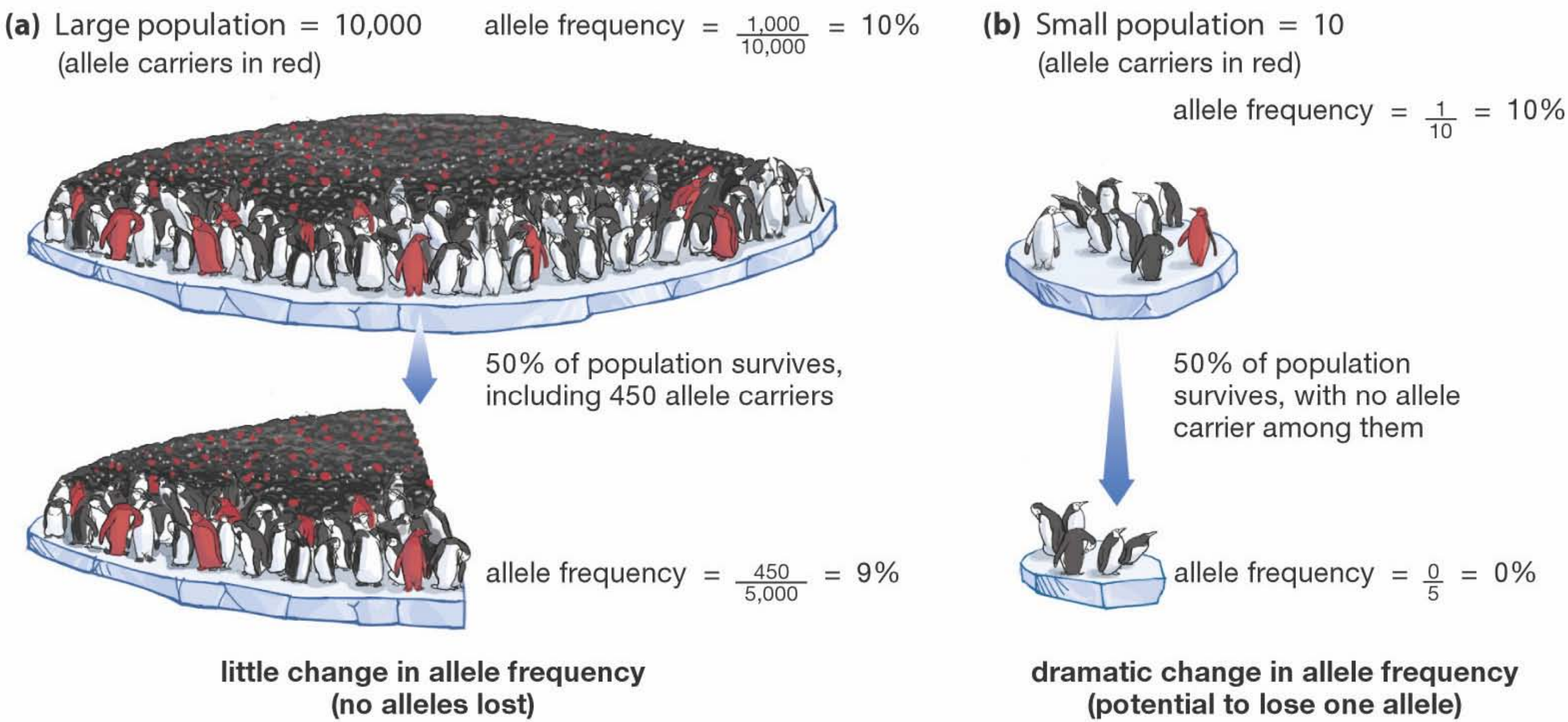
Genetic Mutation



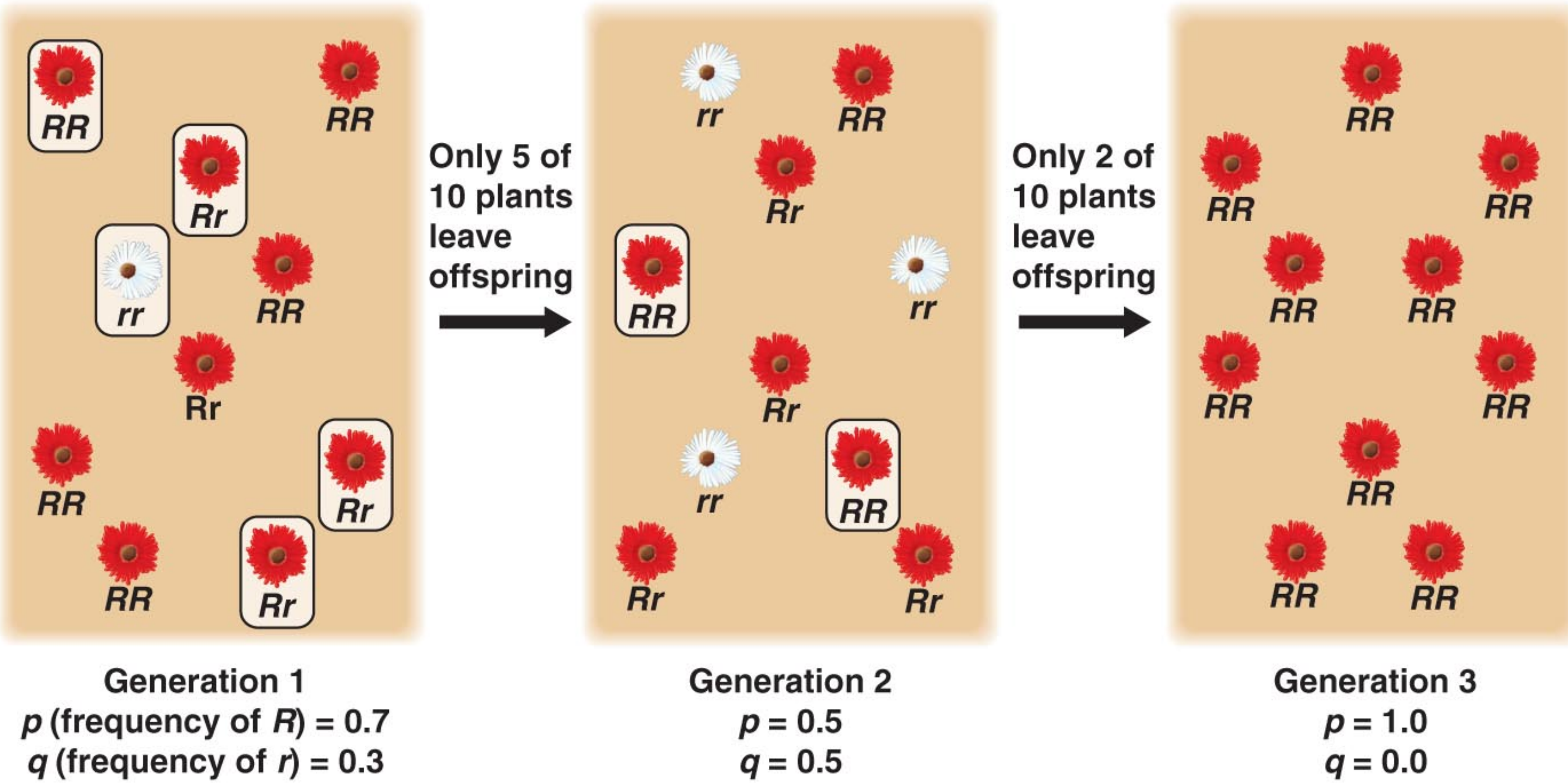
Gene flow


(a) Hawaiian silversword
(b) Tarweeds in California
North America
gene flow
Hawaiian Islands
Pacific Ocean

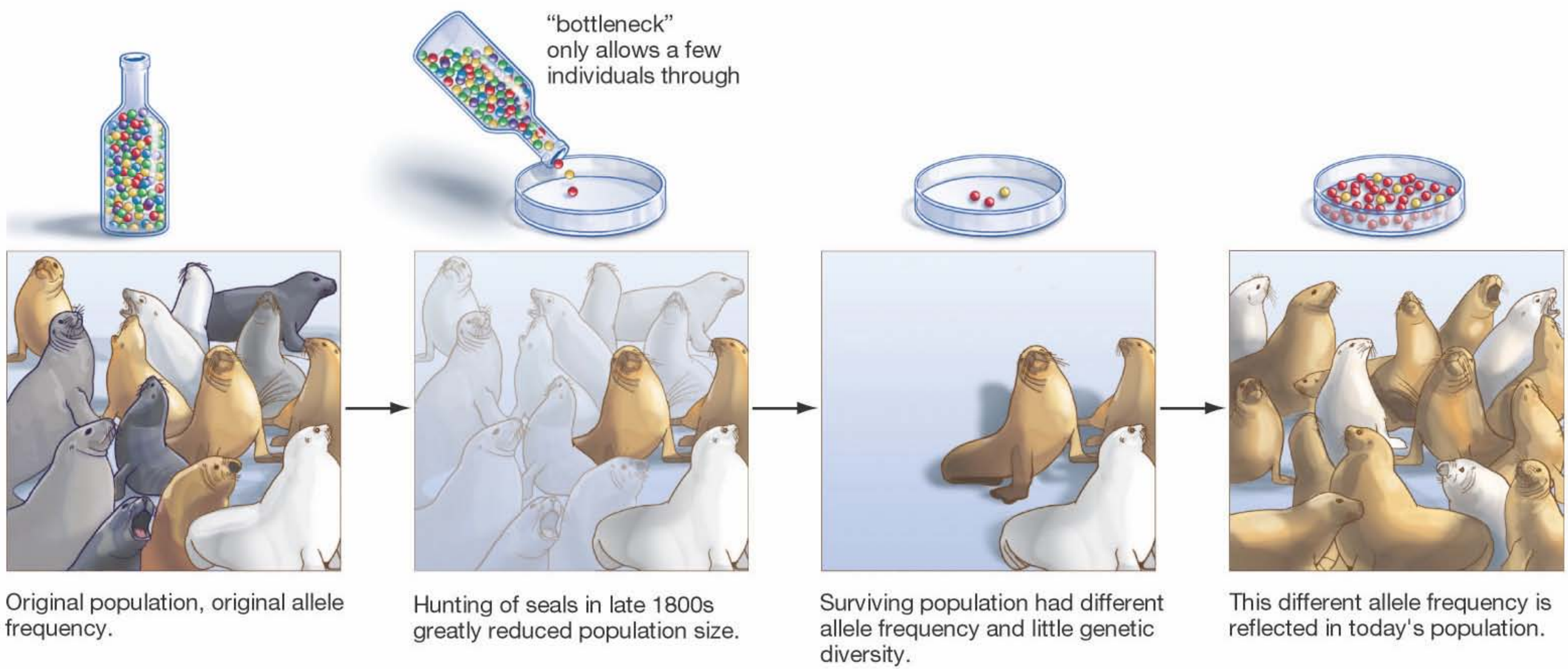
Genetic Drift



Genetic Drift



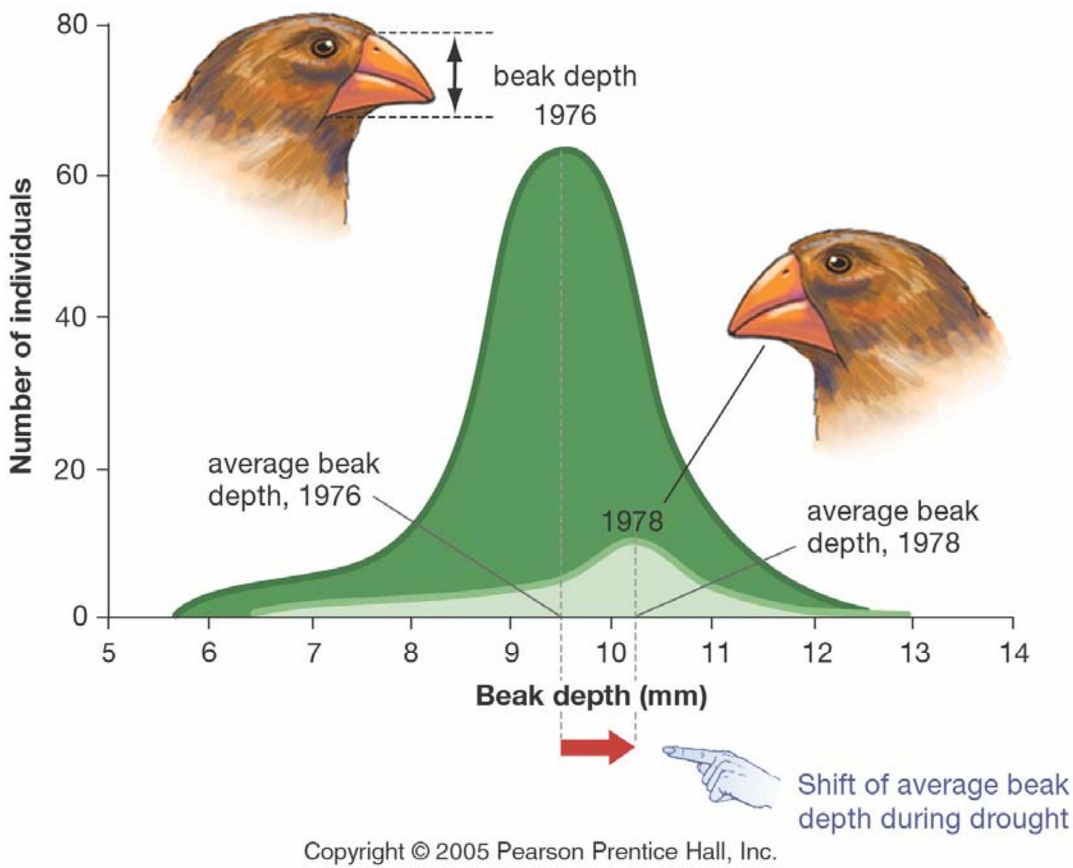
Bottle neck effect --gene flow

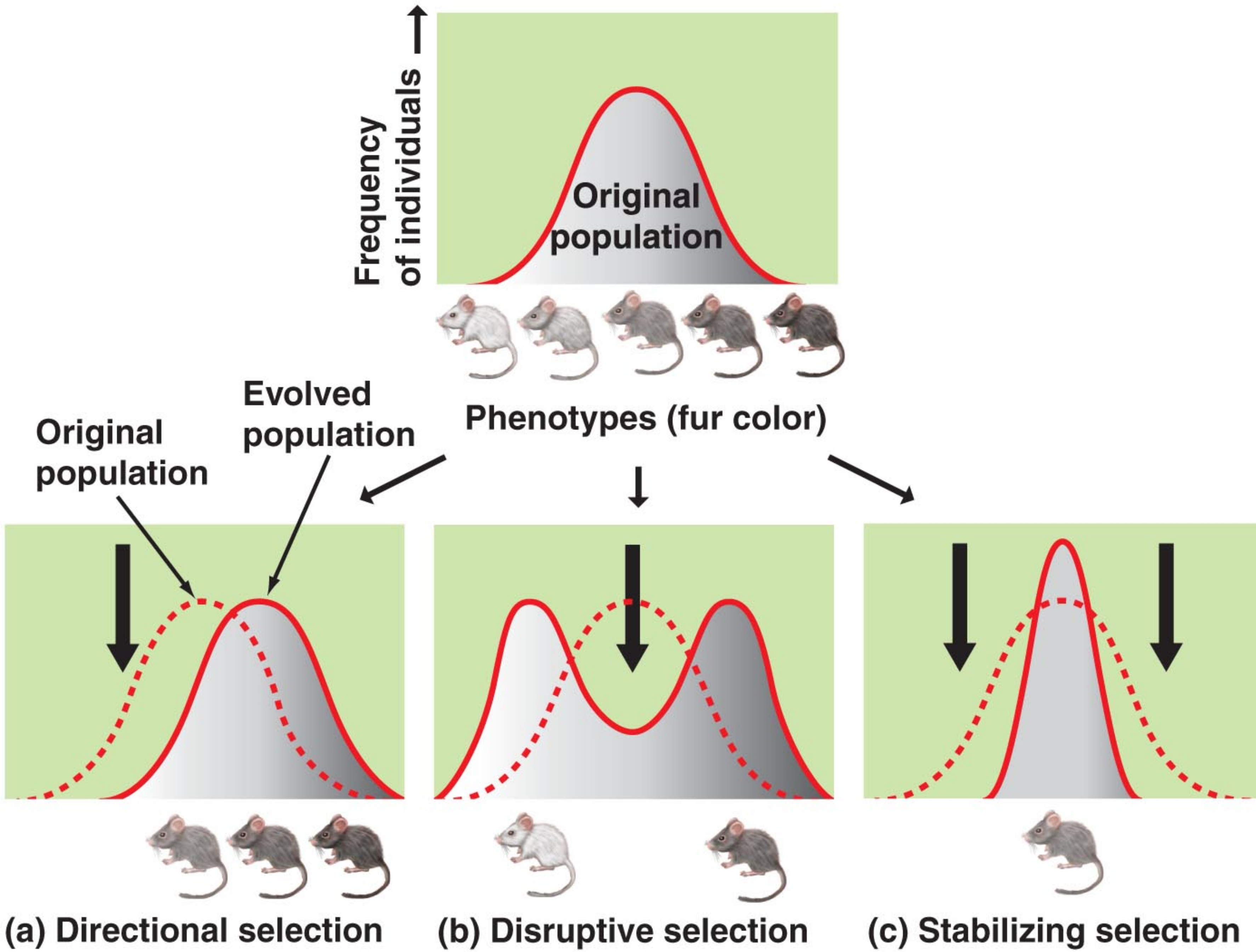


Non Random mating

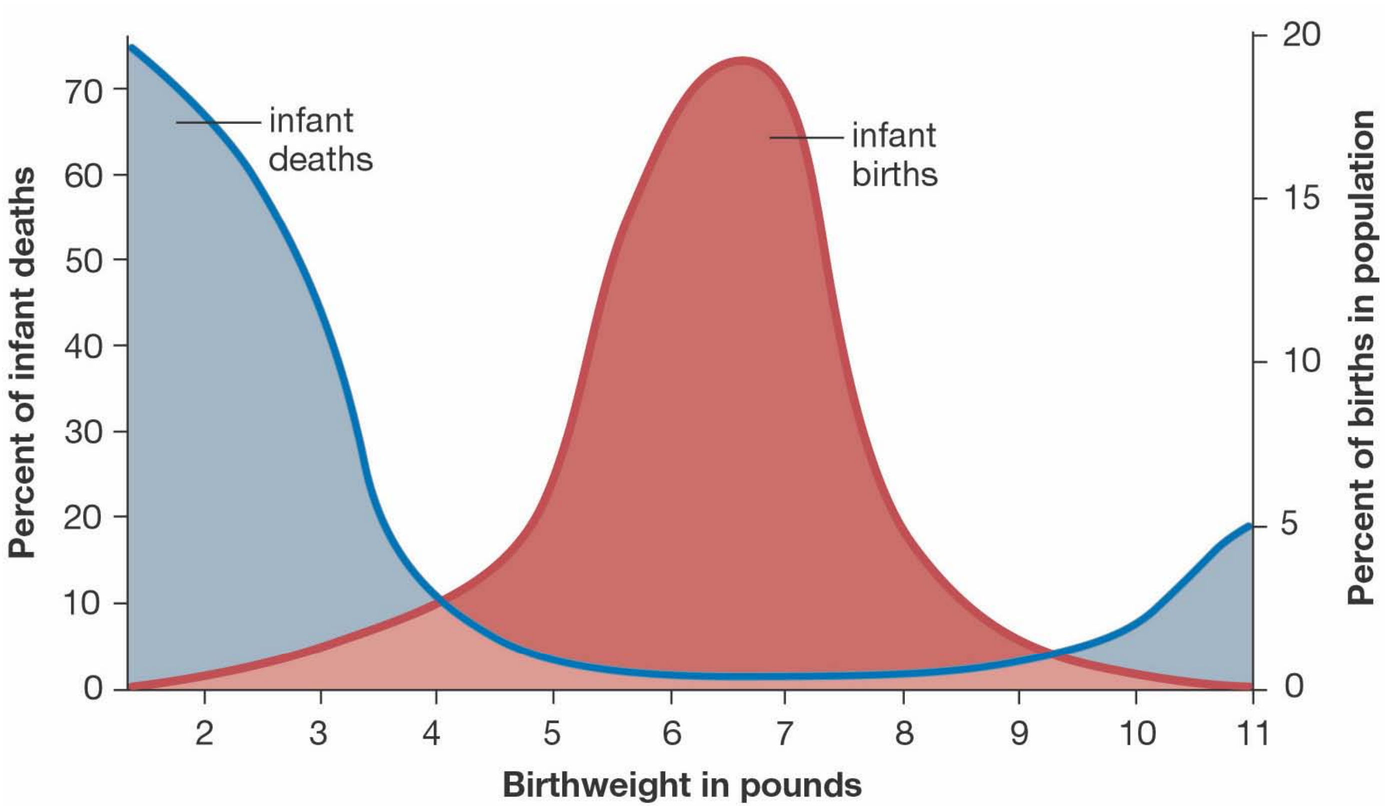
Natural Selection

Directional selection

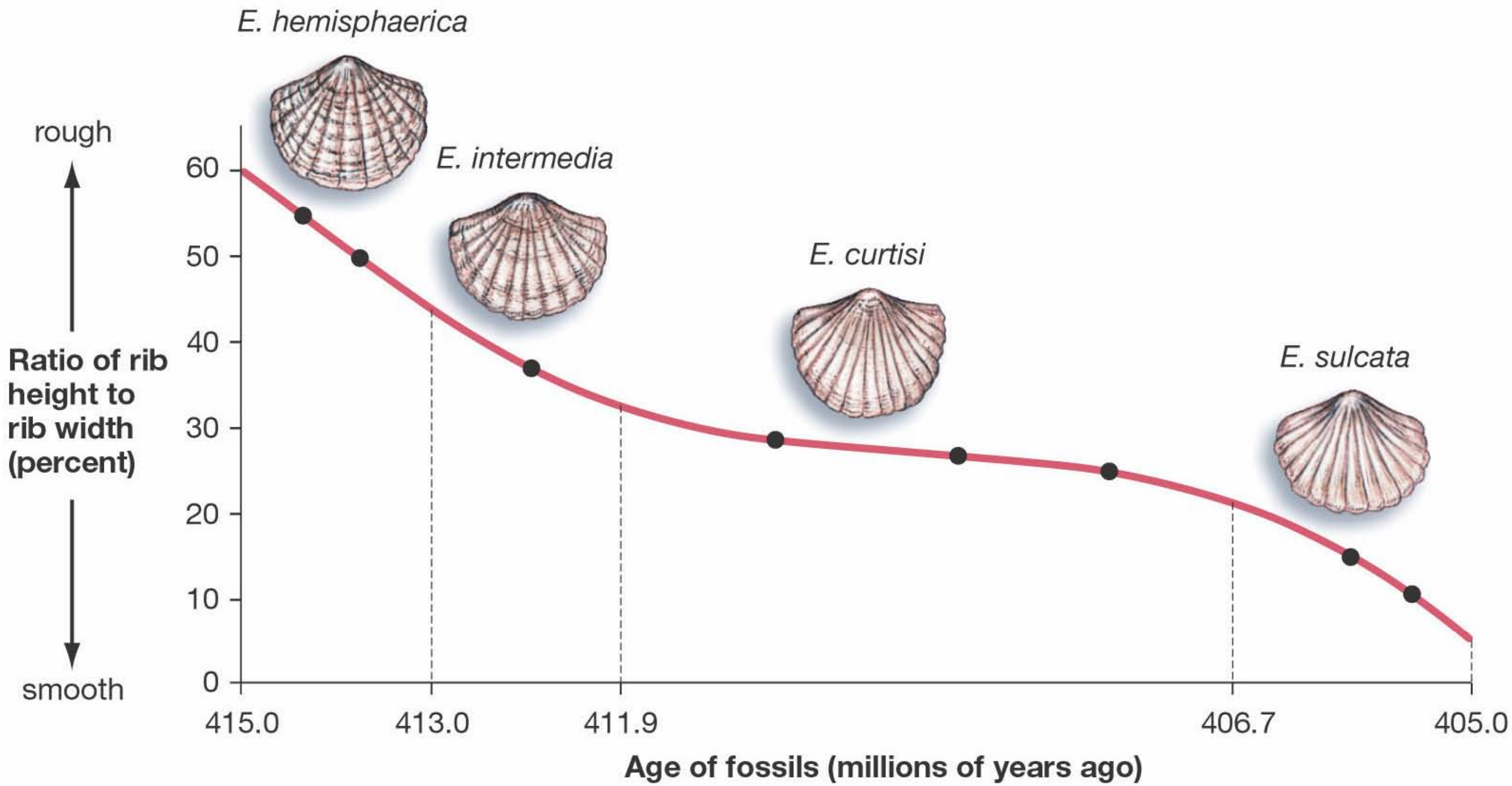



Frequency of individuals
Original population
Phenotypes (fur color)
Original population
Evolved population
(a) Directional selection
(b) Disruptive selection
(c) Stabilizing selection

Stabilizing Selection



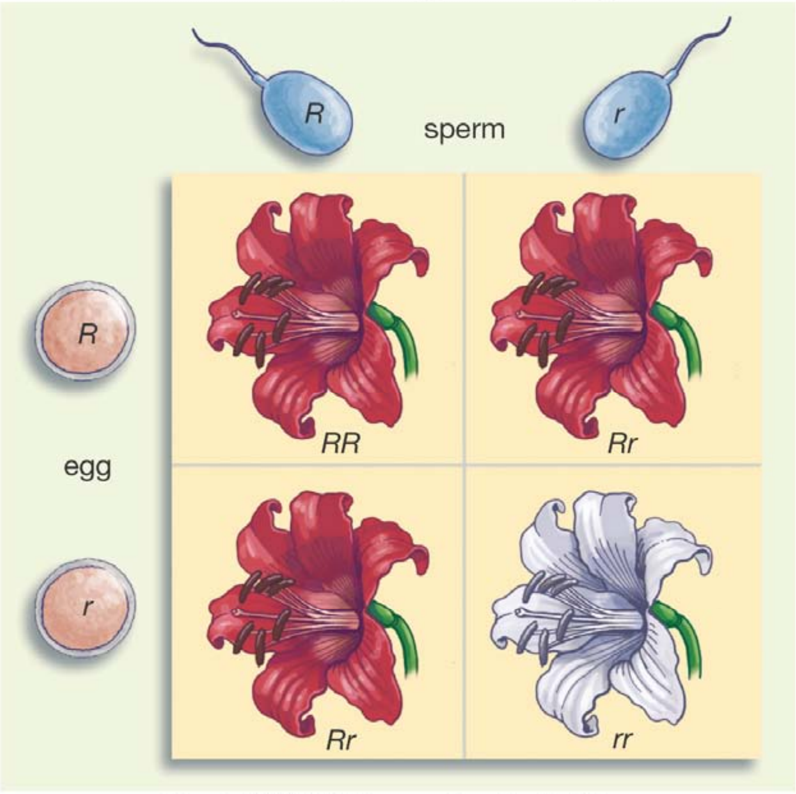
Directional selection



Disruptive Selection

Hardy-Weinberg

Possible genotypes in the population



Hardy-Weinberg equations