BIOL2107, Fall '23

Lecture 12

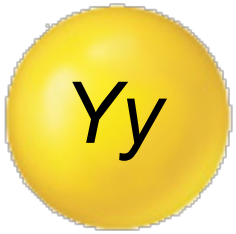
Mendel's 1st law- the law of segregation

Mendel's First Law: Two members of a gene pair segregate from each other into the gametes, whereby one half of the gametes carries one of the traits, the other half carries the other.

Mendel's 2nd law- the law of random/independent assortment

Mendel's Second Law: During gamete formation the segregation of one gene pair is independent of all other gene pairs

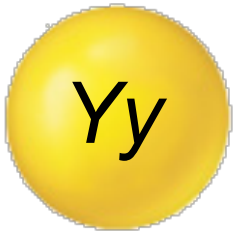
Homozygous DOMINANT



Heterozygous

Homozygous recessive

Homozygous DOMINANT



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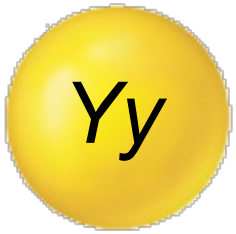
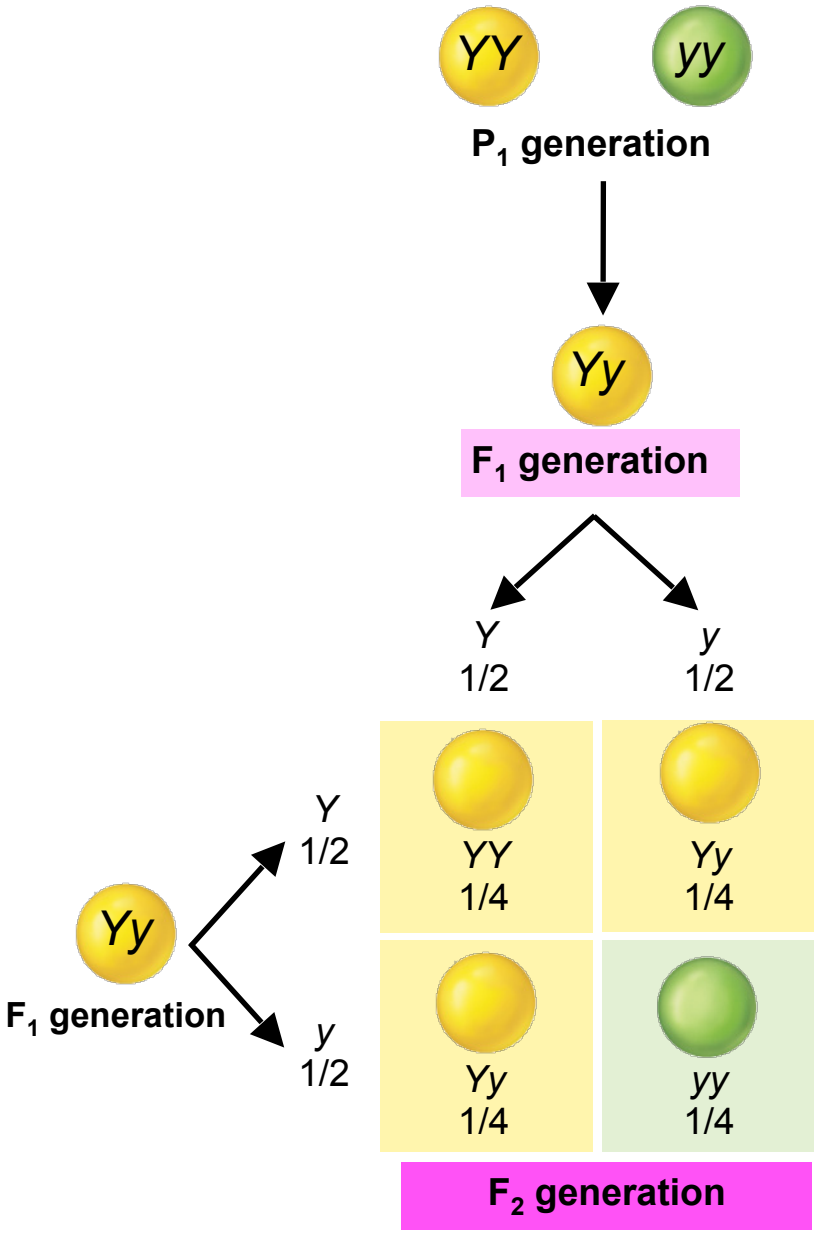


Heterozygous

Homozygous recessive

The Principle of Segregation



Expected ratio of *YY* : *Yy* : *yy* genotypes is 1 : 2 : 1

Expected ratio of dominant:recessive phenotypes is 3 : 1

Mendel's 1st law- the law of segregation

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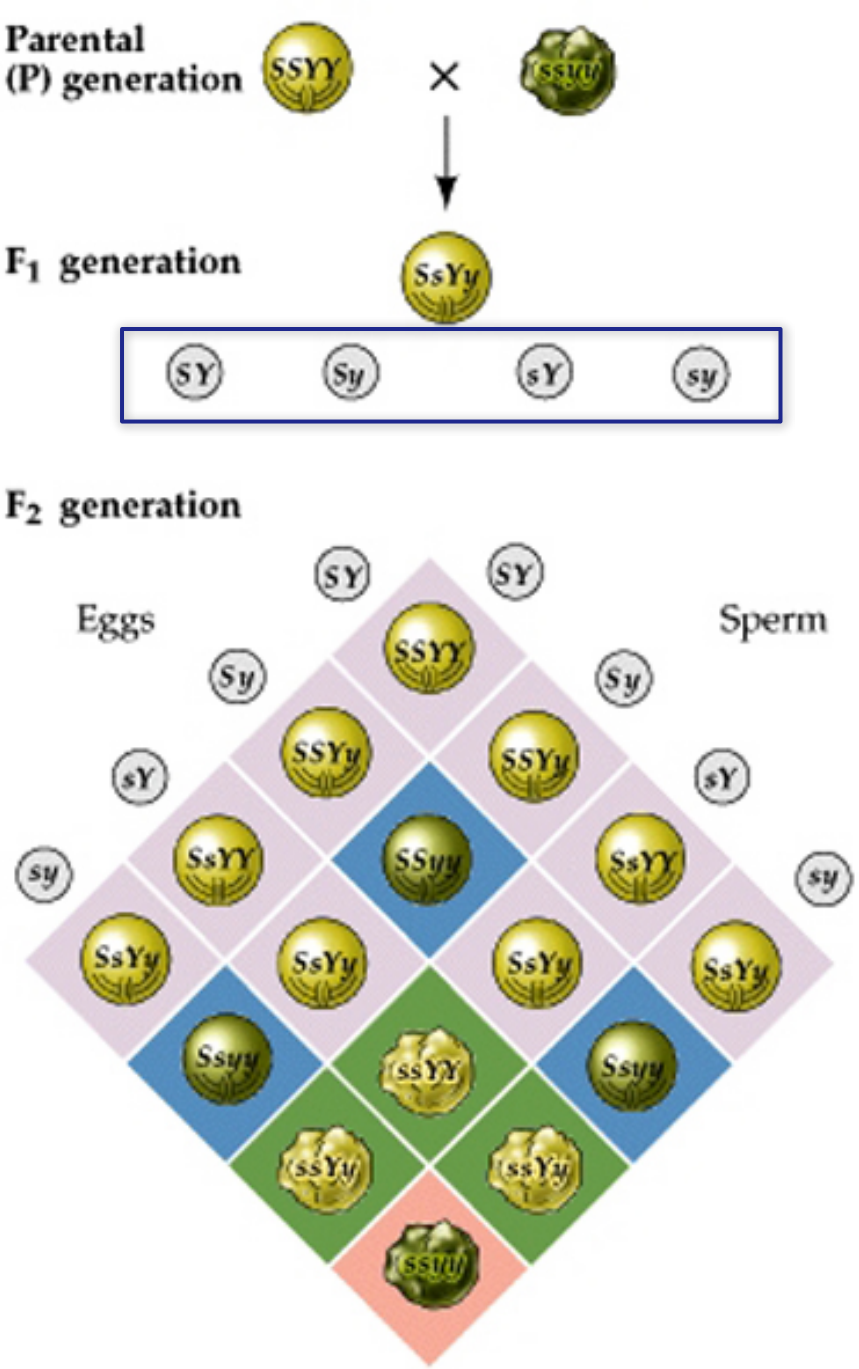
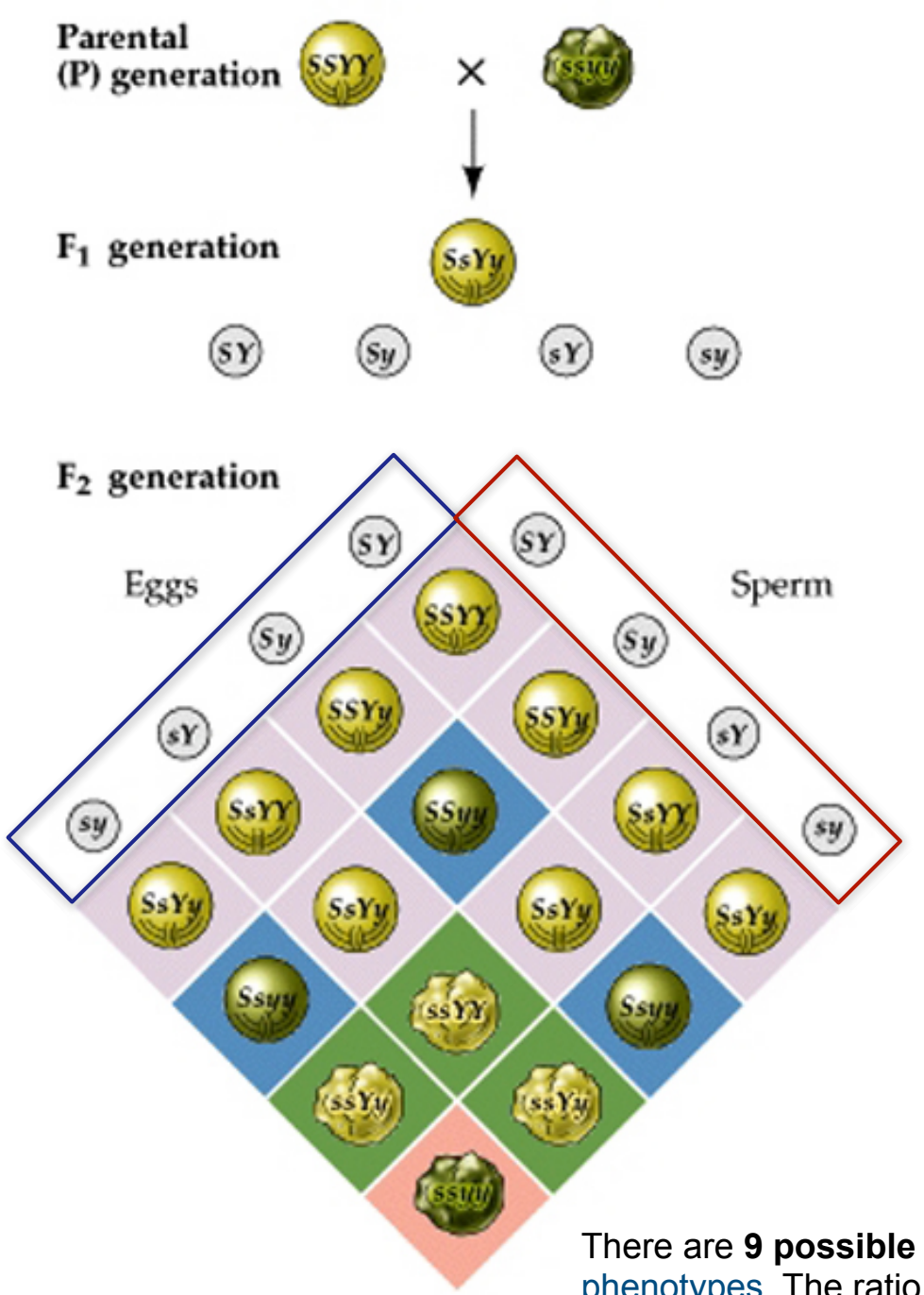
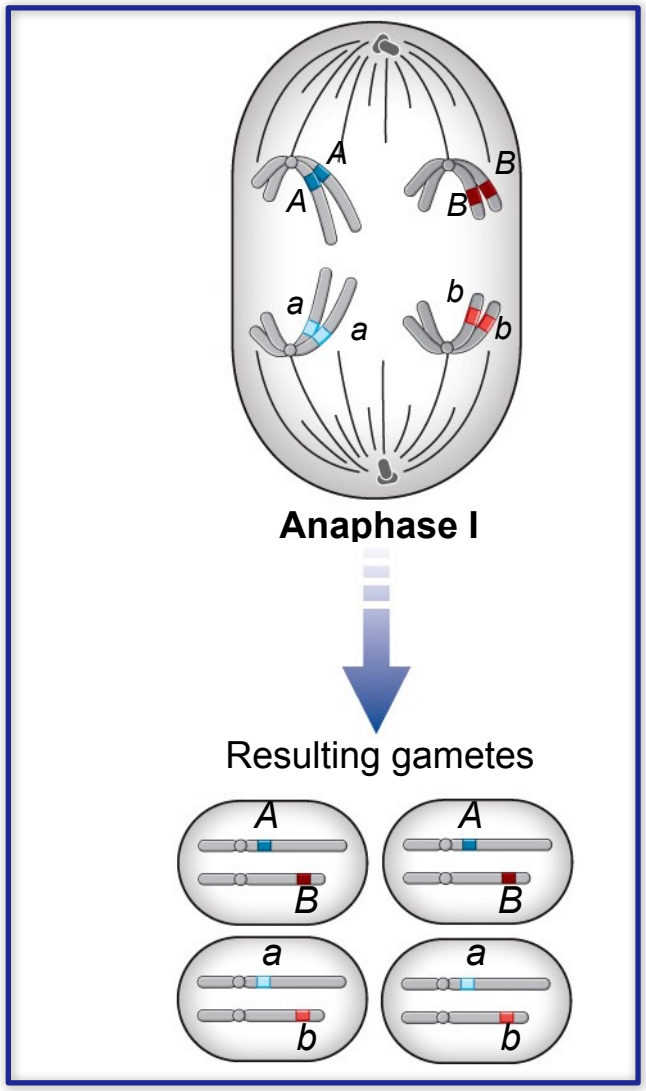

Parental
(P) generation
SSYY
×
ssyy
F1 generation
SsYy
SY
Sy
sY
sy
F2 generation
Eggs
Sperm
SY
SY
Sy
Sy
sY
sY
sy
sy
SSYY
SSYy
SSYy
SSyy
SsYY
SsYY
SsYy
SsYy
SsYy
SsYy
Ssyy
Ssyy
ssYY
ssYy
ssYy
ssyy

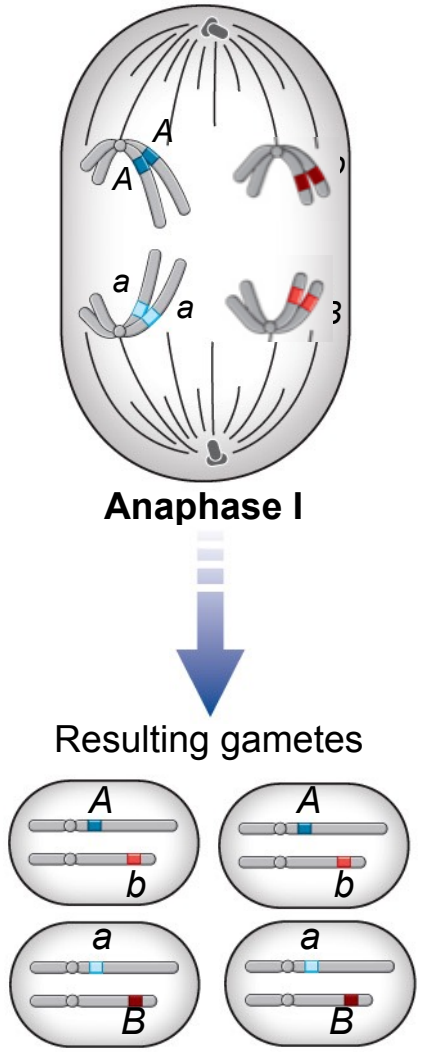


There are **9 possible genotypes** and 4 possible phenotypes. The ratio of phenotypes is **9:3:3:1**.

Independent Assortment

Independent assortment of genes in different chromosomes reflects the fact that non homologous chromosomes can orient in either of two ways that are equally likely.





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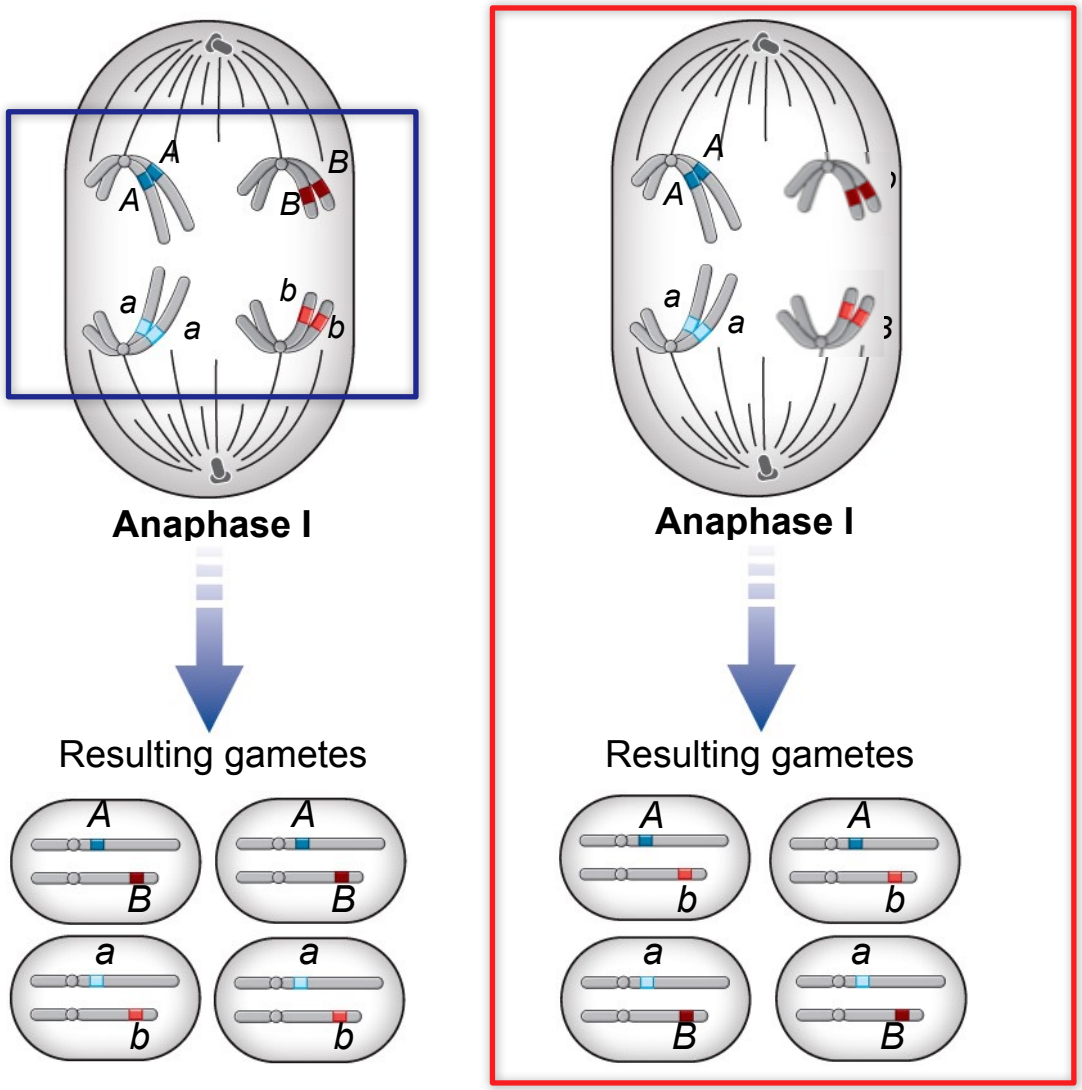

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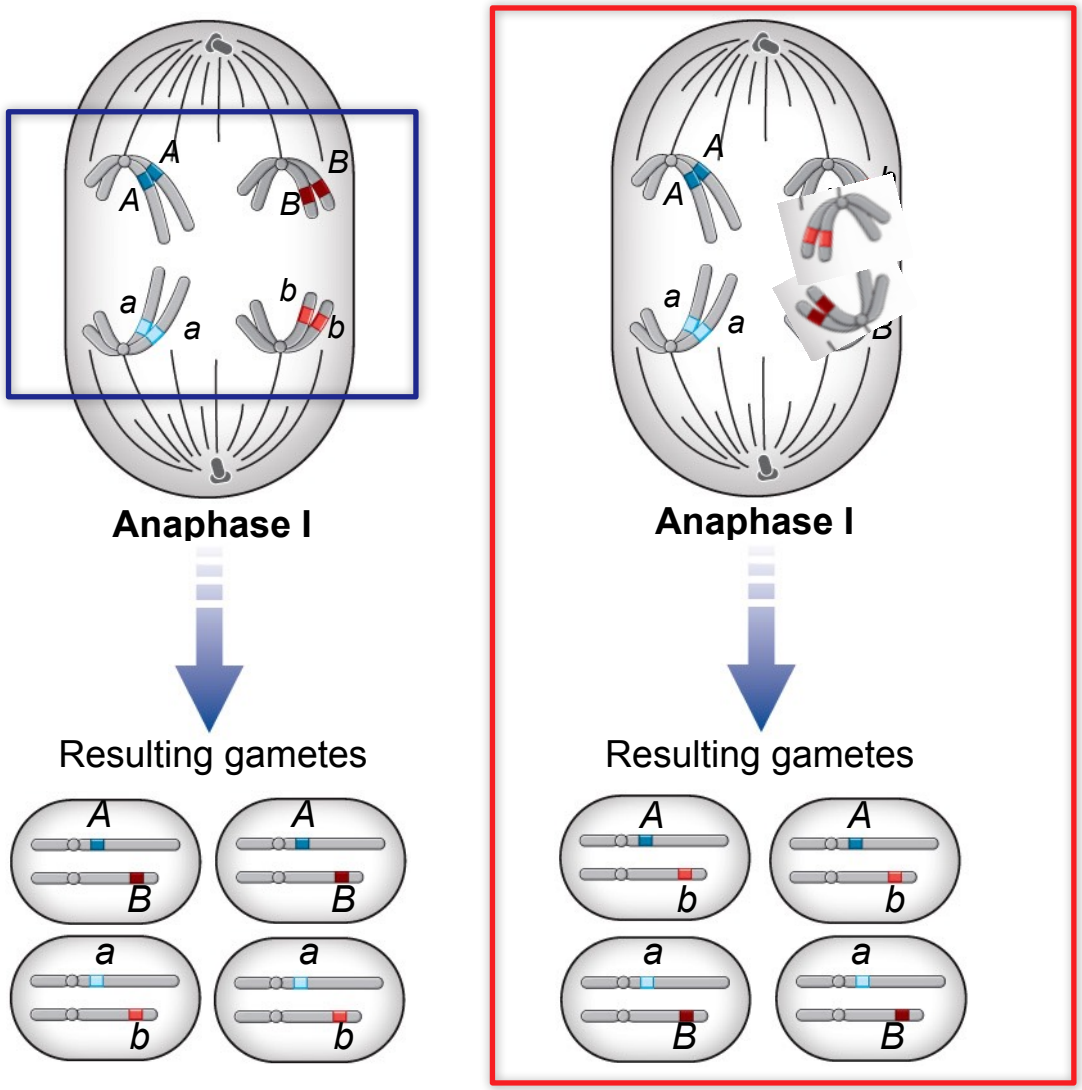

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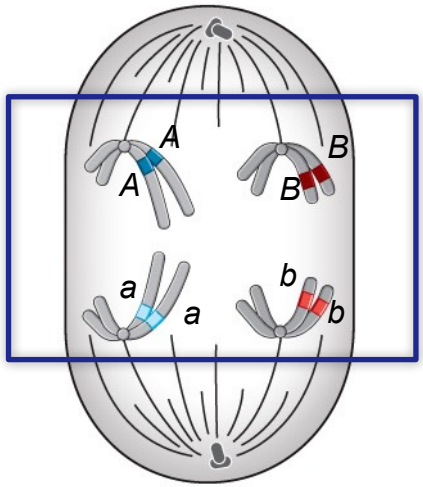
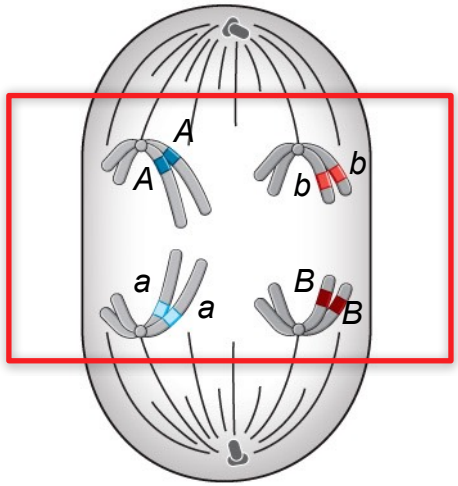
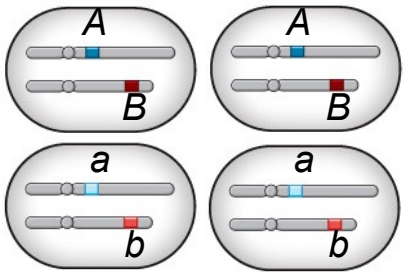


Anaphase I

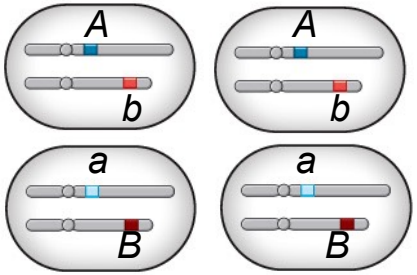


Anaphase I

Resulting gametes



Resulting gametes



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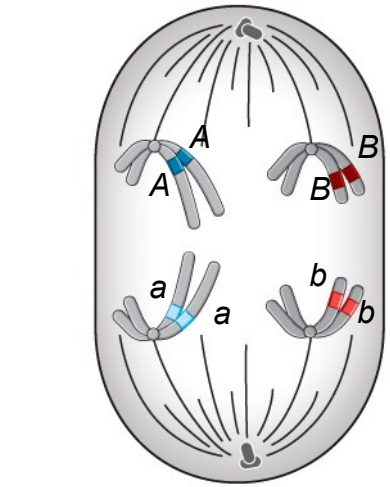
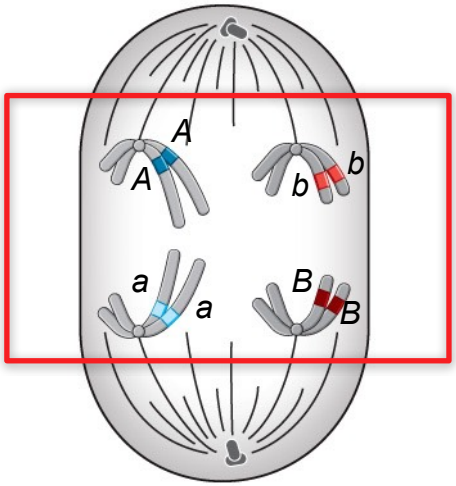


Anaphase I



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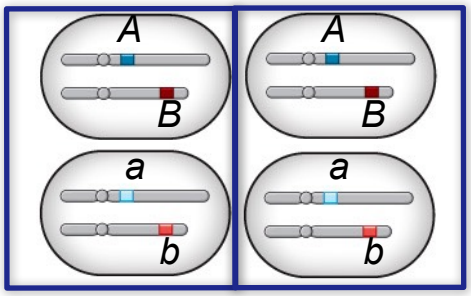
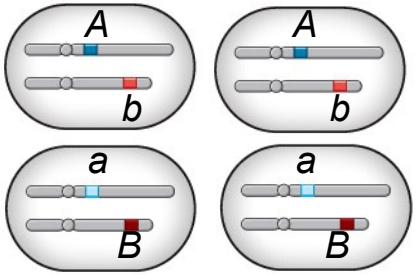


Resulting gametes



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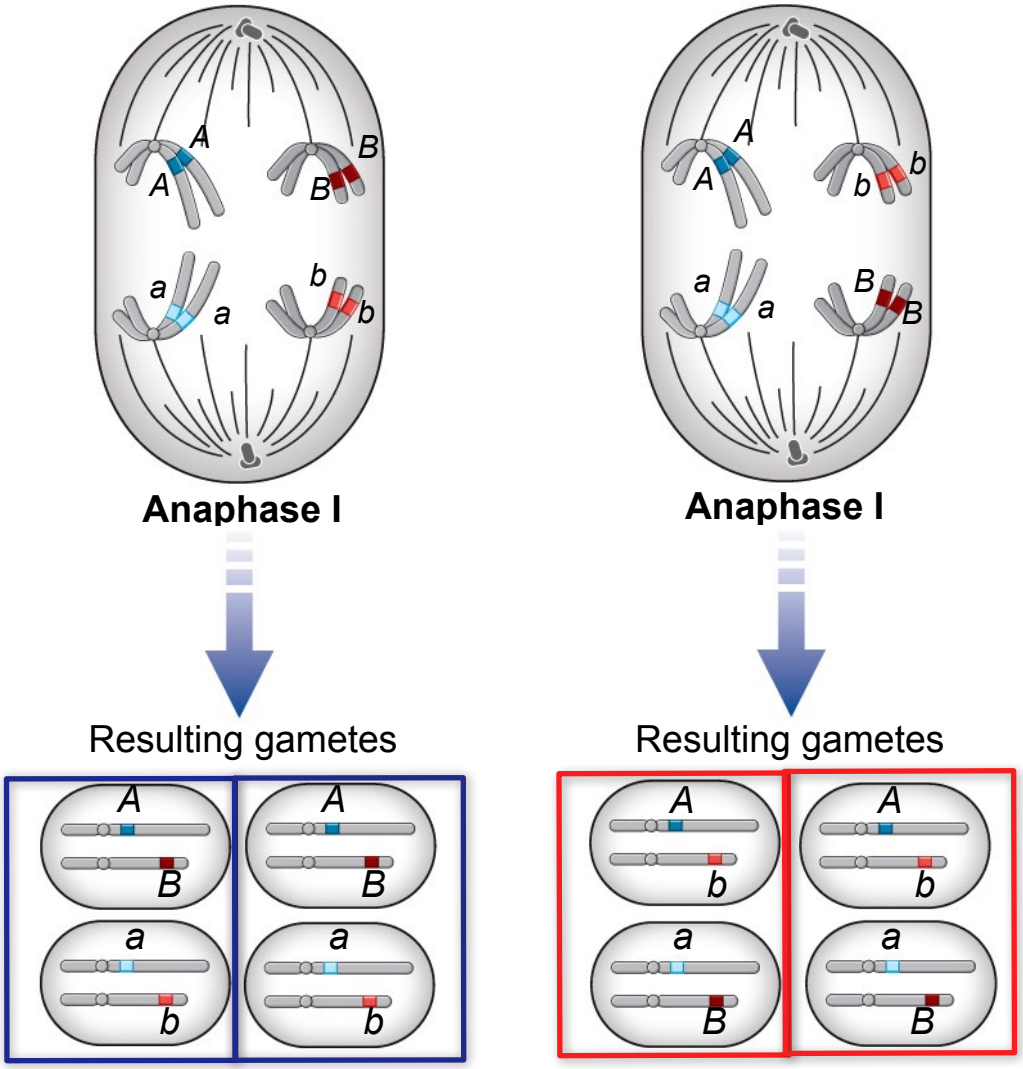
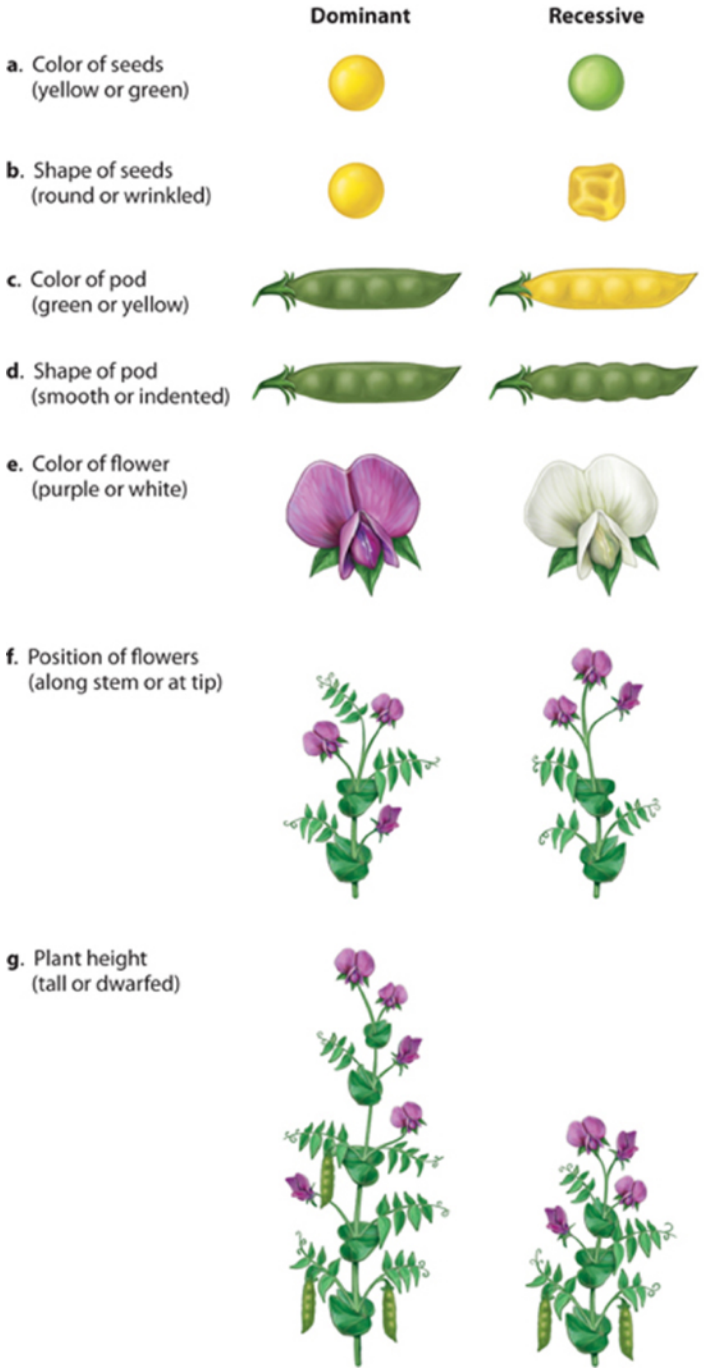
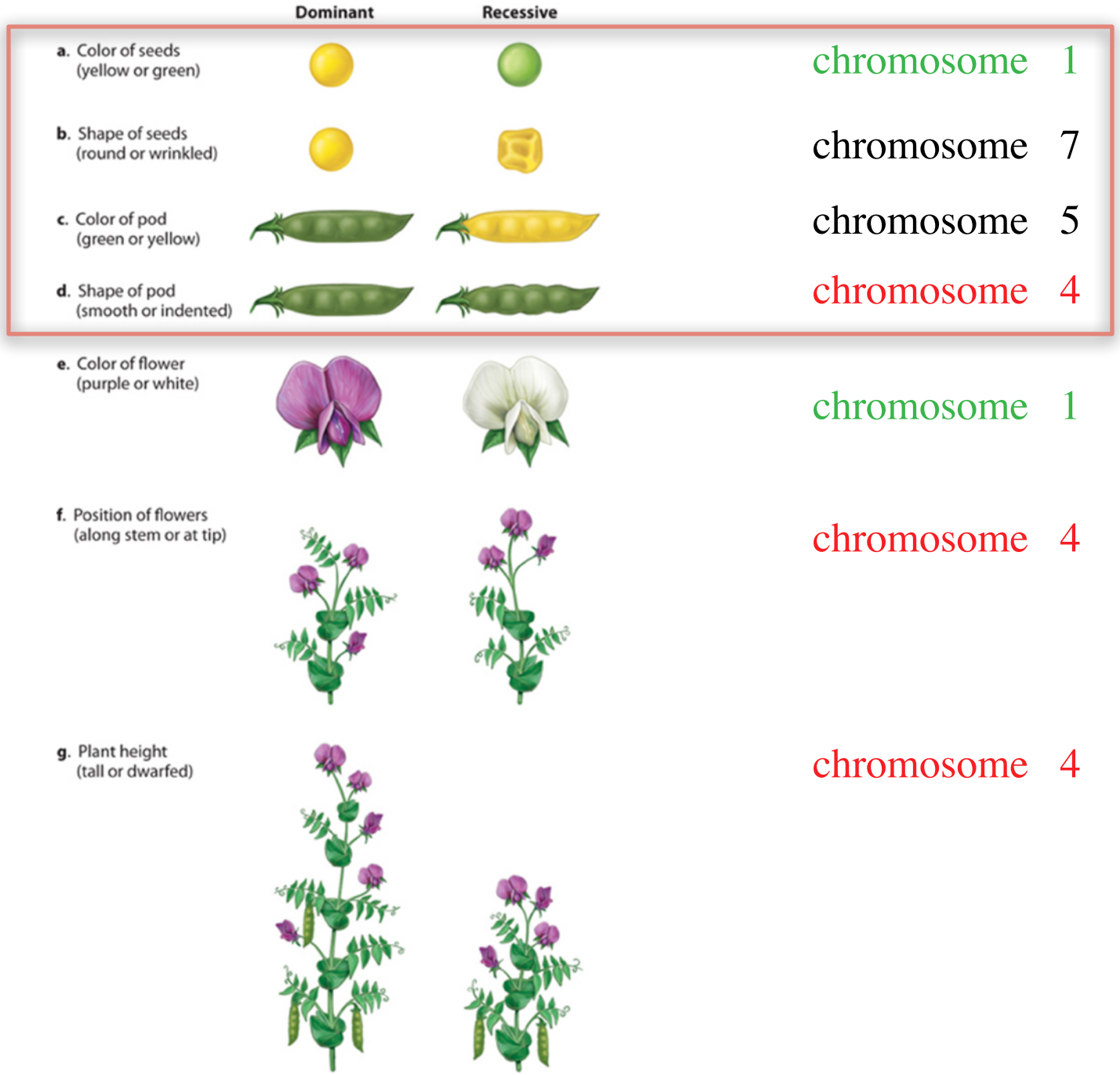
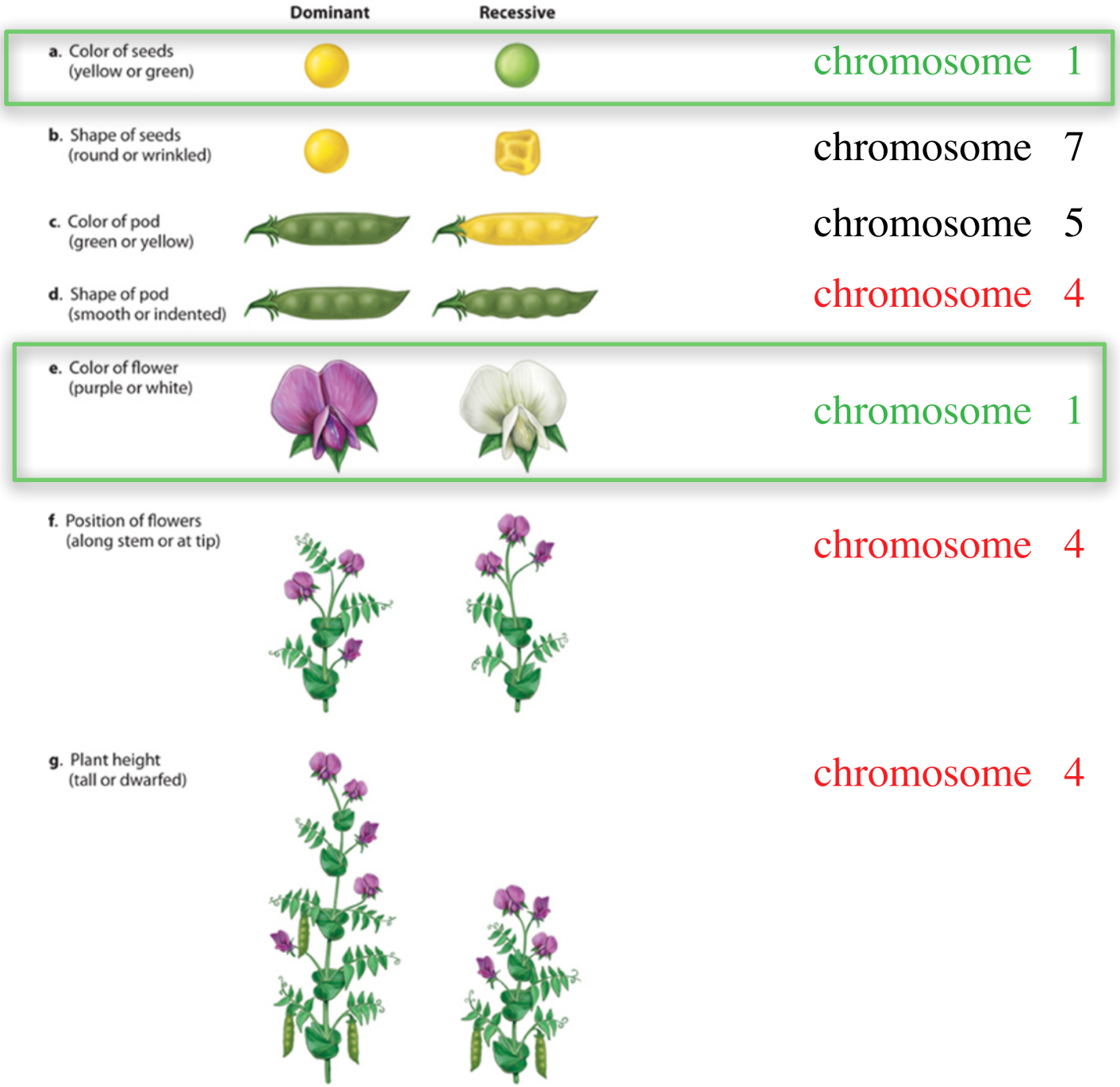



Dominant
Recessive
a. Color of seeds (yellow or green)
b. Shape of seeds (round or wrinkled)
c. Color of pod (green or yellow)
d. Shape of pod (smooth or indented)
e. Color of flower (purple or white)
f. Position of flowers (along stem or at tip)
g. Plant height (tall or dwarfed)


Dominant
Recessive
a. Color of seeds (yellow or green)
chromosome 1
b. Shape of seeds (round or wrinkled)
chromosome 7
c. Color of pod (green or yellow)
chromosome 5
d. Shape of pod (smooth or indented)
chromosome 4
e. Color of flower (purple or white)
chromosome 1
f. Position of flowers (along stem or at tip)
chromosome 4
g. Plant height (tall or dwarfed)
chromosome 4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chromosome 5
d. Shape of pod (smooth or indented)
chromosome 4
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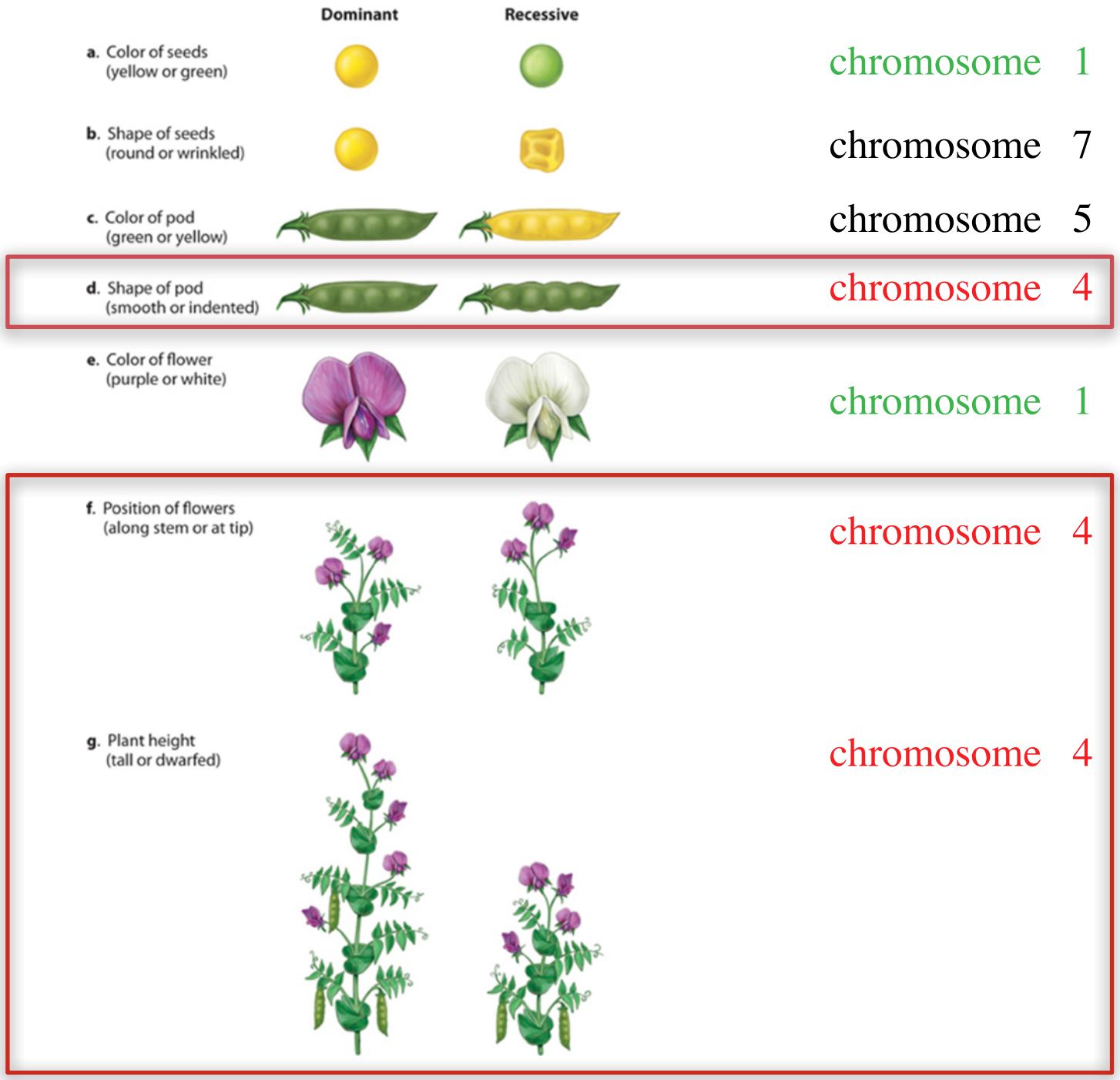
chromosome 1
f. Position of flowers (along stem or at tip)
chromosome 4
g. Plant height (tall or dwarfed)
chromosome 4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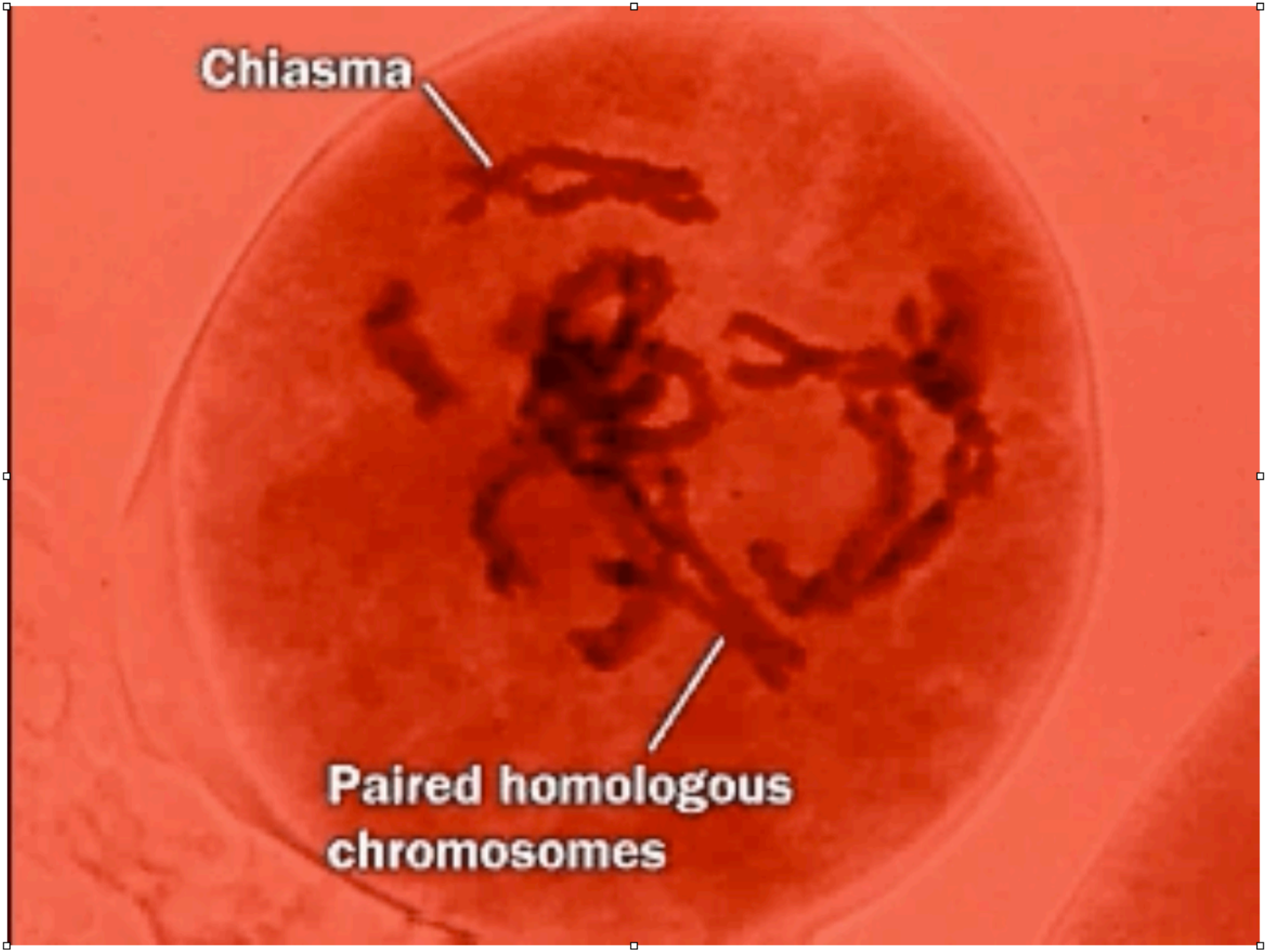
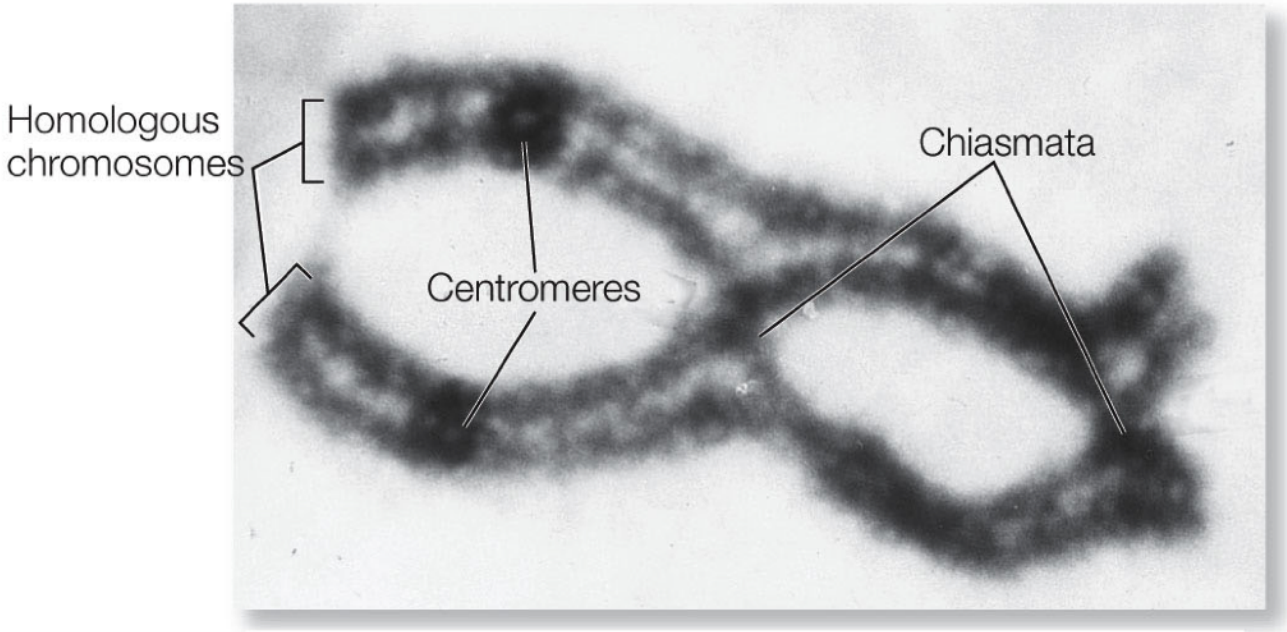
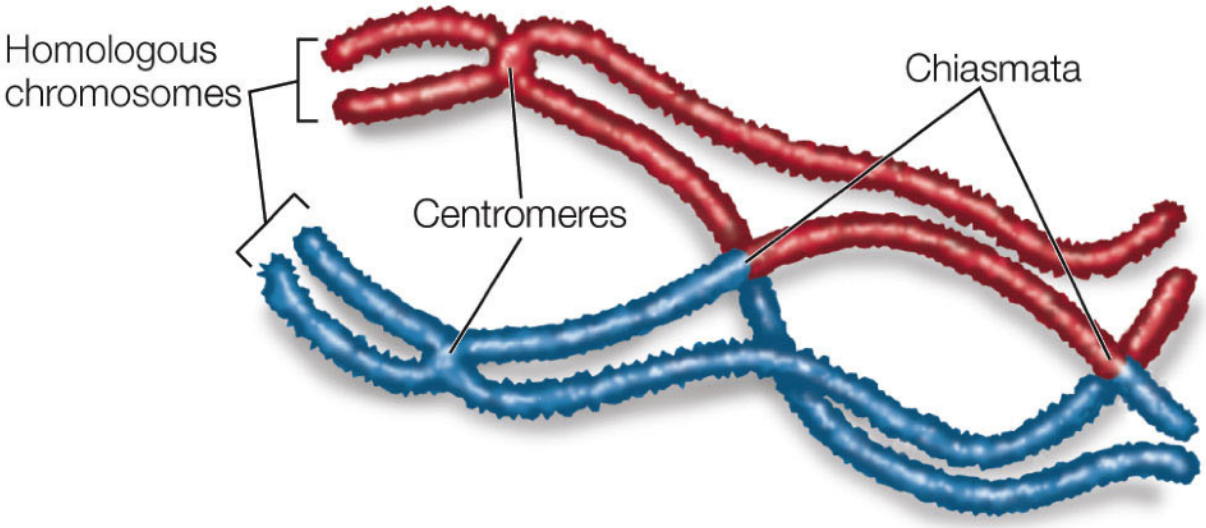
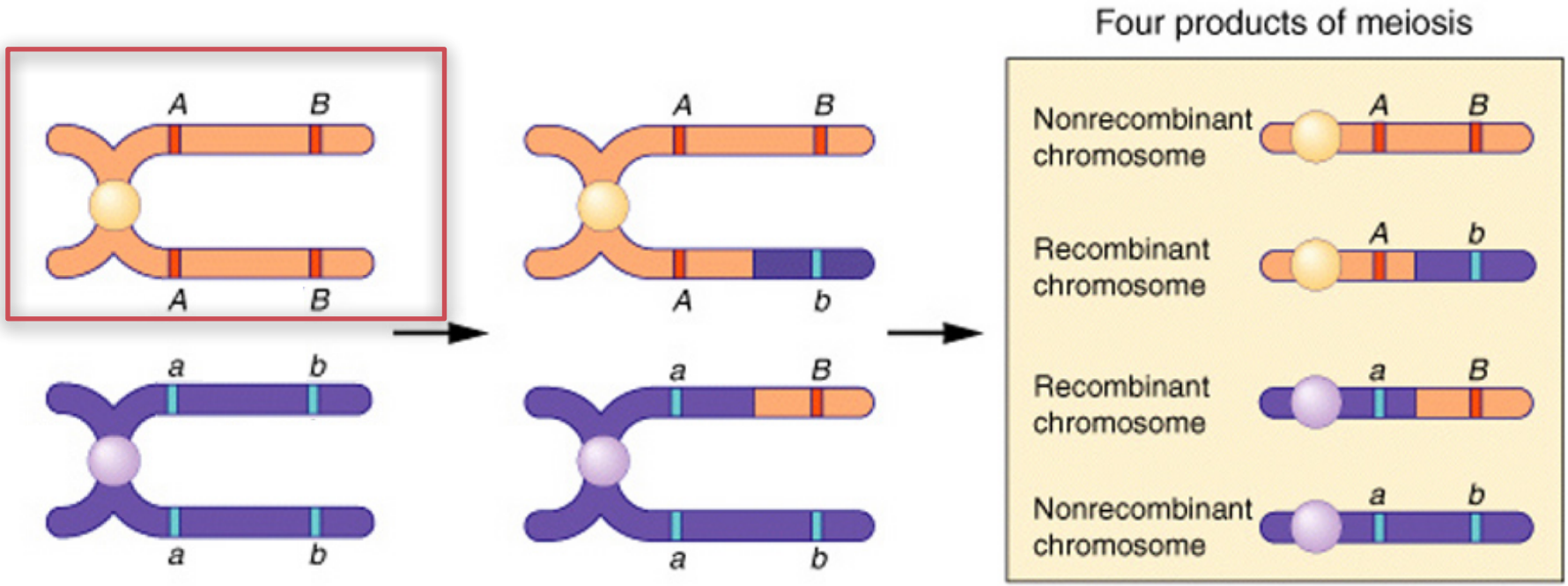
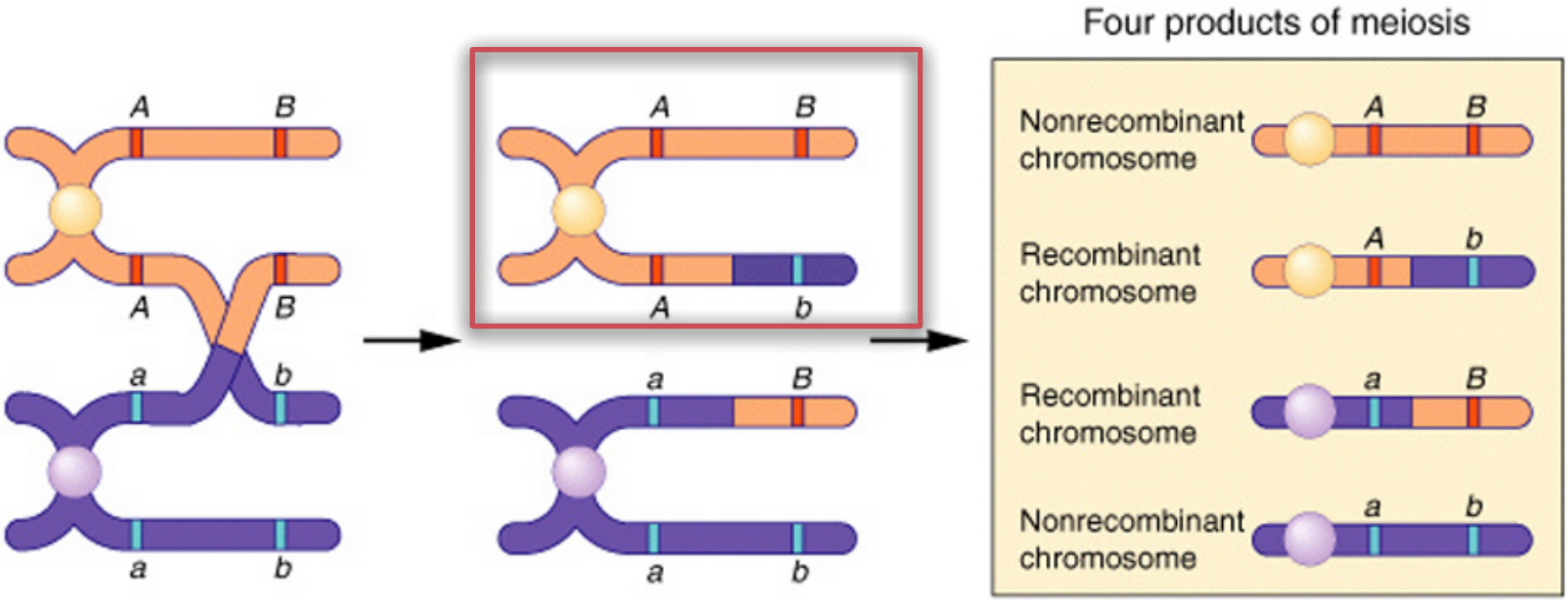
chromosome 1
b. Shape of seeds (round or wrinkled)
chromosome 7
c. Color of pod (green or yellow)
chromosome 5
d. Shape of pod (smooth or indented)
chromosome 4
e. Color of flower (purple or white)
chromosome 1
f. Position of flowers (along stem or at tip)
chromosome 4
g. Plant height (tall or dwarfed)
chromosome 4

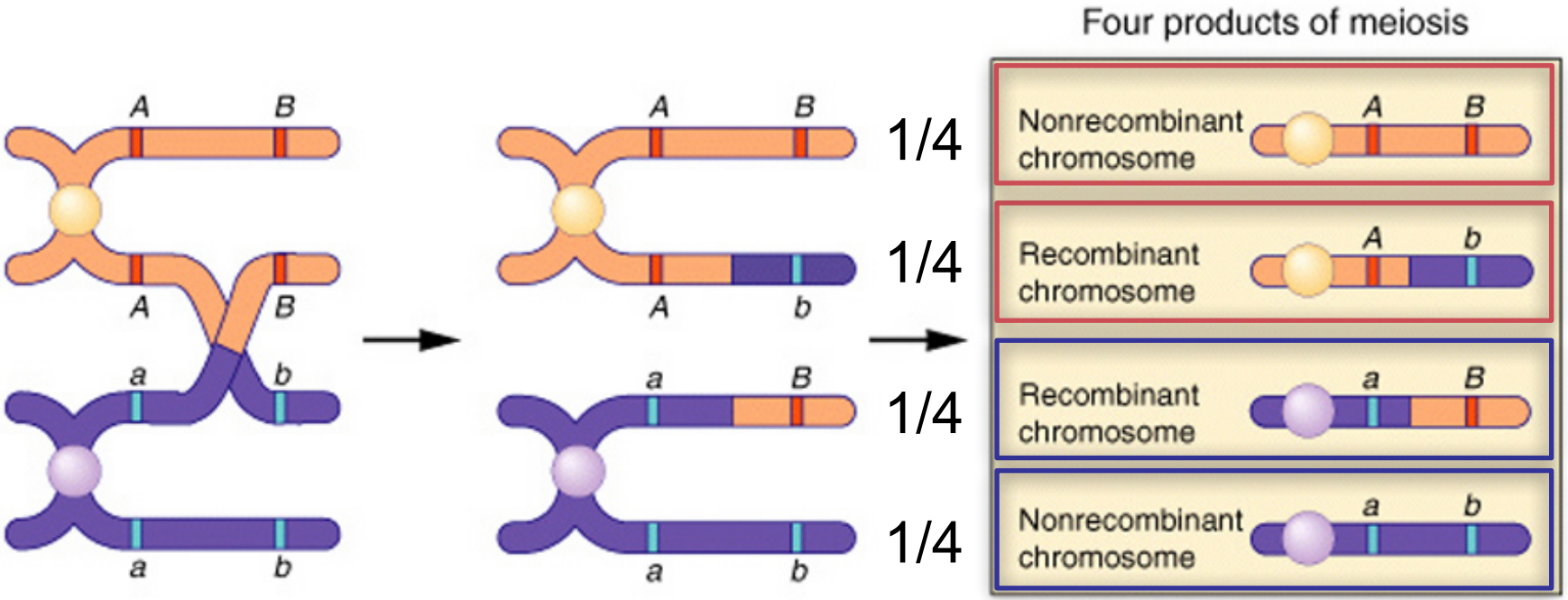

Chiasma
Paired homologous chromosomes


Homologous chromosomes
Chiasmata
Centromeres


Homologous chromosomes
Chiasmata
Centromeres


A
B
A
B
a
b
a
b
A
B
A
b
a
B
a
b
Four products of meiosis
Nonrecombinant chromosome
A
B
Recombinant chromosome
A
b
Recombinant chromosome
a
B
Nonrecombinant chromosome
a
b


A
B
A
B
a
b
a
b
A
B
A
b
a
B
a
b
Four products of meiosis
Nonrecombinant chromosome
A
B
Recombinant chromosome
A
b
Recombinant chromosome
a
B
Nonrecombinant chromosome
a
b



Full agreement with Mendel's 2nd law

Extensions to Mendelian Genetics

Incomplete dominance

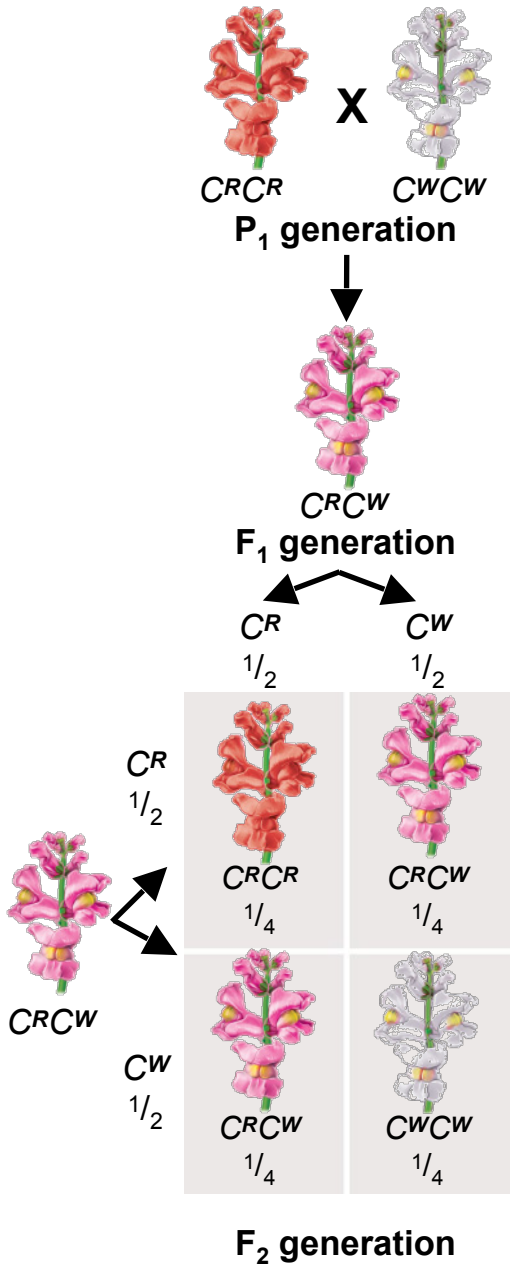
Codominance

Multiple Alleles

Incomplete dominance

Phenotype	Red	Pink	White
Genotype	RR	Rr	rr

Incomplete Dominance



The phenotype of the heterozygous C^RC^W plant is intermediate, an example of incomplete dominance.

The result of segregation can be observed directly, because the ratio of red:pink:white phenotypes is **1 : 2 : 1**, which reflects the ratio of C^RC^R:C^RC^W:C^WC^W genotypes.

Extensions to Mendelian Genetics

Incomplete dominance

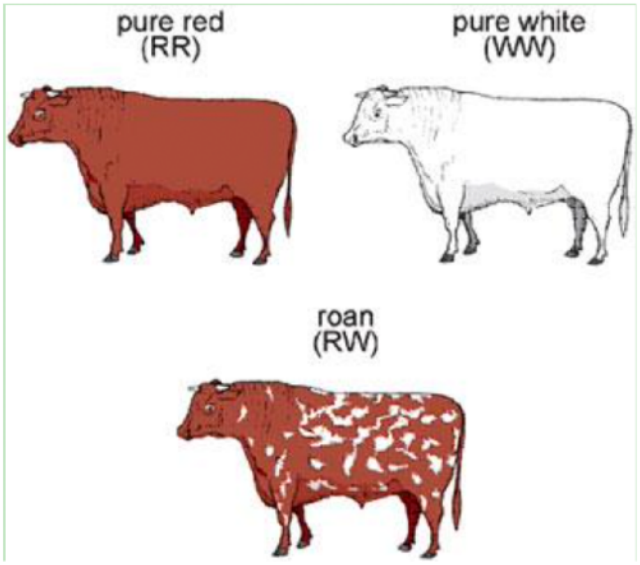
Codominance

Multiple Alleles

Codominance

Phenotype	Red	**Red/white**	White
Genotype	RR	**Rr**	rr

Codominance



Camelias & Cows

Extensions to Mendelian Genetics

Incomplete dominance

Codominance

Lethal Alleles

Multiple Alleles

Huntington's disease

Also called: HD, Huntington's chorea

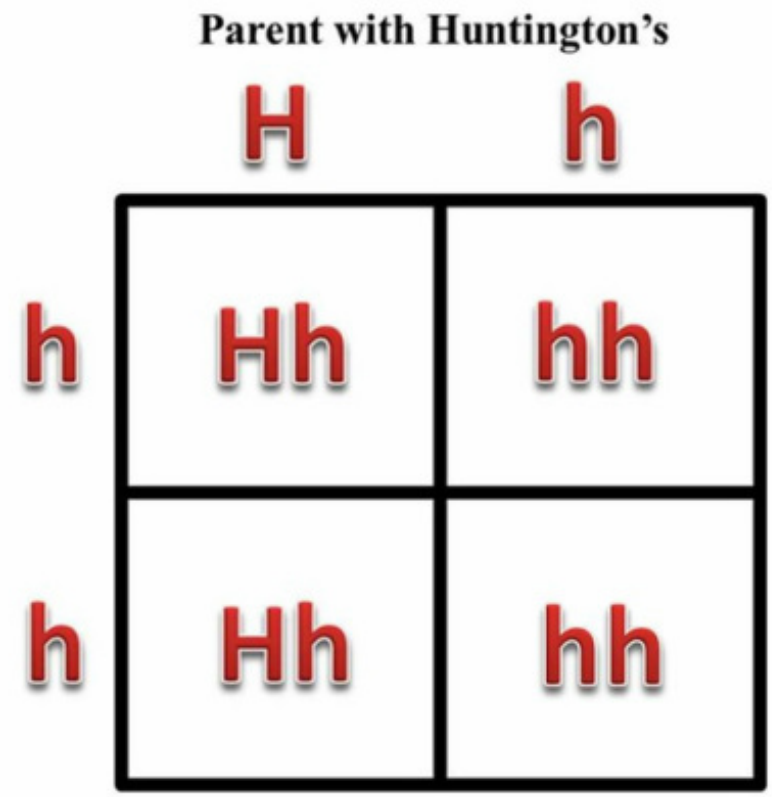
Overview | Symptoms | Treatments | Spe

An inherited condition in which nerve cells in the brain break down over time.

It typically starts in a person's 30s or 40s.
Usually, Huntington's disease results in progressive movement, thinking (cognitive), and psychiatric symptoms.
No cure exists, but drugs, physical therapy, and talk therapy can help manage some symptoms.

HH Hh hh

Hh x hh


Parent with Huntington's
H
h
h
Hh
hh
h
Hh
hh

Parent with Huntington's
H
h
Parent with Huntington's
H
HH
Hh
h
Hh
hh

Hh x hh

	Parent with Huntington's: H	Parent with Huntington's: h
h	Hh	hh
h	Hh	hh

Parent with Huntington's

	H	h
H	HH	Hh
h	Hh	hh

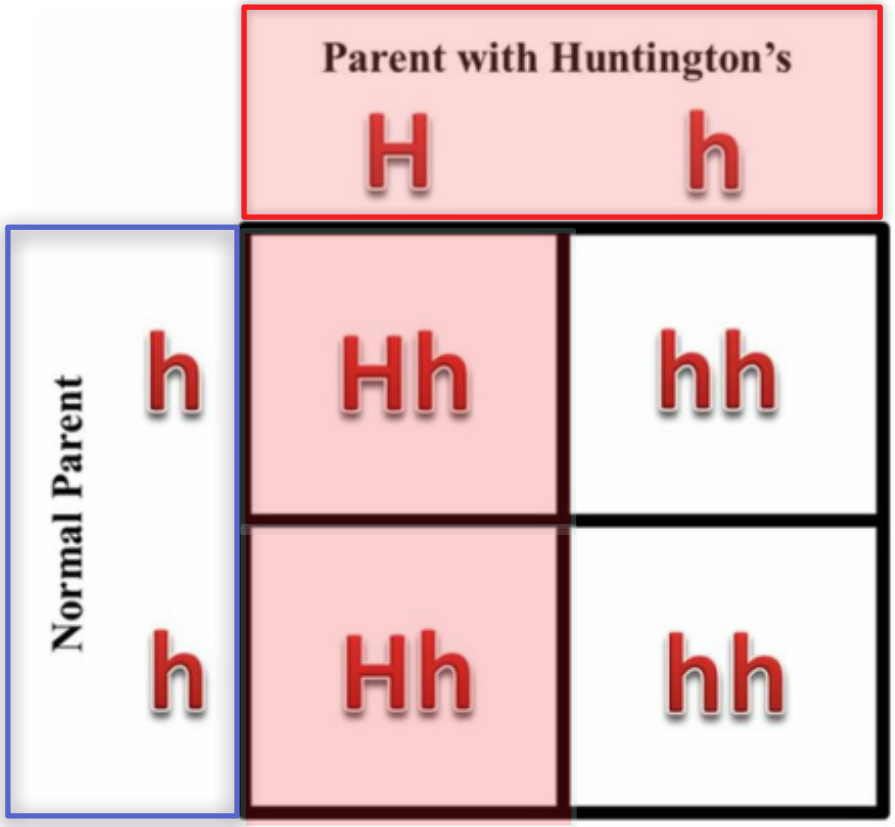
Hh x hh

	Parent with Huntington's: H	Parent with Huntington's: h
h	Hh	hh
h	Hh	hh

Parent with Huntington's

	H	h
H	HH	Hh
h	Hh	hh

Hh x hh


Parent with Huntington's
H
h
Normal Parent
h
h
Hh
hh
Hh
hh

Parent with Huntington's
H
h
Parent with Huntington's
H
h
HH
Hh
Hh
hh

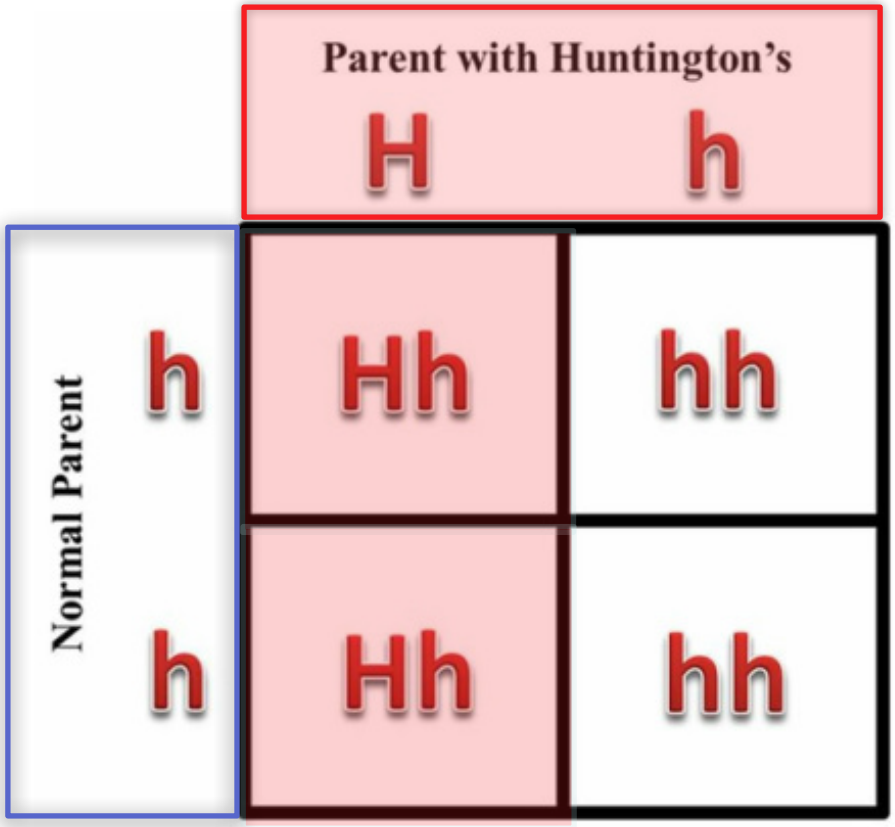
Hh x hh

Normal Parent	Parent with Huntington's H	Parent with Huntington's h
h	Hh	hh
h	Hh	hh

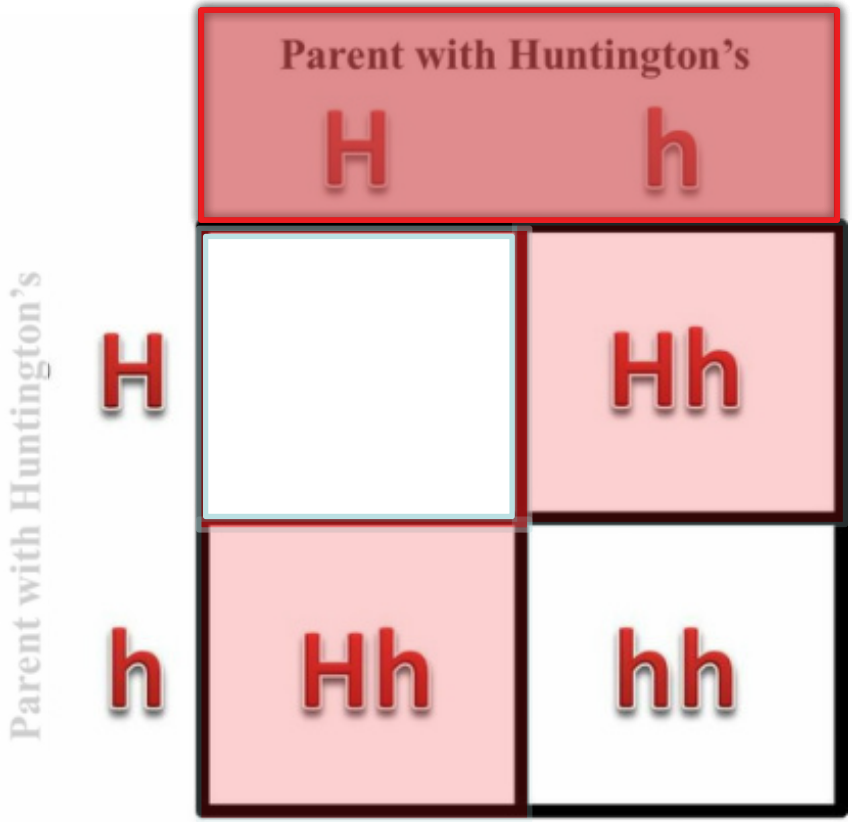
Hh x Hh

Parent with Huntington's	Parent with Huntington's H	Parent with Huntington's h
H	HH	Hh
h	Hh	hh

Hh x hh

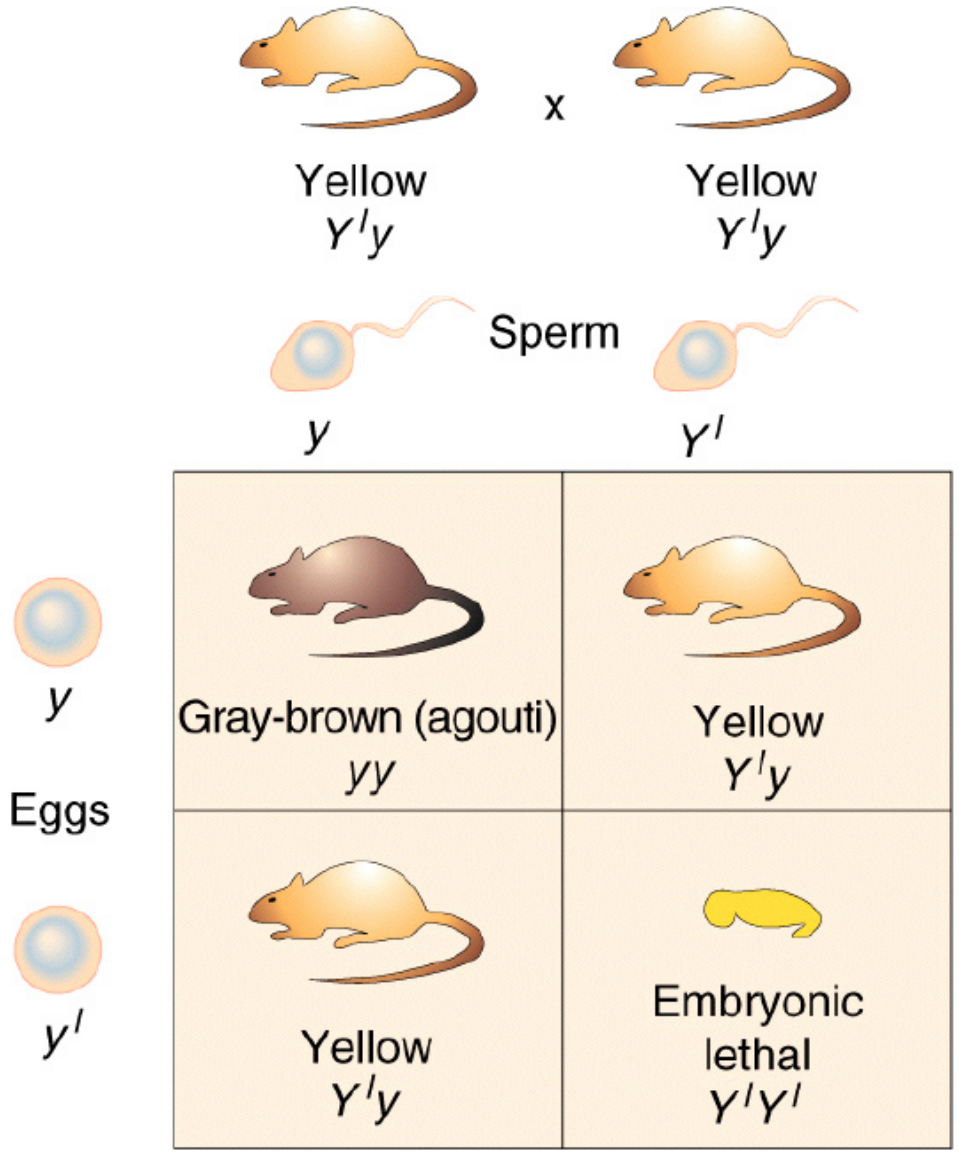

Parent with Huntington's
H
h
Normal Parent
h
h
Hh
hh
Hh
hh

Hh x Hh


Parent with Huntington's
H
h
Parent with Huntington's
H
h
Hh
Hh
hh

Lethal Alleles

Y' is often designated $\mathbf{A}^{y}$ which is dominant over **y** or $\mathbf{a}^{+}$



Extensions to Mendelian Genetics

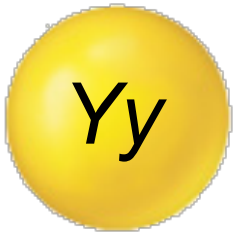
Incomplete dominance

Codominance

Lethal Alleles

Multiple Alleles

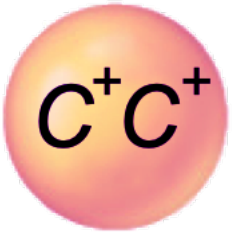
Homozygous DOMINANT



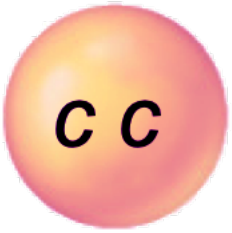
Heterozygous

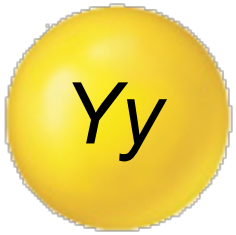
Homozygous recessive

Homozygous DOMINANT

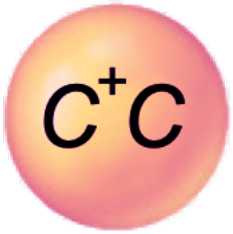


wild type

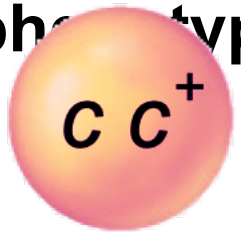




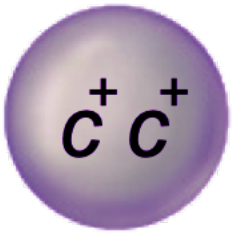
Heterozygous



wild type (ph type)



Homozygous recessive



wild type

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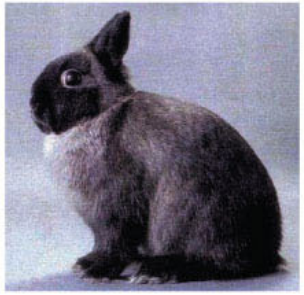
HH Hh hh

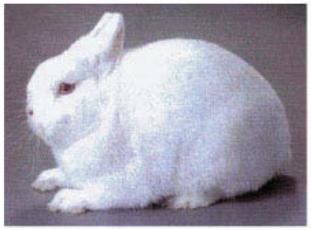
wild type

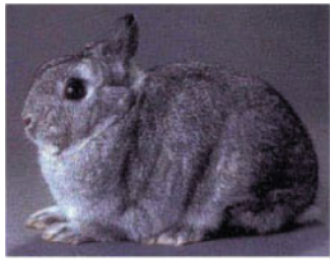
Multiple Alleles

Possible genotypes	CC, Cc^{ch}, Cc^{h}, Cc	$c^{ch}c^{ch}$	$c^{ch}c^{h}$, $c^{ch}c$	$c^{h}c^{h}$, $c^{h}c$	cc
Phenotype	Dark gray	Chinchilla	Light gray	Point restricted	Albino









Genotype	Phenotype
cc	White hairs over the entire body
$c^h c^h$	Black hairs on the extremities; white hairs everywhere else
$c^{ch} c^{ch}$	White hair with black tips on the body
$c^+ c^+$	Colored hairs over the entire body

Albino

Himalayan

Chinchilla

Wild-type

	Genotype	Phenotype
 Albino	cc	White hairs over the entire body
 Himalayan	$c^h c^h$	Black hairs on the extremities; white hairs everywhere else
 Chinchilla	$c^{ch} c^{ch}$	White hair with black tips on the body
 Wild-type	$c^+ c^+$	Colored hairs over the entire body

c^+c
c^+c^{ch}
c^+c^h

Wild-type

$c^{ch}c$

Light chinchilla

$c^{ch}c^h$

Light chinchilla with black tips

c^hc

Himalayan

Genotype

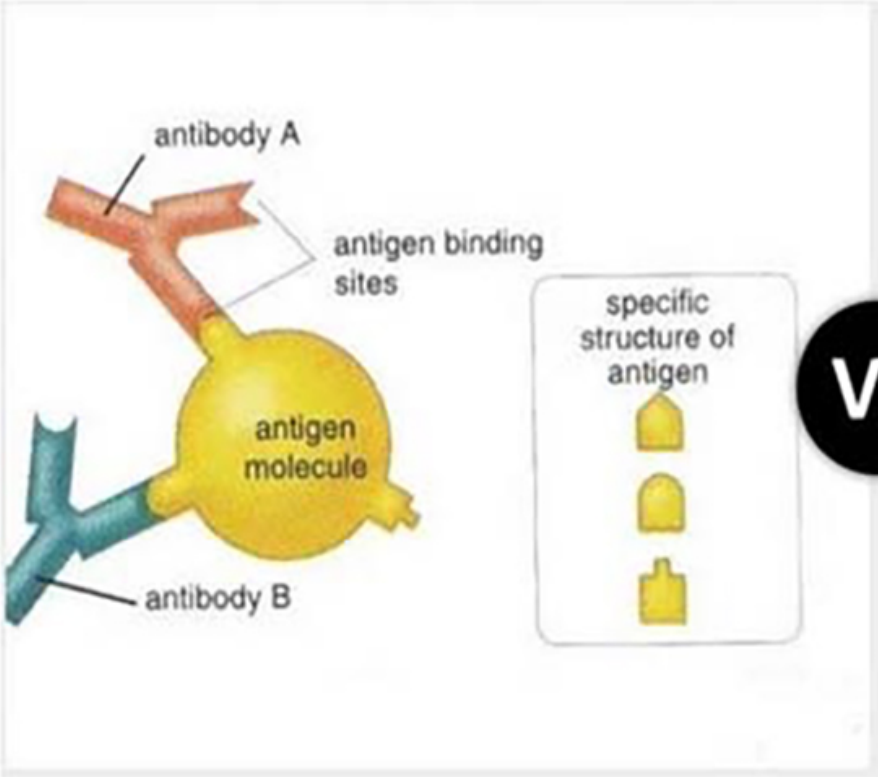
cc

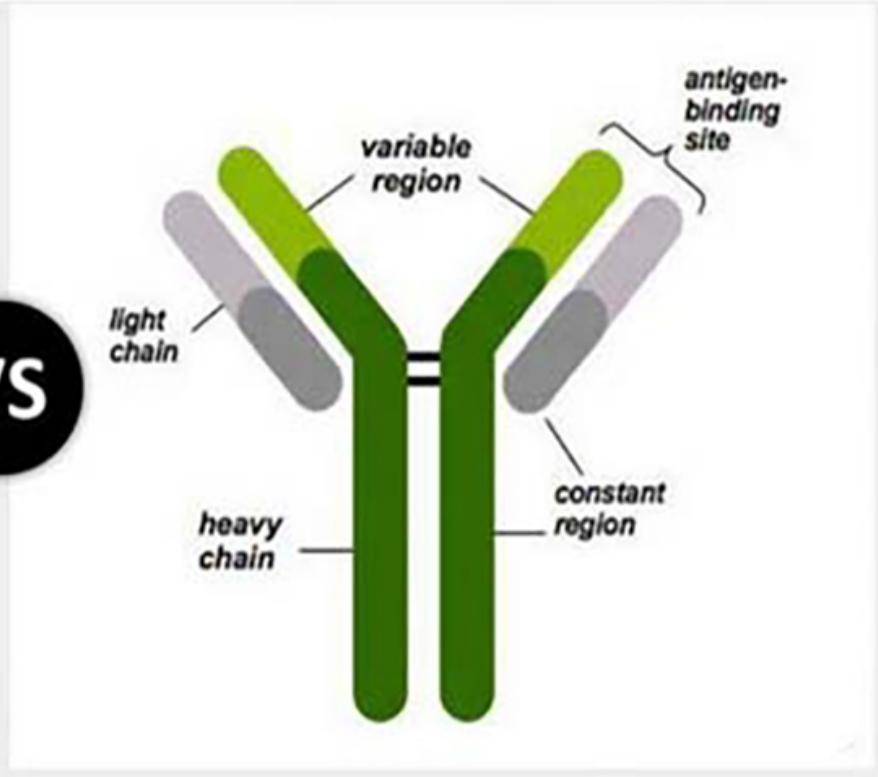
Albino

Figure 4.4 Phenotypes of different combinations of *c* alleles in rabbits. The alleles form a series, with the wild-type allele, c^+, dominant over all the other alleles and the null allele, *c* (albino), recessive to all the other alleles; one hypomorphic allele, c^{ch} (chinchilla), is partially dominant over the other, c^h (himalayan).

Antigens

Antibody





Some of the differences are:

S.N.	Characteristics	Antigen	Antibody
1	Molecule Type	Usually, proteins may also be polysaccharides, lipids or nucleic acids.	Proteins
2	Definition	These are substances that provoke an immune response.	These are Glycoproteins that are secreted by immune cells (plasma cells) in response to a foreign substance (antigen).
3	Effect	Cause disease or allergic reactions.	Protect the system by lysis of antigenic material.
4	Origin	Within the body or externally.	Within the body.

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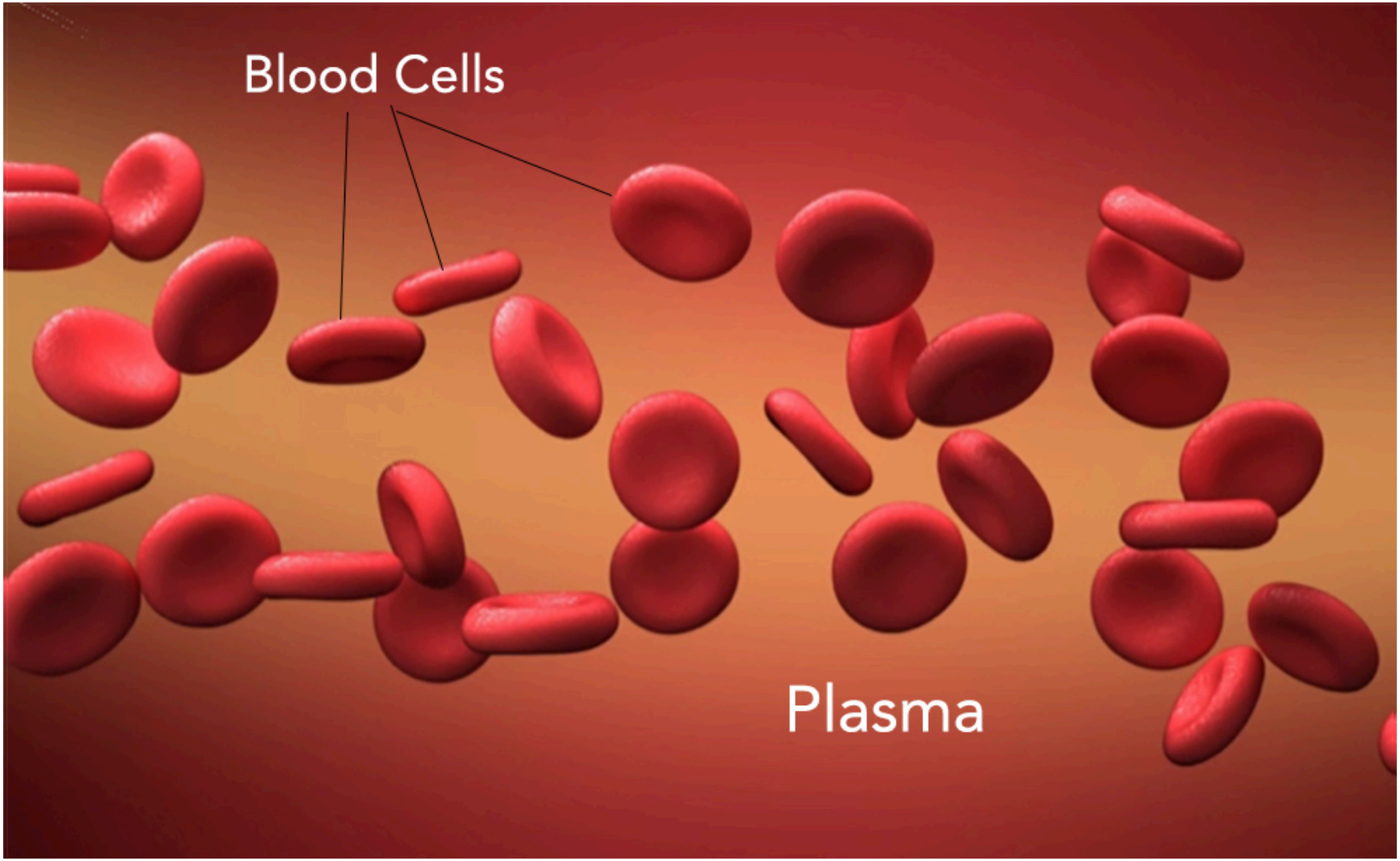
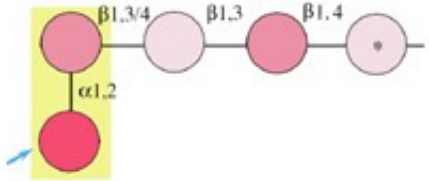
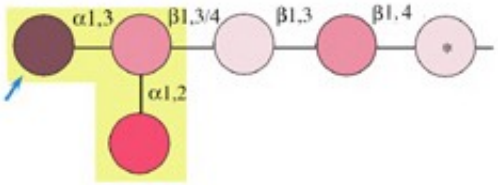

Blood Cells
Plasma

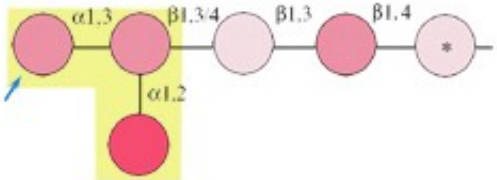
H - antigen =

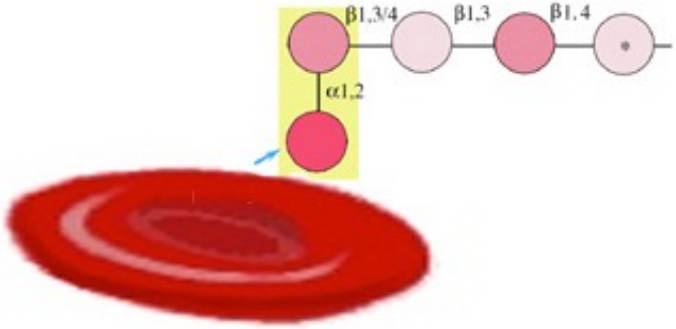


A - antigen =



B - antigen =





An example of "co-dominant" alleles in humans

The ABO Blood Group System

Antigens: molecules, usually on the outside of a cell, that provoke an immune response

Genetics of the ABO System

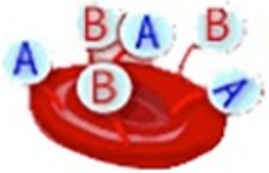
A person with at least one A gene will produce the A protein

Type A

A person with at least one B gene will produce the B protein

Type B

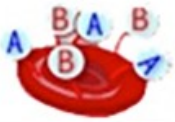
A person with one A gene and one B gene will produce both proteins



Type AB

A person with neither A nor B gene will not produce either protein

Type O

		Potential Donors			
Blood Type	**Antibodies Produced**				
A		+	-	-	+
B		-	+	-	+
AB	None	+	+	+	+
O		-	-	-	+

RECIPIENT

DONOR

Alleles & Antibodies	O anti-A anti-B	A anti-B	B anti-A	AB None
O	None	None	None	None
A	Clump	None	Clump	None
B	Clump	Clump	None	None
AB	Clump	Clump	Clump	None

Extensions to Mendelian Genetics

Incomplete dominance

Codominance

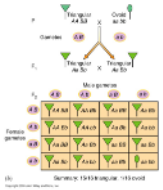
Lethal Alleles

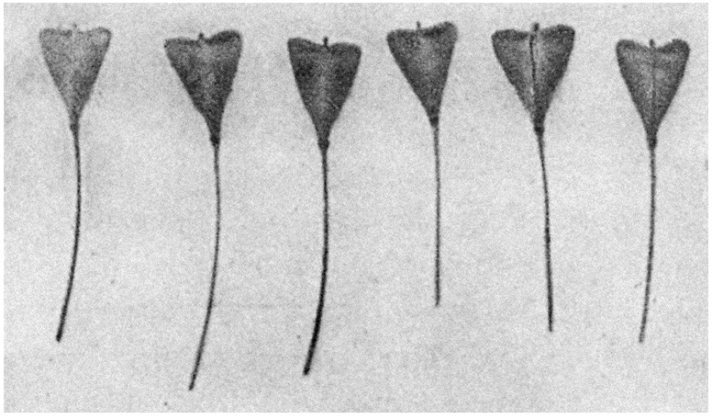
Multiple Alleles

Multiple Genes

Duplicate Genes

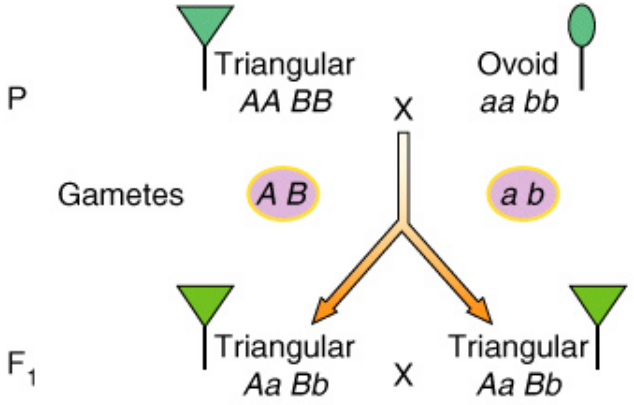
Courtesy New York Public Library

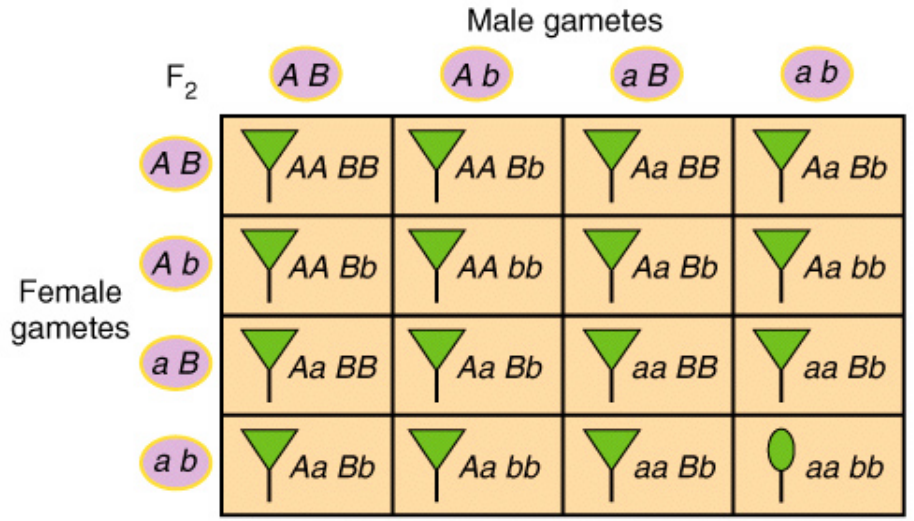




Courtesy New York Public Library

Duplicate Genes

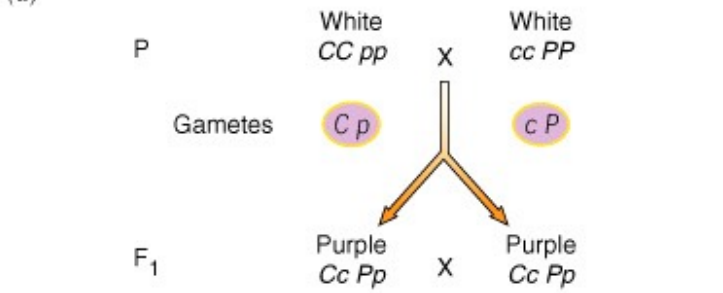




(b) Summary: 15/16 triangular, 1/16 ovoid

Complementary Genes

(a)



Male gametes

F2	C P	C p	c P	c p
C P	CC PP Purple	CC Pp Purple	Cc PP Purple	Cc Pp Purple
C p	CC Pp Purple	CC pp White	Cc Pp Purple	Cc pp White
c P	Cc PP Purple	Cc Pp Purple	cc PP White	cc Pp White
c p	Cc Pp Purple	Cc pp White	cc Pp White	cc pp White

Female gametes

(b)

Summary: 9/16 purple, 7/16 white

Two Genes
E and B

(A)

Epistasis

(B)

(C)

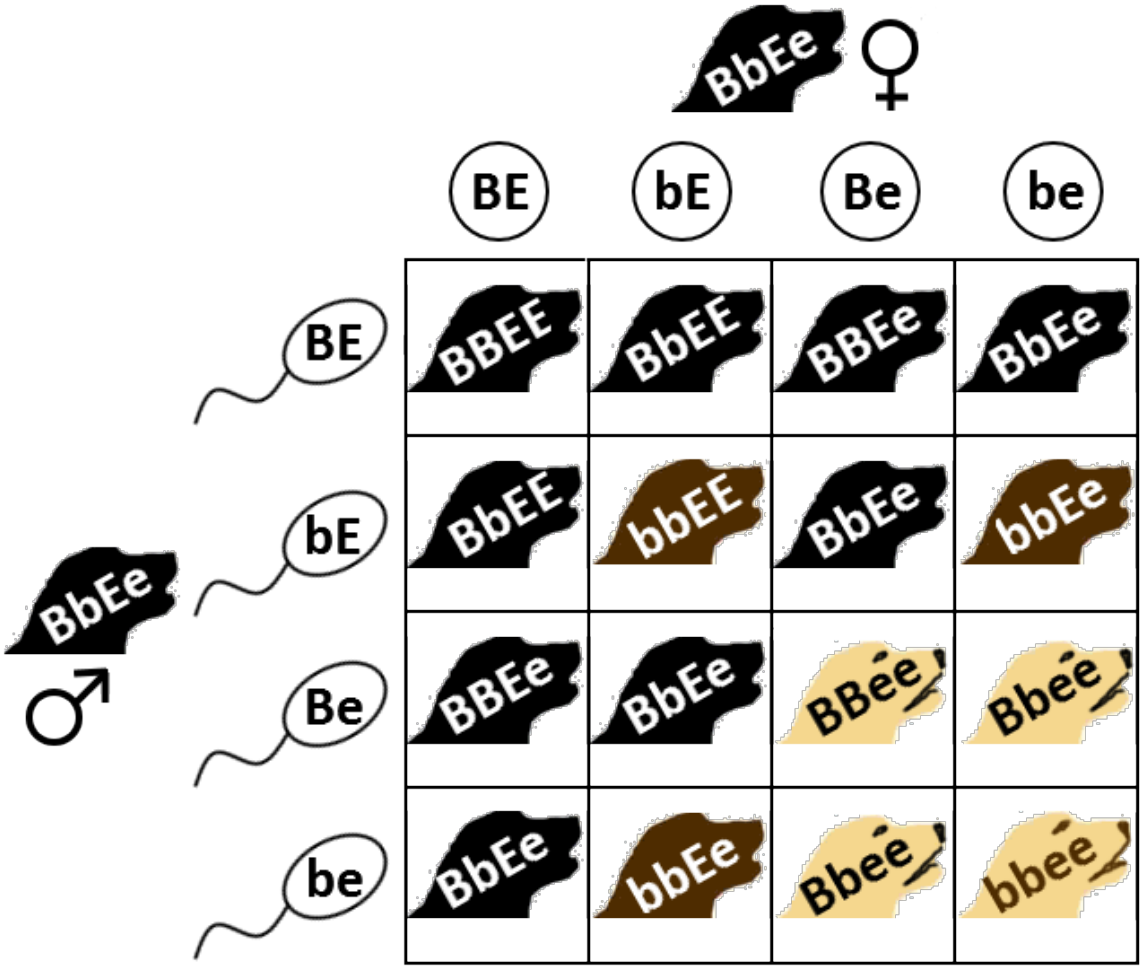
Epistasis

(A) Black labrador (*B_E_*)

(B) Chocolate labrador (*bbE_*)

(C) Yellow labrador (*_ _ee*)

Epistasis



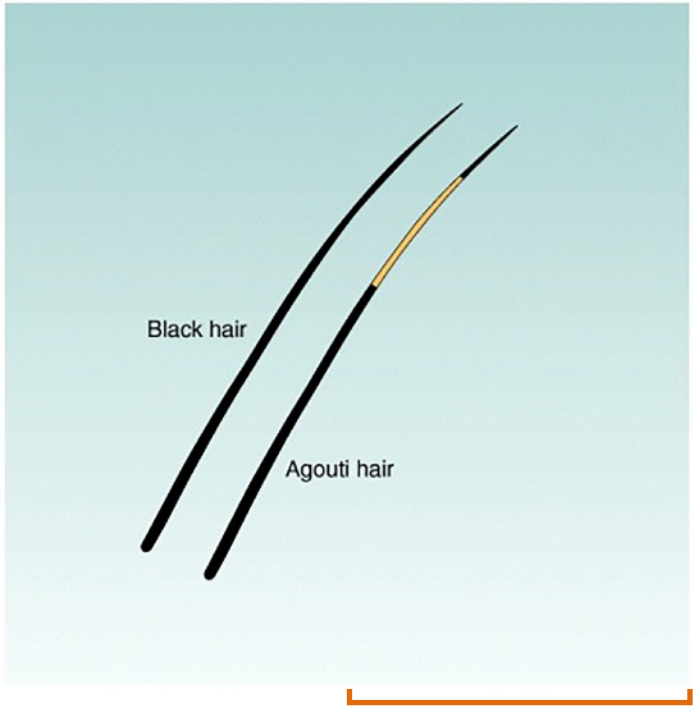
9 B-E- Black

3 bbE- Chocolate

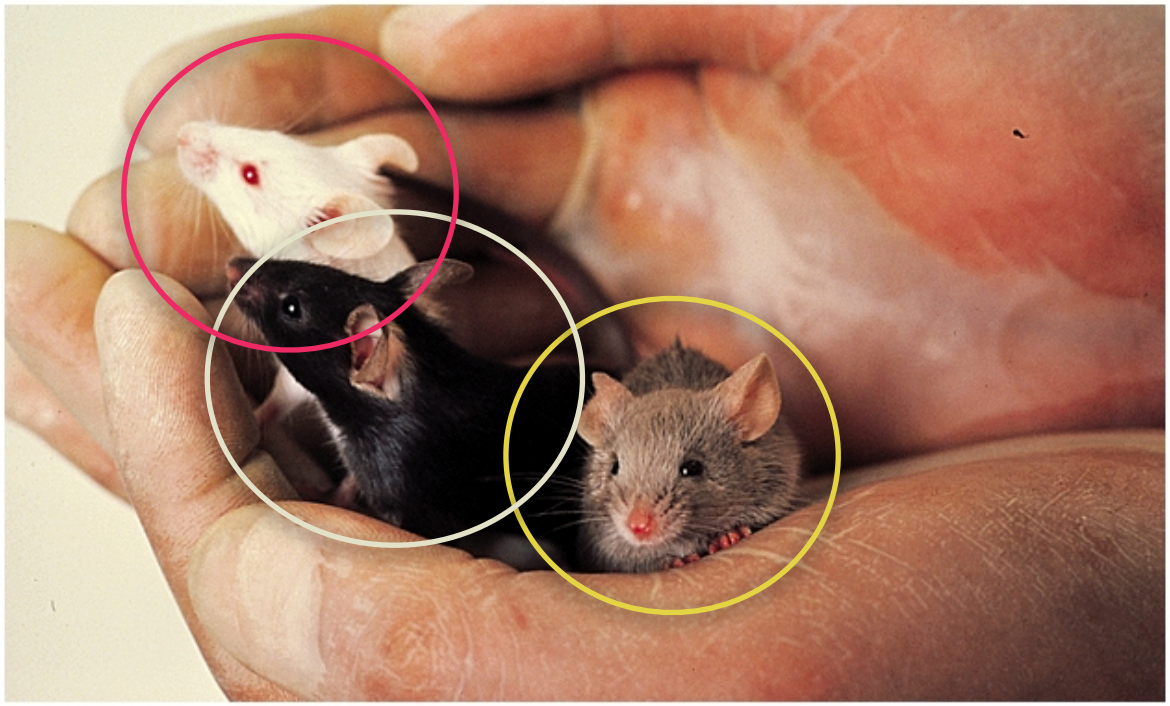
4 --ee Yellow

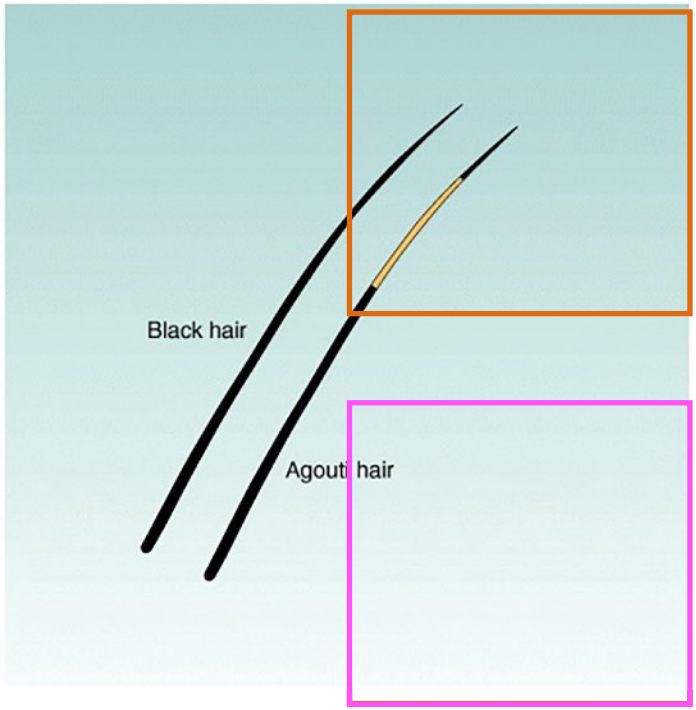
1 is Dudley (bbee)

Several Interactive genes



An **F2 phenotypic ratio** of an initial parental cross between a **BB, AA** and a **bb, aa** would be:





An **F2 phenotypic ratio** of an initial parental cross between a **BB, AA** and a **bb, aa** would be:

A 3rd gene **C**, which when present in **CC** or **Cc** allows all colours that we have mentioned.. but if present as **cc**, then it BLOCKS ALL coloration of fur and eye colour -giving **albino**

Multiple or "Poly" Genes

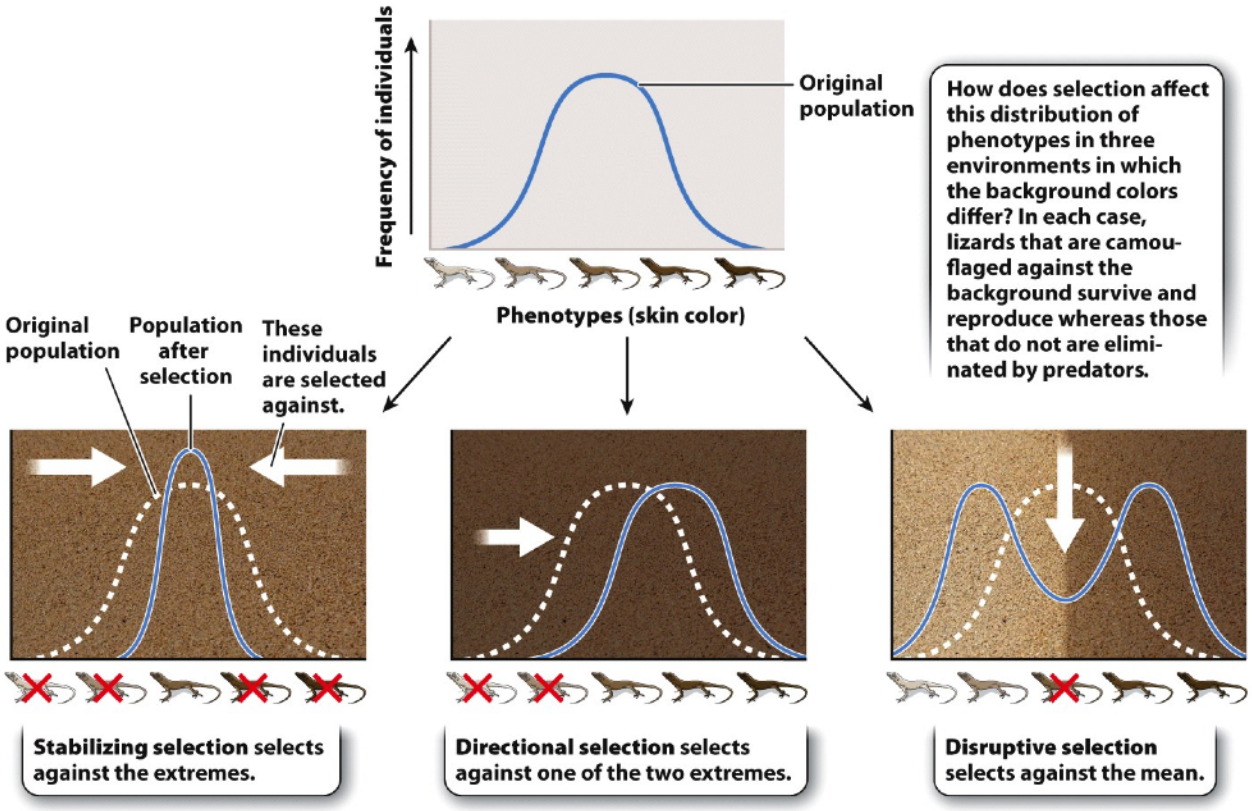


Figure 21.9
Biology: How Life Works

Multiple or “Poly” Genes

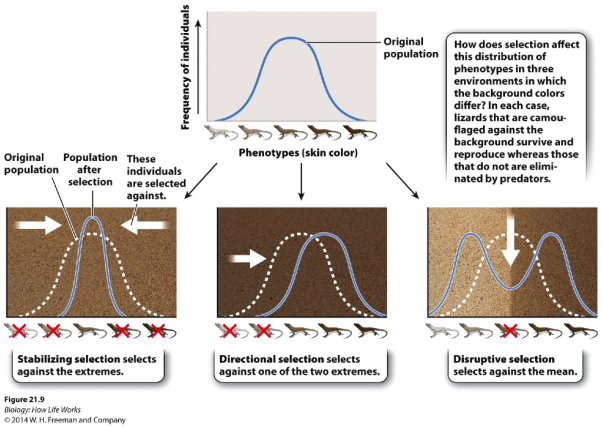


Figure 21.9
Biology: How Life Works

>50 Genes involved directly in structural height integrity in humans

Multiple Genes affecting the same trait (Polygenes)

Human Height?

Science News *from research organizations*

Number of genes linked to height revealed by study

Date: October 5, 2014

Source: Boston Children's Hospital

Summary: The largest genome-wide association study to date, involving more than 300 institutions and more than 250,000 subjects, roughly doubles the number of known gene regions influencing height to more than 400. The study provides a better glimpse at the biology of height and offers a model for investigating traits and diseases caused by many common gene changes acting together.

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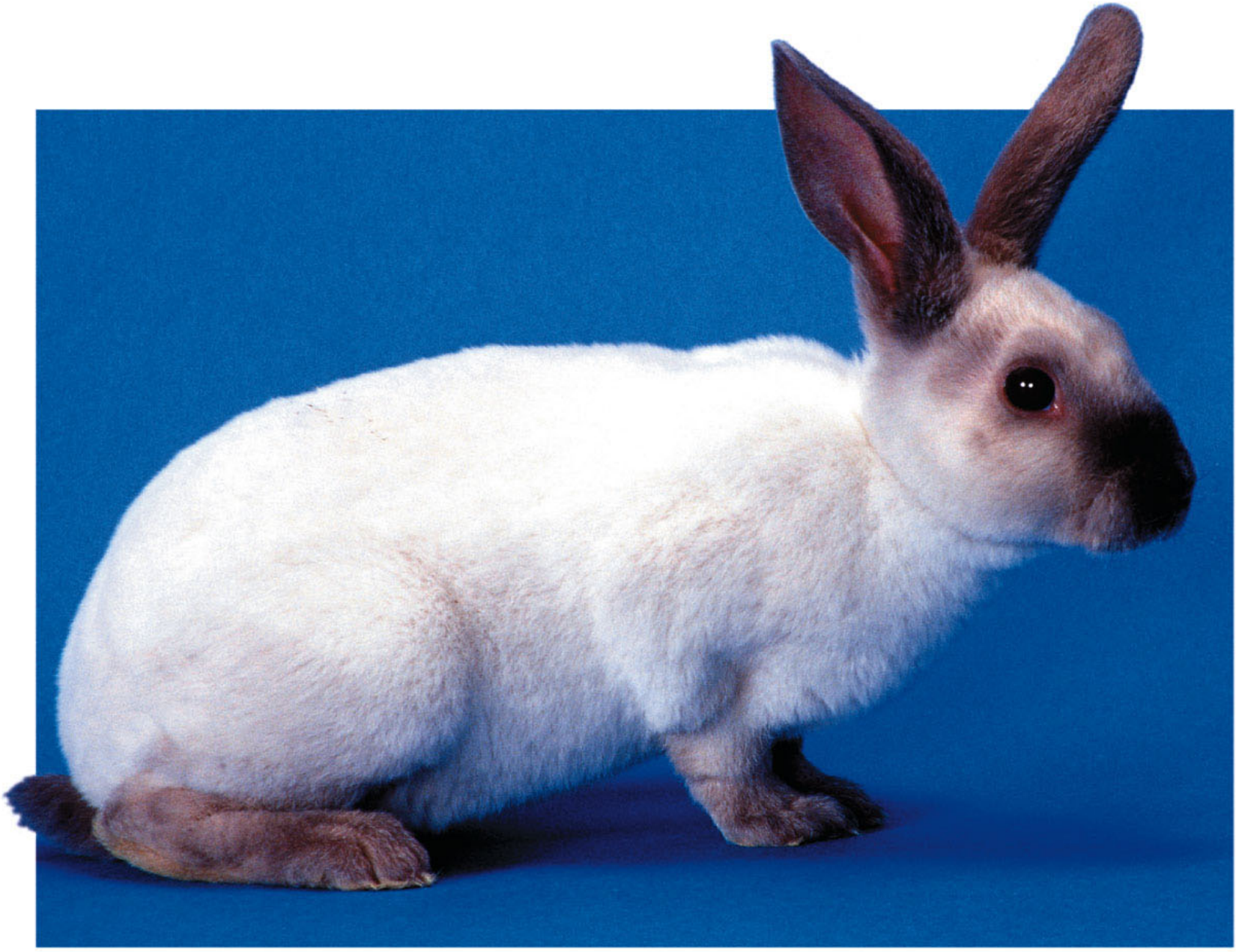

***LIFE* 8e, Figure 10.16**