

Genetics and Heredity

History

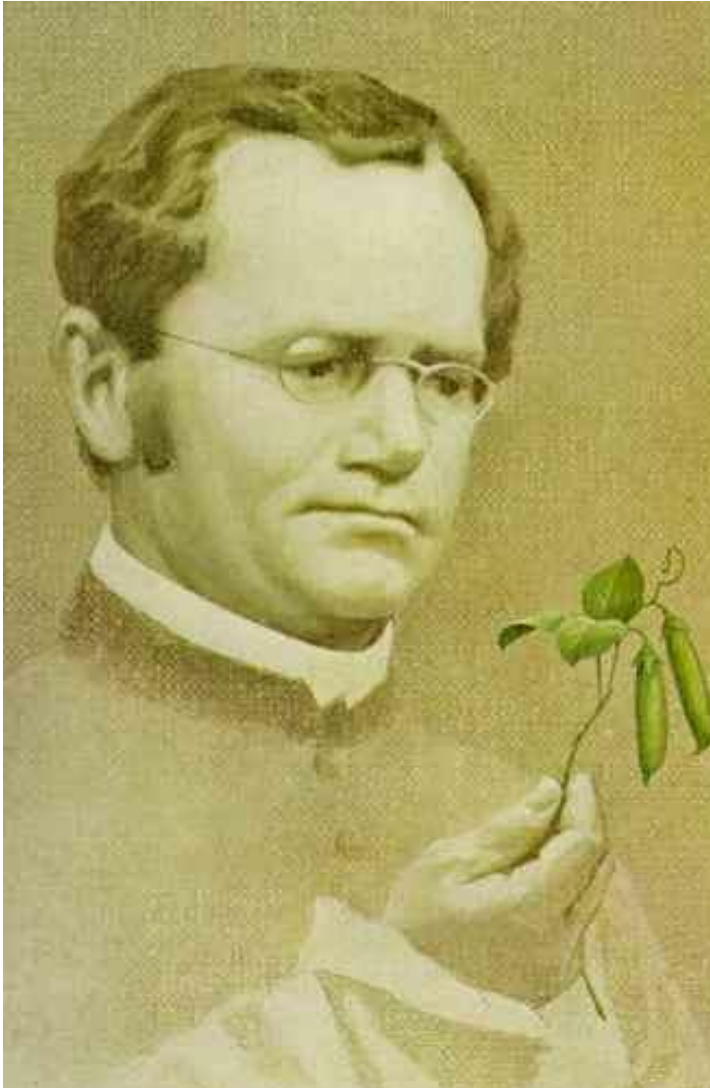
- **Genetics** is the study of genes.
- **Inheritance** is how traits, or characteristics, are passed on from generation to generation.
- Chromosomes are made up of genes, which are made up of DNA.
- Genetic material (genes, chromosomes, DNA) is found inside the nucleus of a cell.
- **Gregor Mendel** is considered “The Father of Genetics”

Gregor Mendel

- Austrian Monk.
- Experimented with “pea plants”.
- Used pea plants because:
 - They were available
 - They reproduced quickly
 - They showed obvious differences in the traits

Understood that there was something that carried traits from one generation to the next- **“FACTOR”**.

Mendel cont.....



In the mid-1800s, the rules underlying patterns of inheritance were uncovered in a series of experiments performed by an Austrian monk named Gregor Mendel.

Mendel's Plant Breeding Experiments

Gregor Mendel was one of the first to apply an experimental approach to the question of inheritance.

For seven years, Mendel bred pea plants and recorded inheritance patterns in the offspring.

Particulate Hypothesis of Inheritance

Parents pass on to their offspring separate and distinct factors (today called genes) that are responsible for inherited traits.

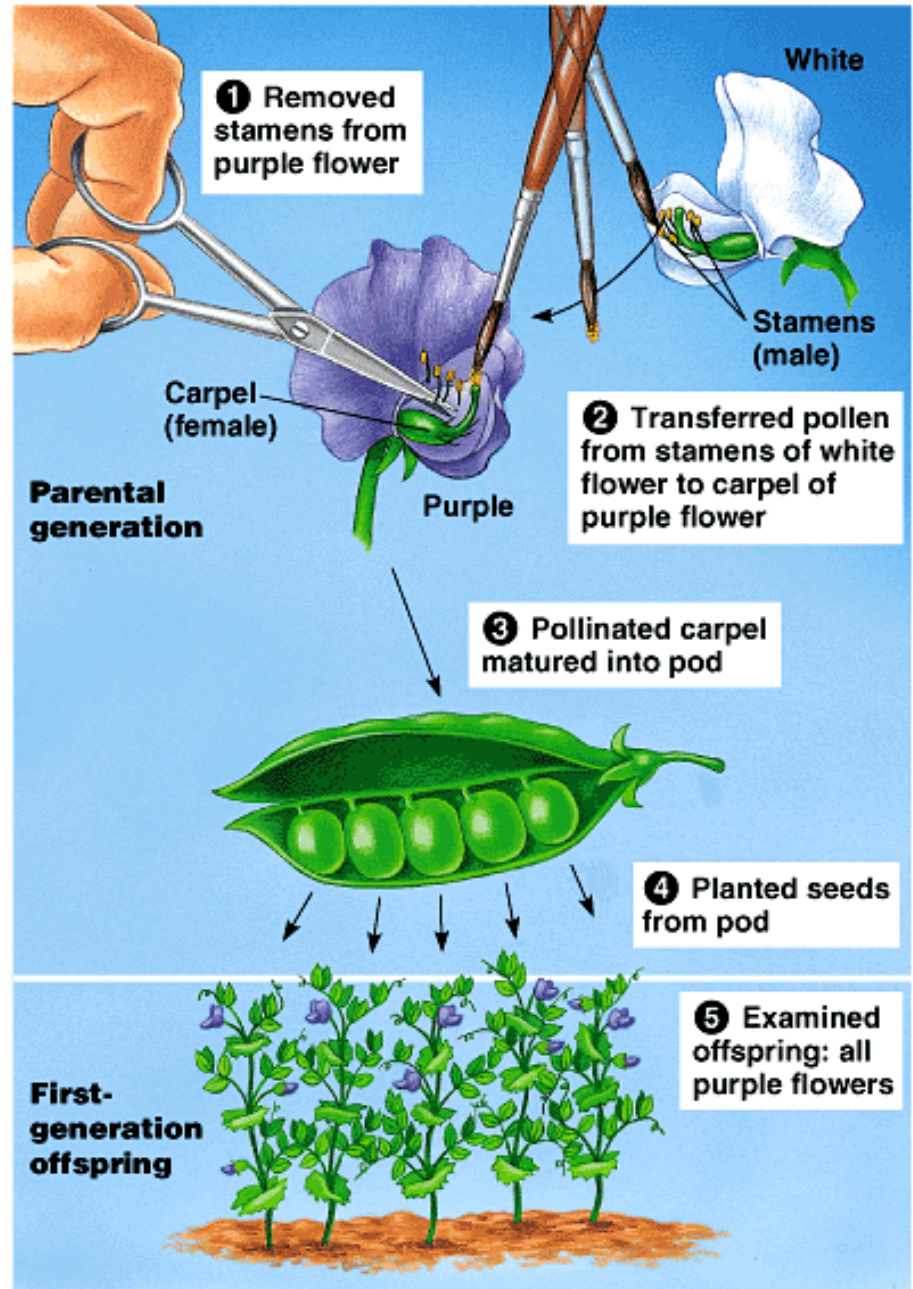


Mendelian Genetics

- **Dominant** traits- traits that are expressed.
- **Recessive** traits- traits that are covered up.
- **Alleles**- the different forms of a characteristic.
- **Punnett Squares**- show how crosses are made.
- **Probability**- the chances/ percentages that something will occur.
- **Genotype**- the types of genes (Alleles) present.
- **Phenotype**- what it looks like.
- **Homozygous**- two of the same alleles.
- **Heterozygous**- two different alleles.

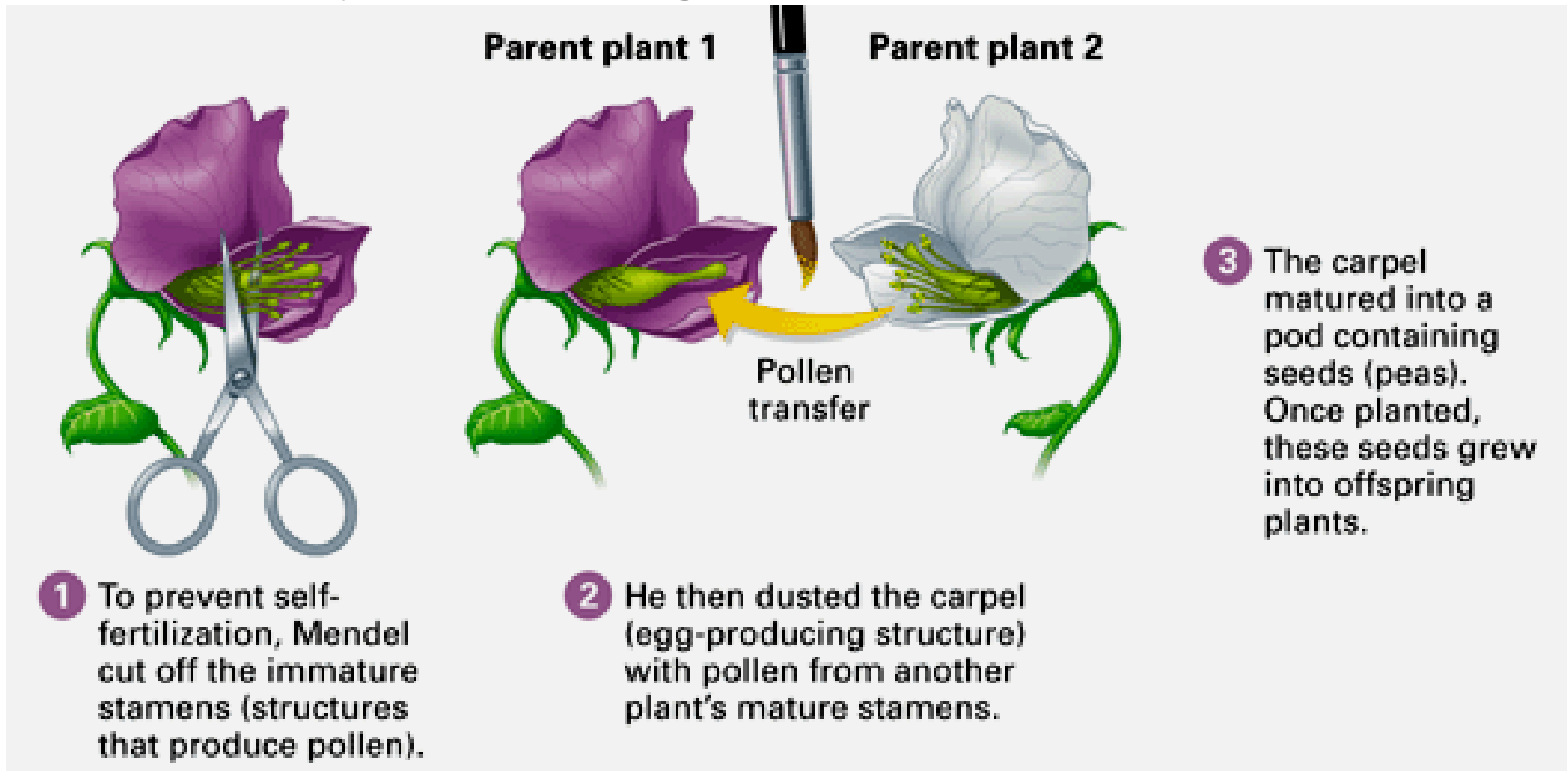
Mendel was fortunate he chose the Garden Pea

- Mendel probably chose to work with peas because they are available in many varieties.
- The use of peas also gave Mendel strict control over which plants mated.
- Fortunately, the pea traits are distinct and were clearly contrasting.



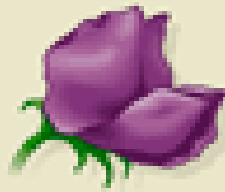
To test the particulate hypothesis, Mendel crossed true-breeding plants that had two distinct and contrasting traits—for example, purple or white flowers.

What is meant by “true breeding?”



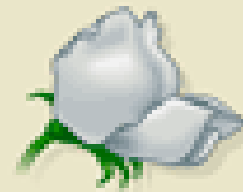
Mendel cross-fertilized his plants by hand. Why is it important to control which plants would serve as the parents?

P Generation
(true-breeding
parents)



Purple flowers

×



White flowers



F₁ Generation
(hybrids)

All plants have
purple flowers.



Self-fertilization in
F₁ plants

F₂ Generation



$\frac{3}{4}$ of plants
have purple flowers.



$\frac{1}{4}$ of plants
have white flowers.

For each **monohybrid cross**, Mendel cross-fertilized true-breeding plants that were different in just one character—in this case, flower color. He then allowed the hybrids (the F₁ generation) to self-fertilize.

Typical breeding experiment

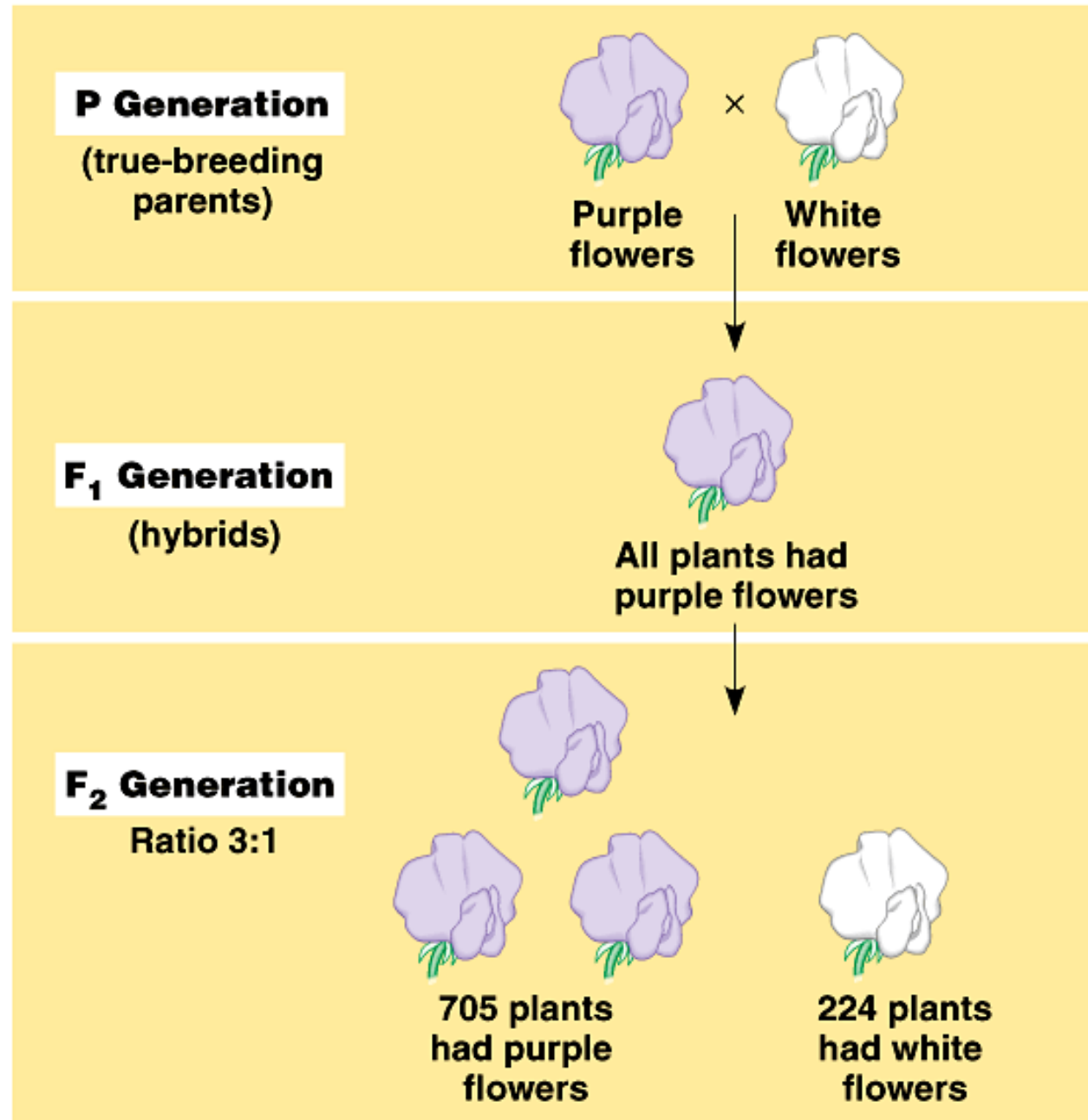
P generation (parental generation)

F₁ generation (first filial generation, the word filial from the Latin word for "son") are the hybrid offspring.





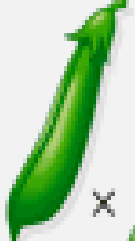
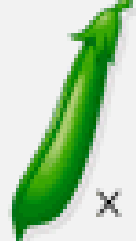












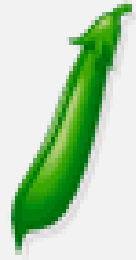
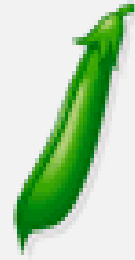
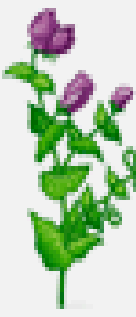
Allowing these F₁ hybrids to self-pollinate produces:















F₂ generation (second filial generation).

It is the analysis of this that lead to an understanding of genetic crosses.



Mendel studies seven characteristics in the garden pea

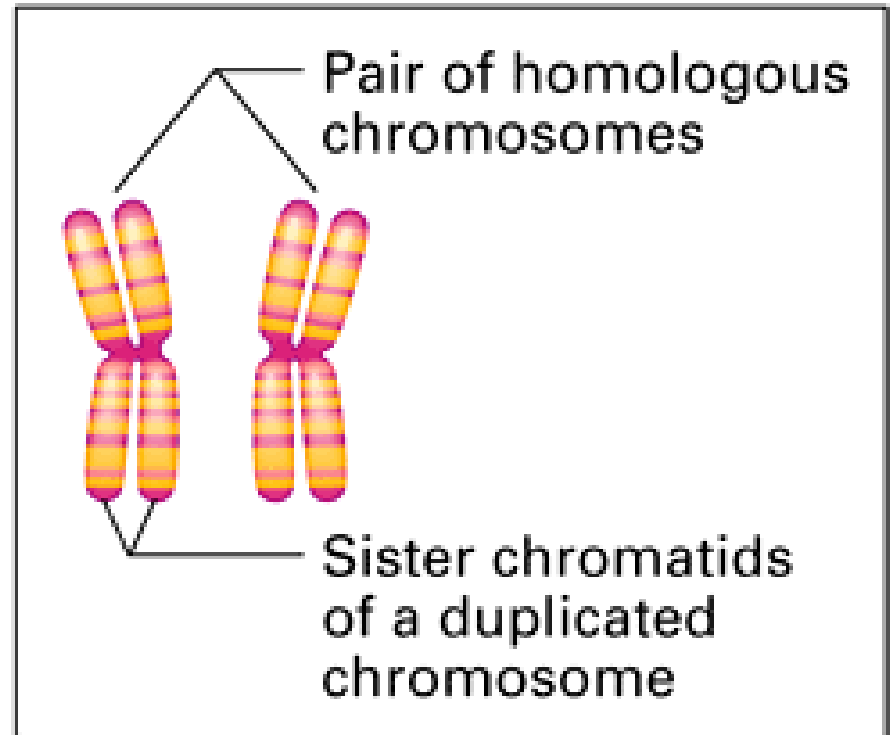
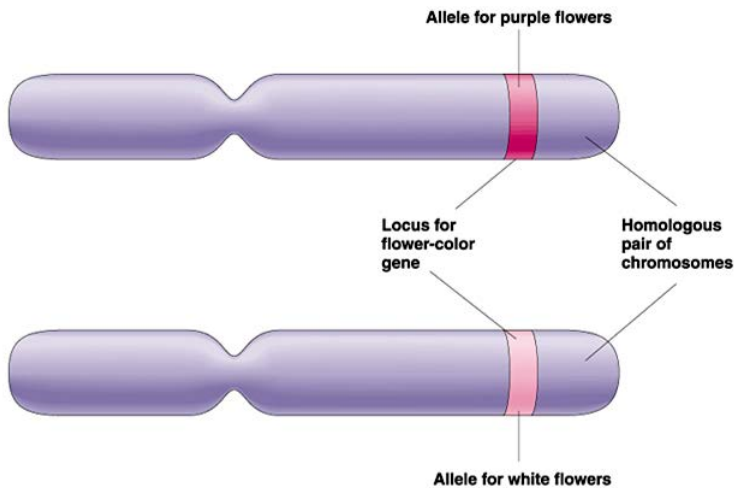
	Flower color	Flower position	Seed color	Seed shape	Pod shape	Pod color	Stem length
P	Purple 	Axial 	Yellow 	Round 	Inflated 	Green 	Tall 
	White 	Terminal 	Green 	Wrinkled 	Constricted 	Yellow 	Dwarf 
F ₁	Purple 	Axial 	Yellow 	Round 	Inflated 	Green 	Tall 

Character	Dominant Trait	×	Recessive Trait	F ₂ Generation Dominant:Recessive	Ratio
Flower color	 Purple	×	 White	705:224	3.15:1
Flower position	 Axial	×	 Terminal	651:207	3.14:1
Seed color	 Yellow	×	 Green	6022:2001	3.01:1
Seed shape	 Round	×	 Wrinkled	5474:1850	2.96:1
Pod shape	 Inflated	×	 Constricted	882:299	2.95:1
Pod color	 Green	×	 Yellow	428:152	2.82:1
Stem length	 Tall	×	 Dwarf	787:277	2.84:1

Statistics indicated a pattern.

Chromosomes

Homologous chromosome: one of a matching pair of chromosomes, one inherited from each parent.

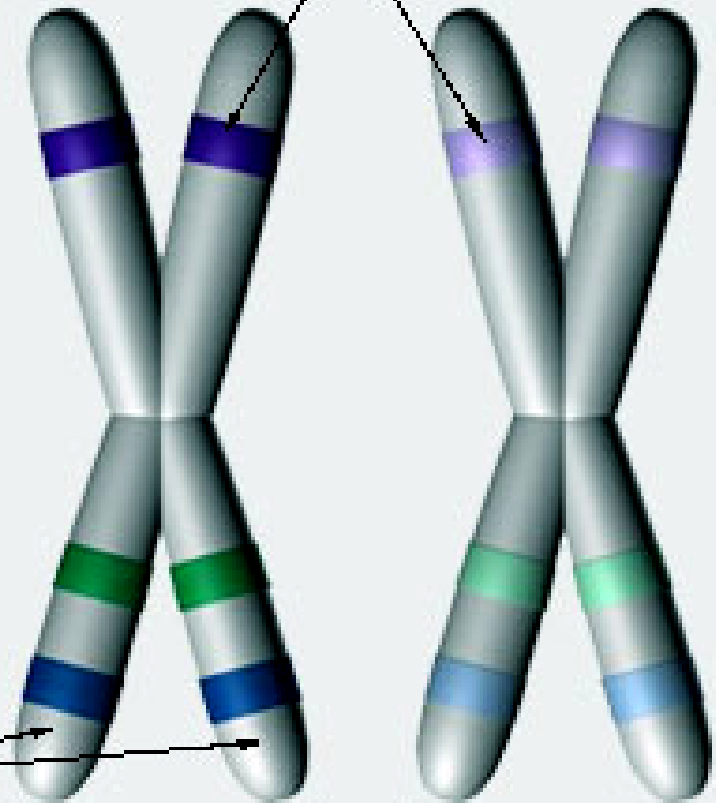
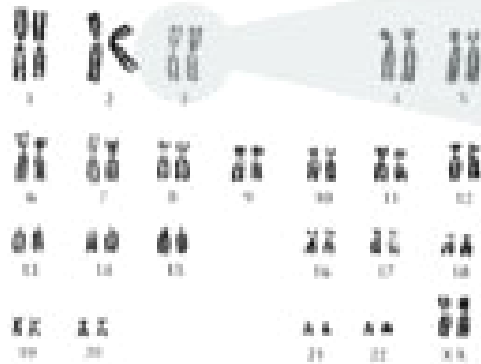


Sister chromatids are identical

Homologous Chromosomes

Homologous chromosomes contain DNA that codes for the same genes. In this example, both chromosomes have all the same genes in the same locations (represented with colored strips), but different 'versions' of those genes (represented by the different shades of each color).

Homologous regions code for the same gene.



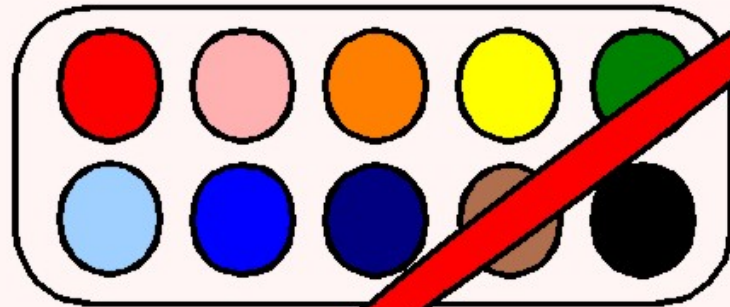
Sister chromatids are exact replicas...
but homologous chromosomes are not.

What genetic principles account for the transmission of such traits from parents to offspring?

The Blending Hypothesis of Inheritance

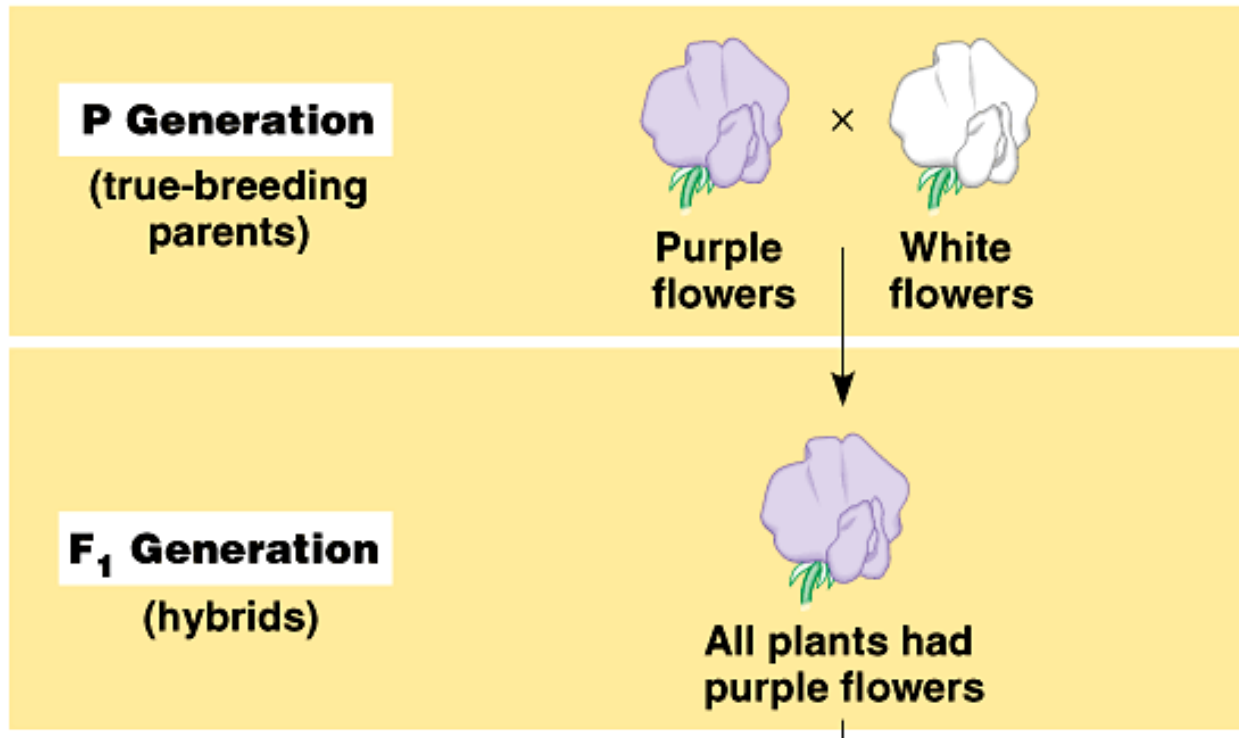
In the early 1800's the blending hypothesis was proposed. Genetic material contributed by the two parents mixes in a manner analogous to the way blue and yellow paints blend to make green.

What would happen if this was the case?



Law of Dominance

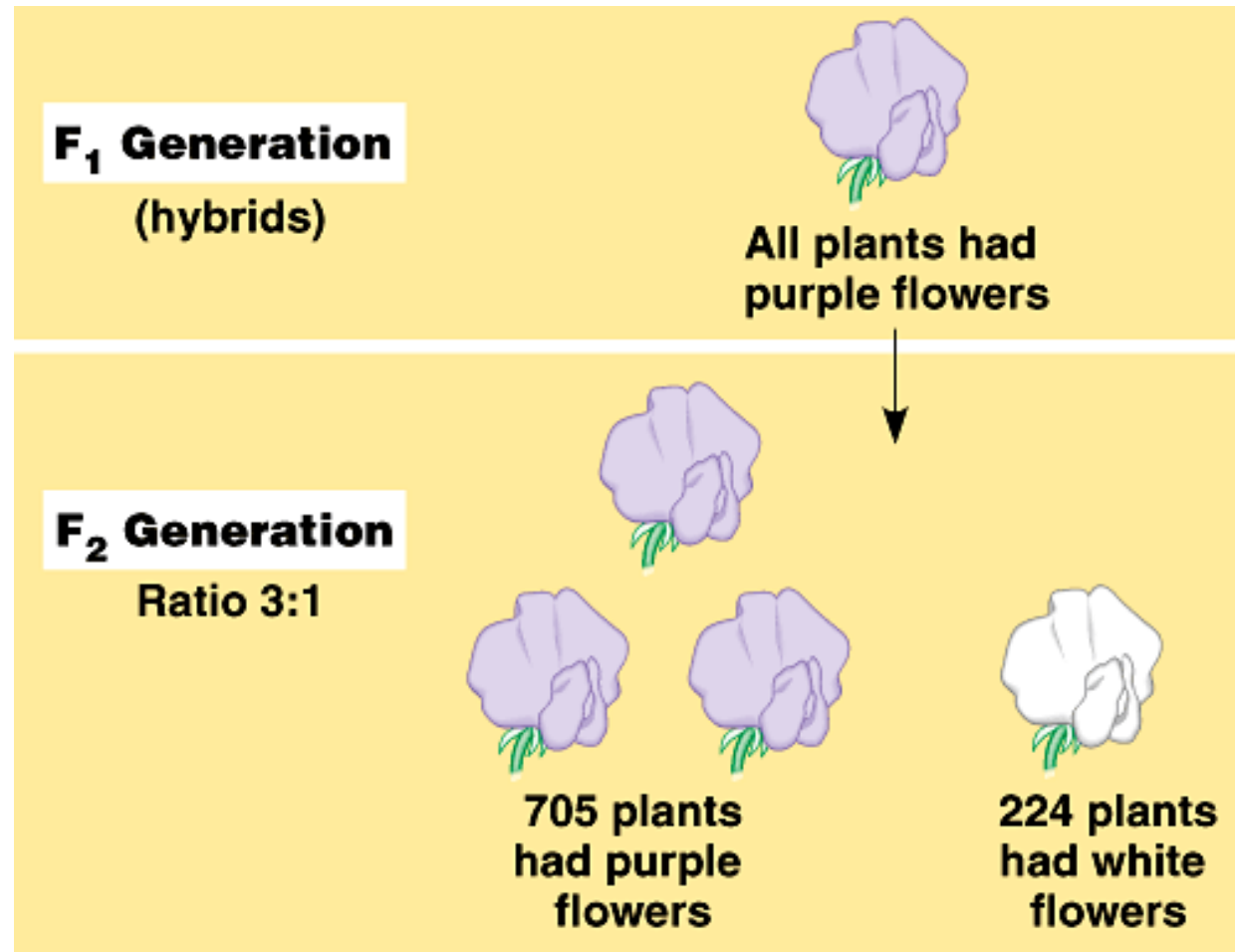
In the monohybrid cross (mating of two organisms that differ in only one character), one version disappeared.



What happens when the F₁'s are crossed?

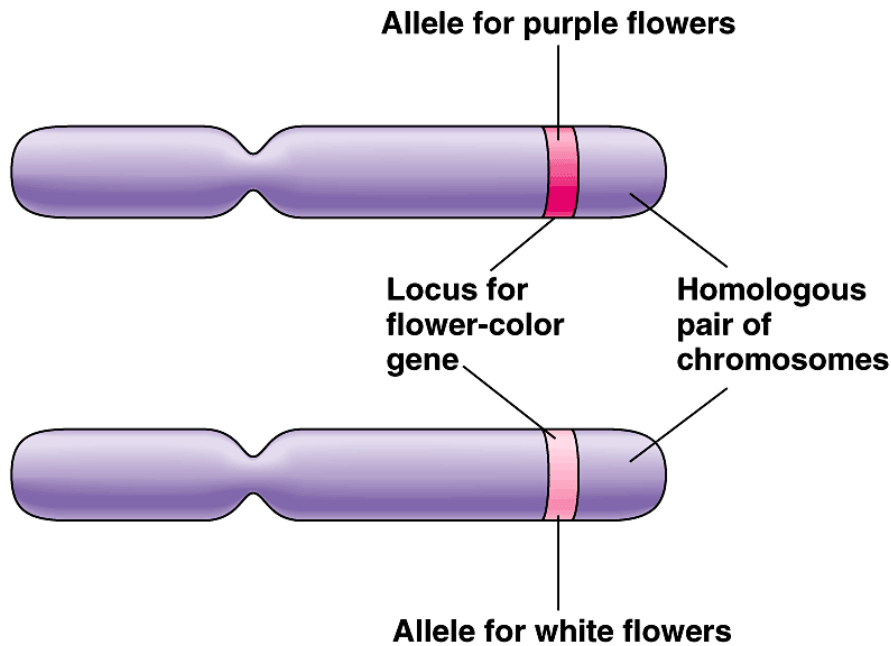
The F₁ crossed produced the F₂ generation and the lost trait appeared with predictable ratios.

This led to the formulation of the current model of inheritance.

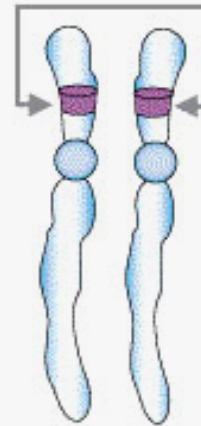


Alleles: alternative versions of a gene.

The gene for a particular inherited character resides at a specific locus (position) on homologous chromosome.

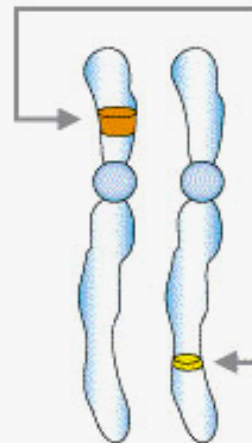


For each character, an organism inherits two alleles, one from each parent



Alleles

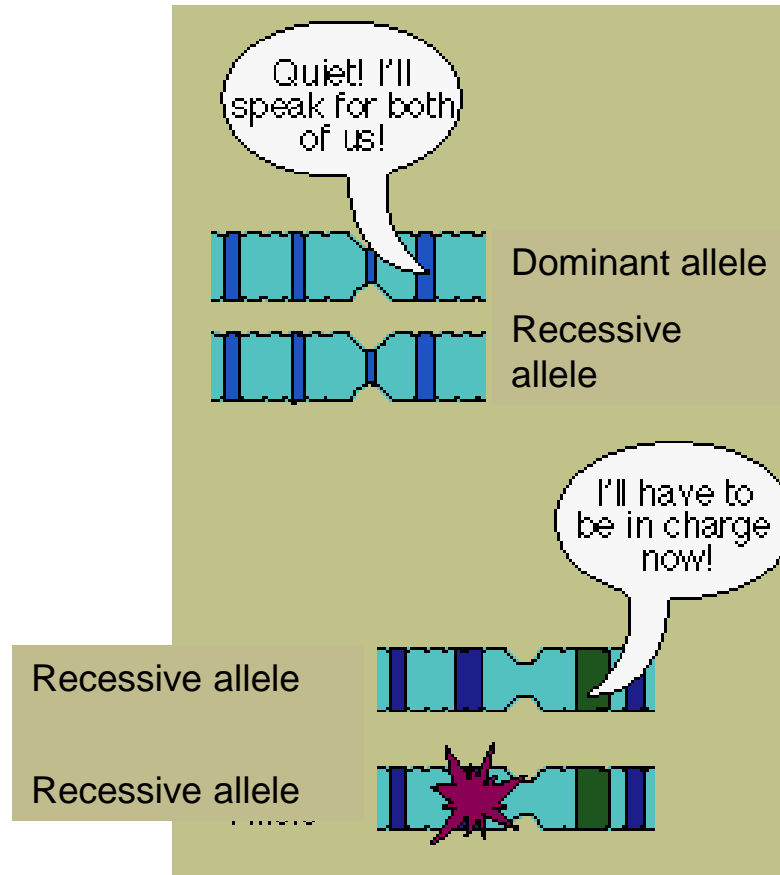
(code for same trait, same location on chromosome)



Genes, but *not* alleles

(code for different trait, different locations on chromosome)

How do alleles differ?

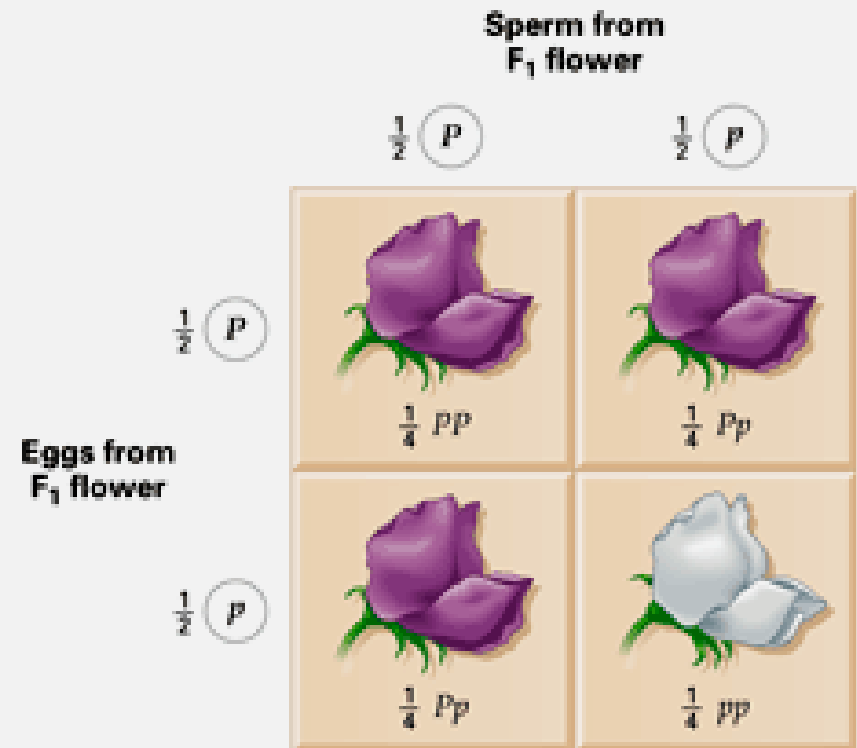
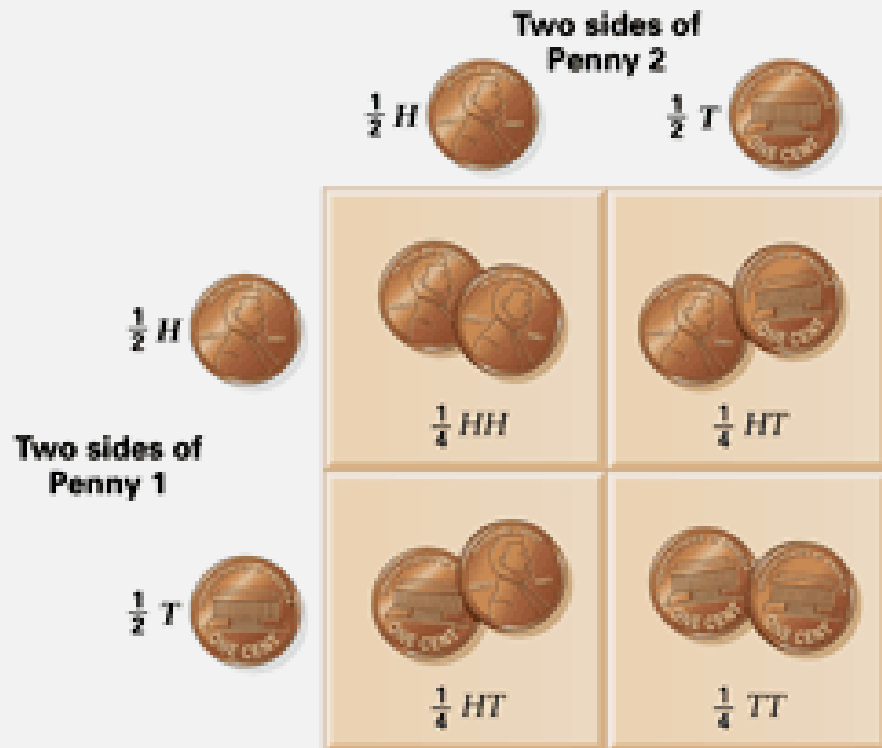


Dominant - a term applied to the trait (allele) that is expressed irregardless of the second allele.

Recessive - a term applied to a trait that is only expressed when the second allele is the same (e.g. short plants are homozygous for the recessive allele).

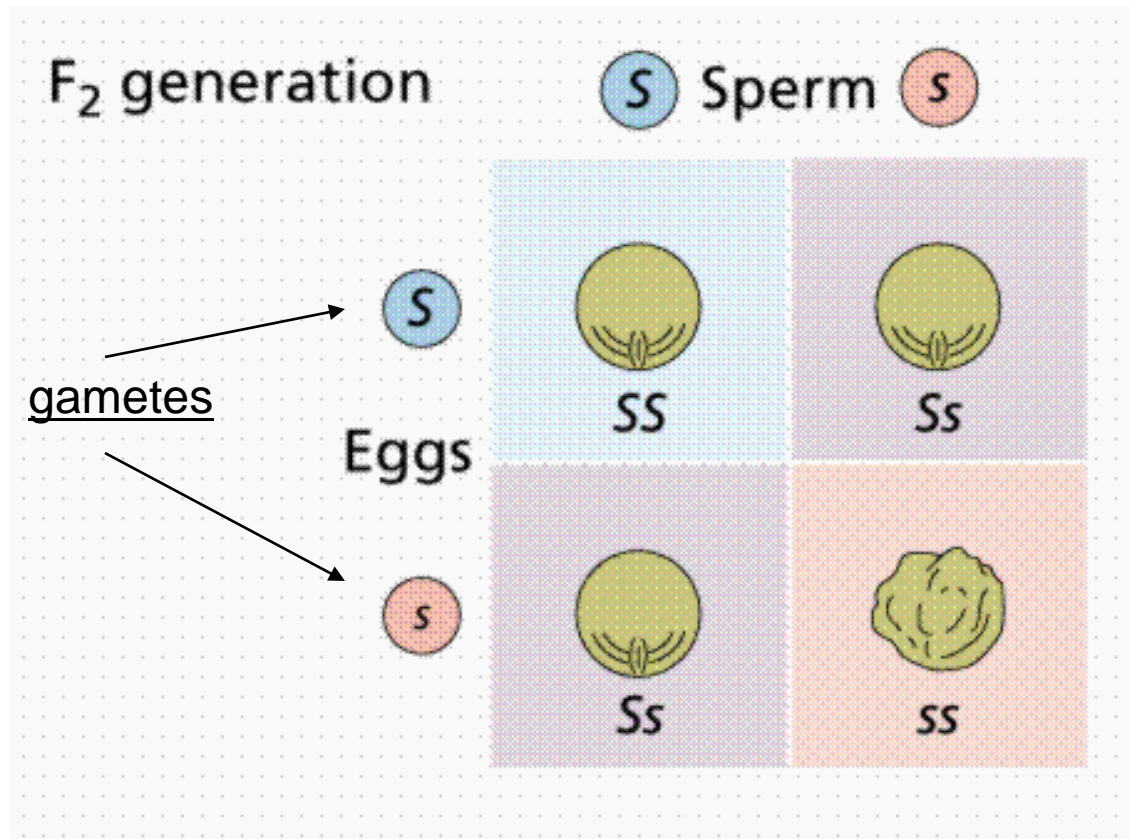
Probability and Punnett Squares

Punnett square: diagram showing the probabilities of the possible outcomes of a genetic cross



Punnett squares - probability diagram illustrating the possible offspring of a mating.

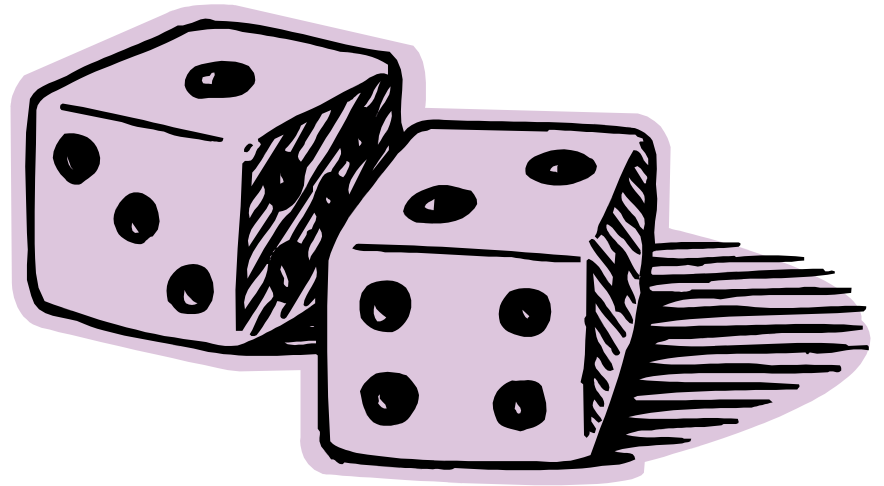
$Ss \times Ss$



Probability & Genetics

the number of times an event is expected to happen divided by the number of opportunities for event to happen

The Rules Of Probability

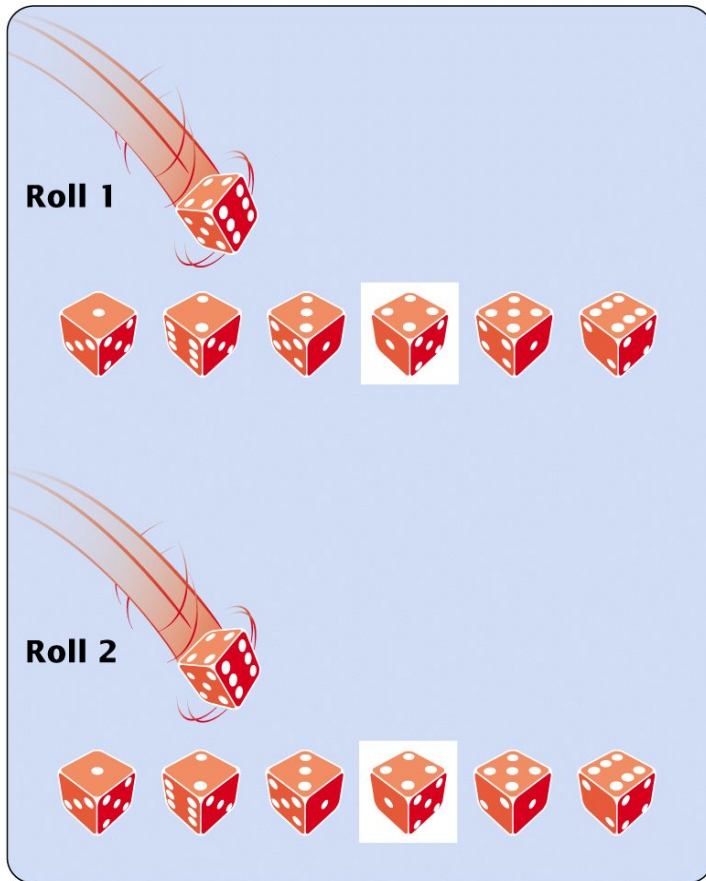


The Product Rule

probability of two events occurring
simultaneously is product of their
probabilities

Multiplication Rule

(a) The multiplication rule



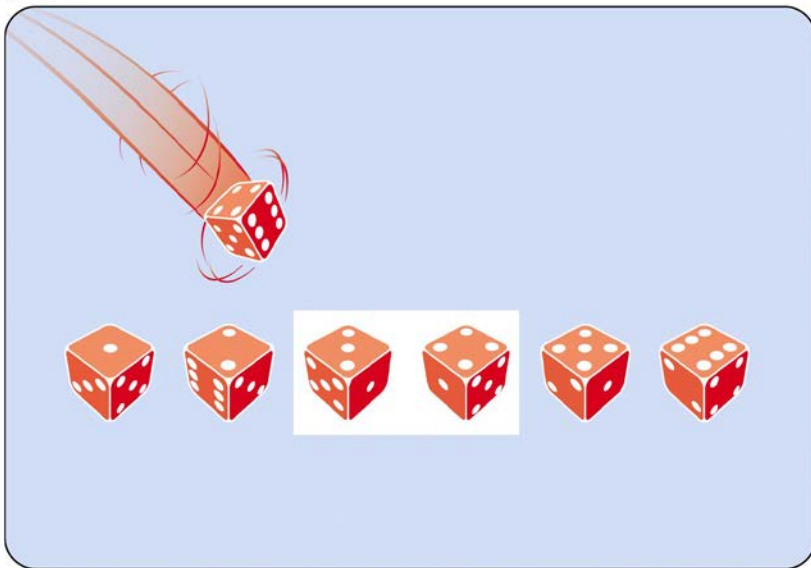
- Roll 1
 - $1/6$ chance of “4”
 - on any roll
- Roll 2
 - $1/6$ chance of “4”
- Two sequential rolls
 - $1/6 \times 1/6 = 1/36$

The Sum Rule

probability of either one of two mutually exclusive events is sum of their individual probabilities

Addition Rule

(b) The addition rule



- $P(3) = 1/6$
- $P(4) = 1/6$
- $P(3 \text{ or } 4) =$
 - $1/6 + 1/6 =$
 - $2/6 = 1/3$

Probability

expressed as ratio
(decimal or fraction)



Genetic Applications

- Multiplication rule
 - Probability of getting each allele
 - Probability of getting desired combination
- Addition rule
 - Overall phenotypic ratio



Genotype versus phenotype.

Genotype

1 { PP
(homozygous)

2 { Pp
(heterozygous)
 Pp
(heterozygous)

1 { pp
(homozygous)

Ratio 1:2:1



Phenotype

Purple

Purple

Purple

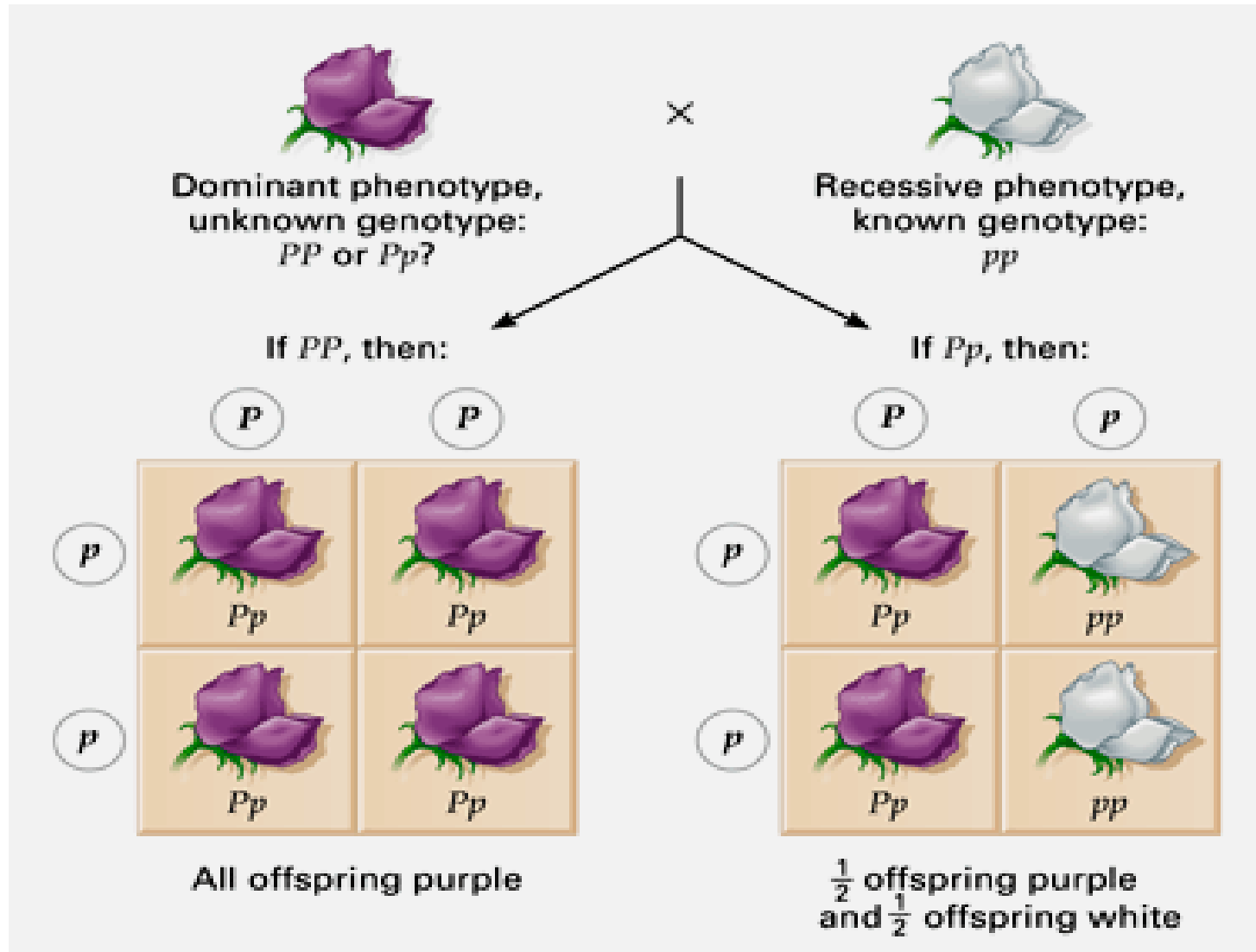
White

Ratio 3:1

How does a genotype ratio differ from the phenotype ratio?

Testcross

A testcross is designed to reveal whether an organism that displays the dominant phenotype is homozygous or heterozygous.



- The offspring of a **testcross** can reveal the genotype of a parent.

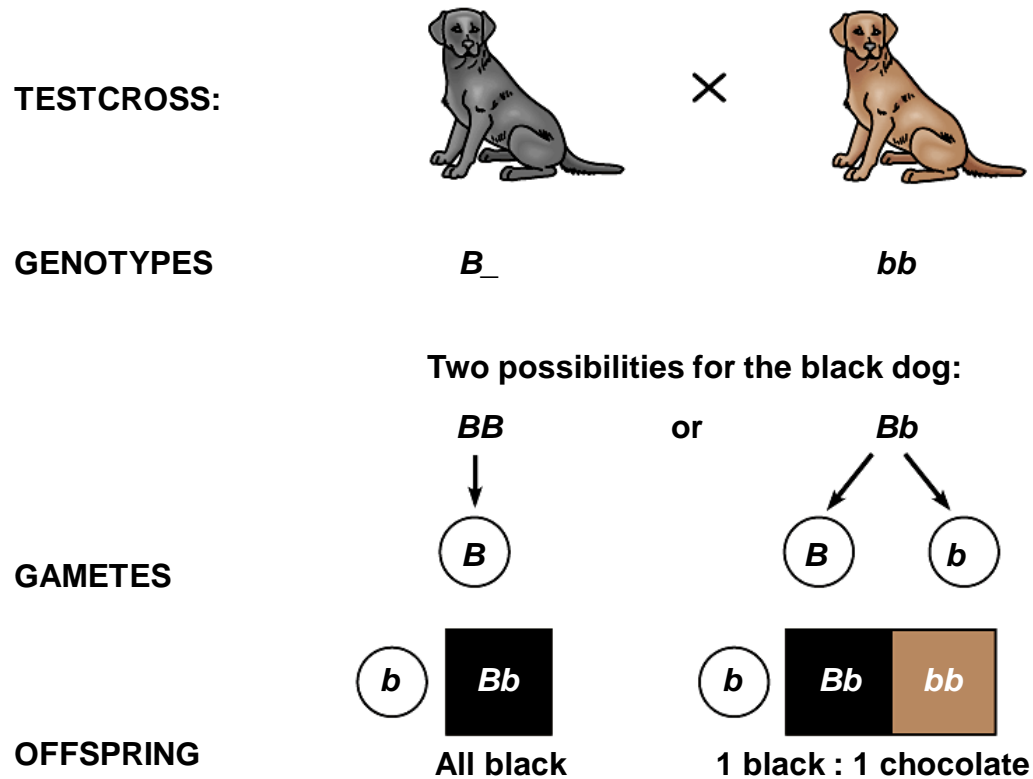


Figure 9.6

Law of Independent Assortment

- **During gamete formation, genes for different traits separate independently into gametes**
- Why? random alignment of homologues at Meiosis I
- A sperm or egg carries only one allele of each pair

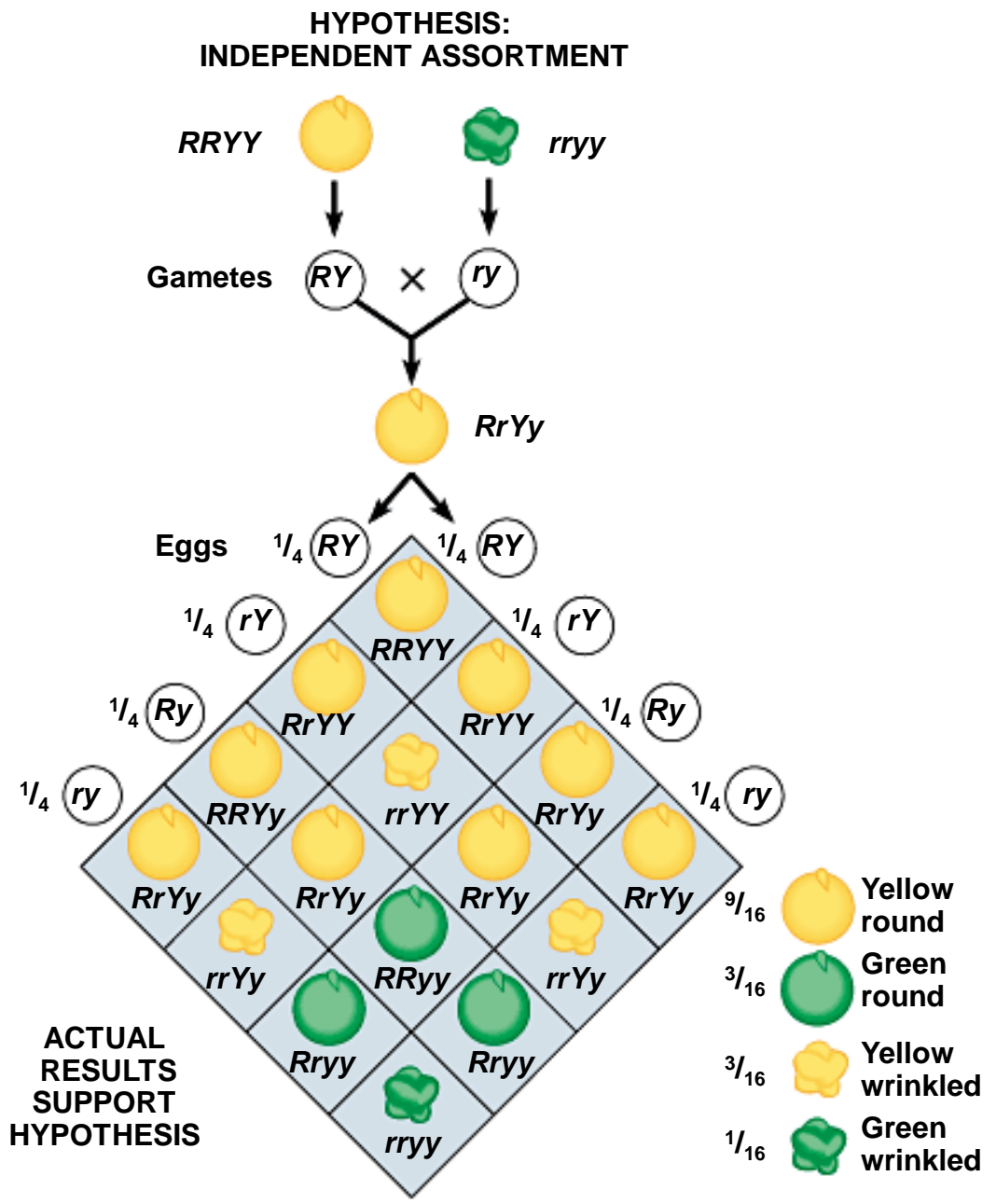
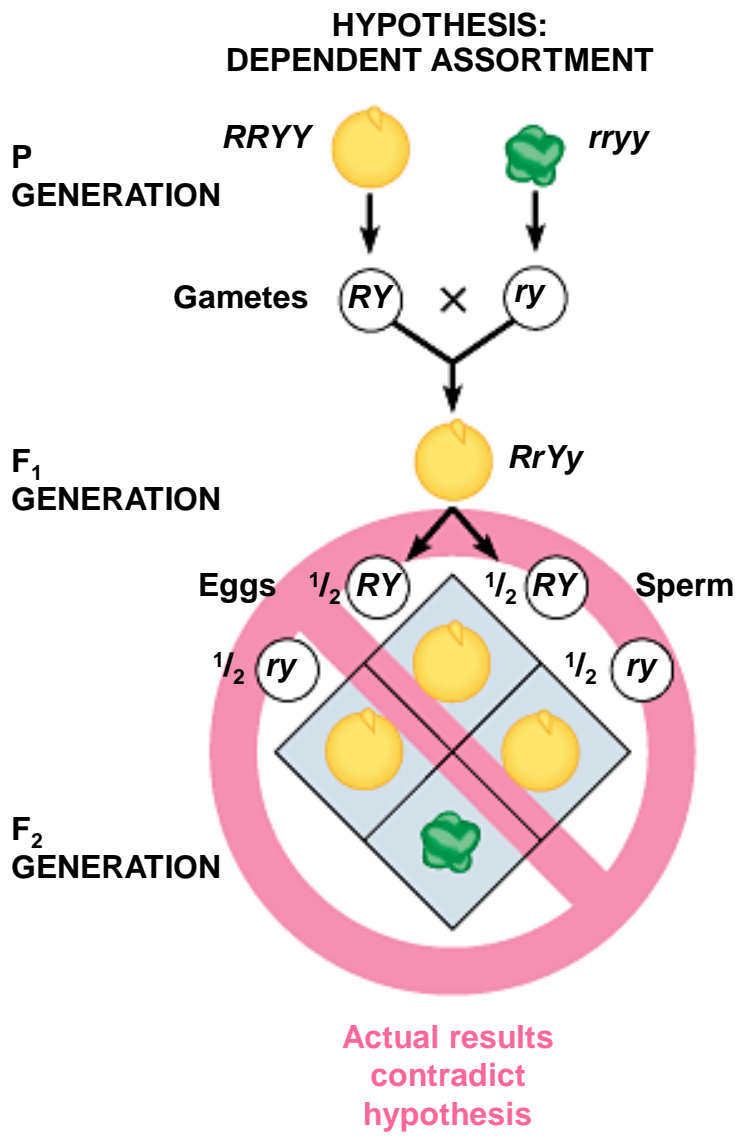


Figure 9.5A

- Independent assortment of two genes in the Labrador retriever

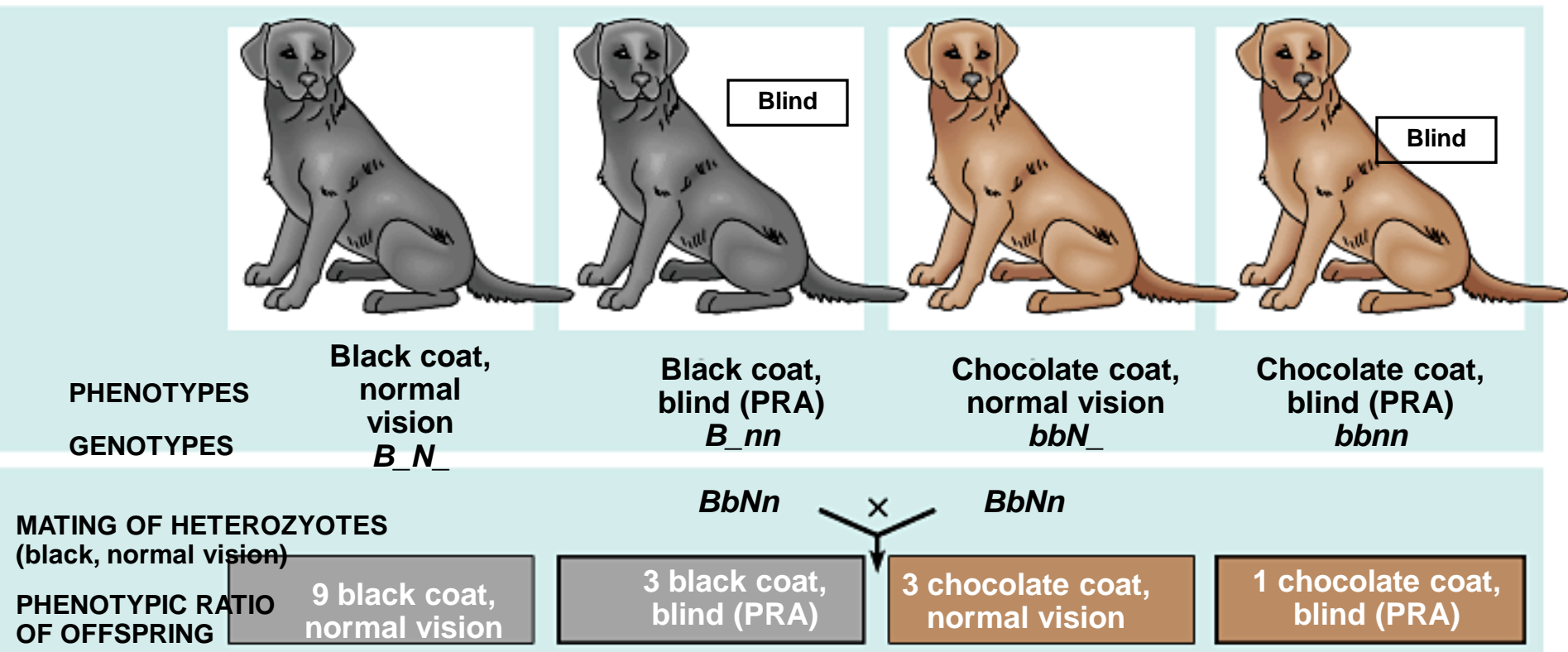


Figure 9.5B

Chromosome behavior accounts for Mendel's principles

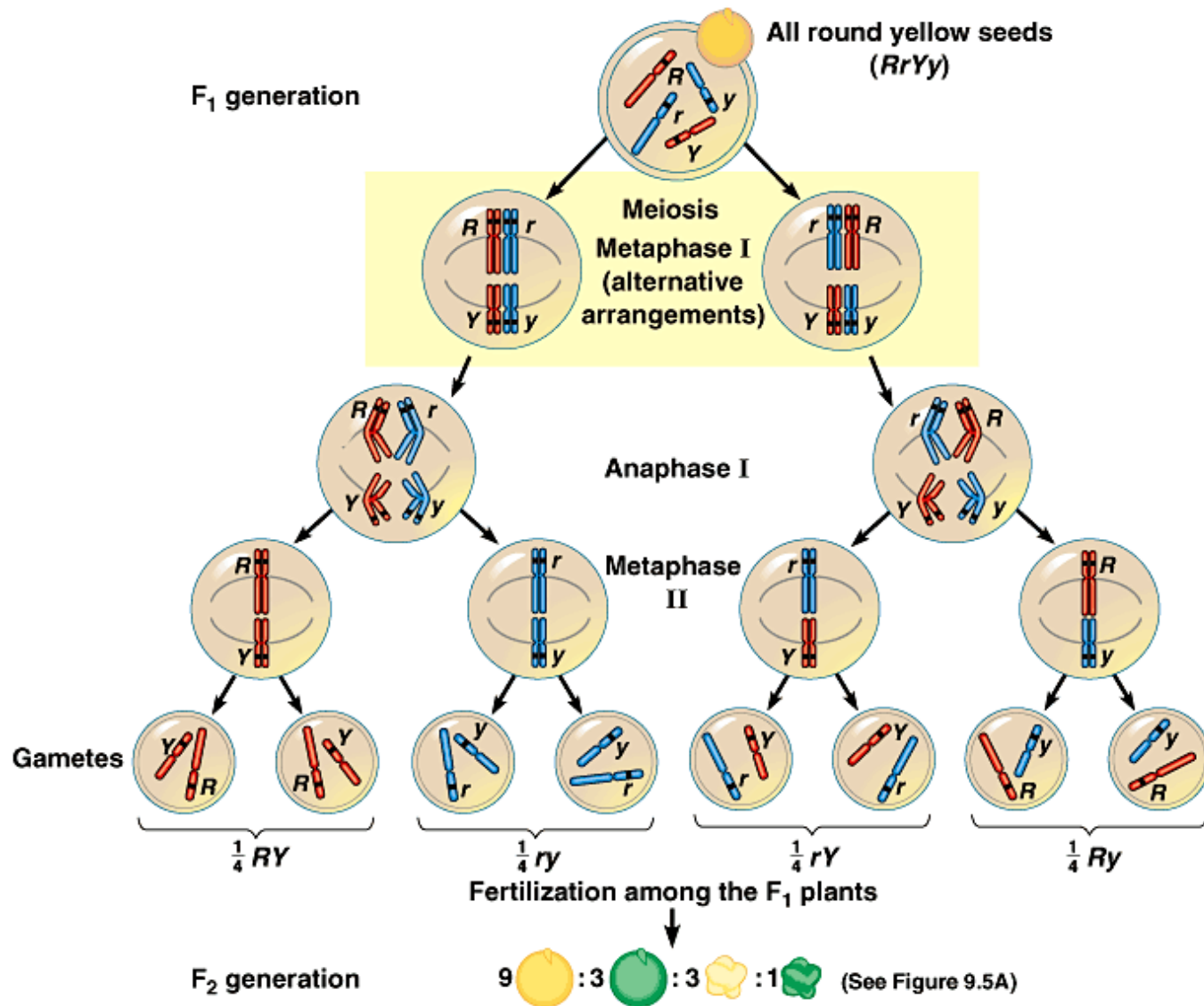
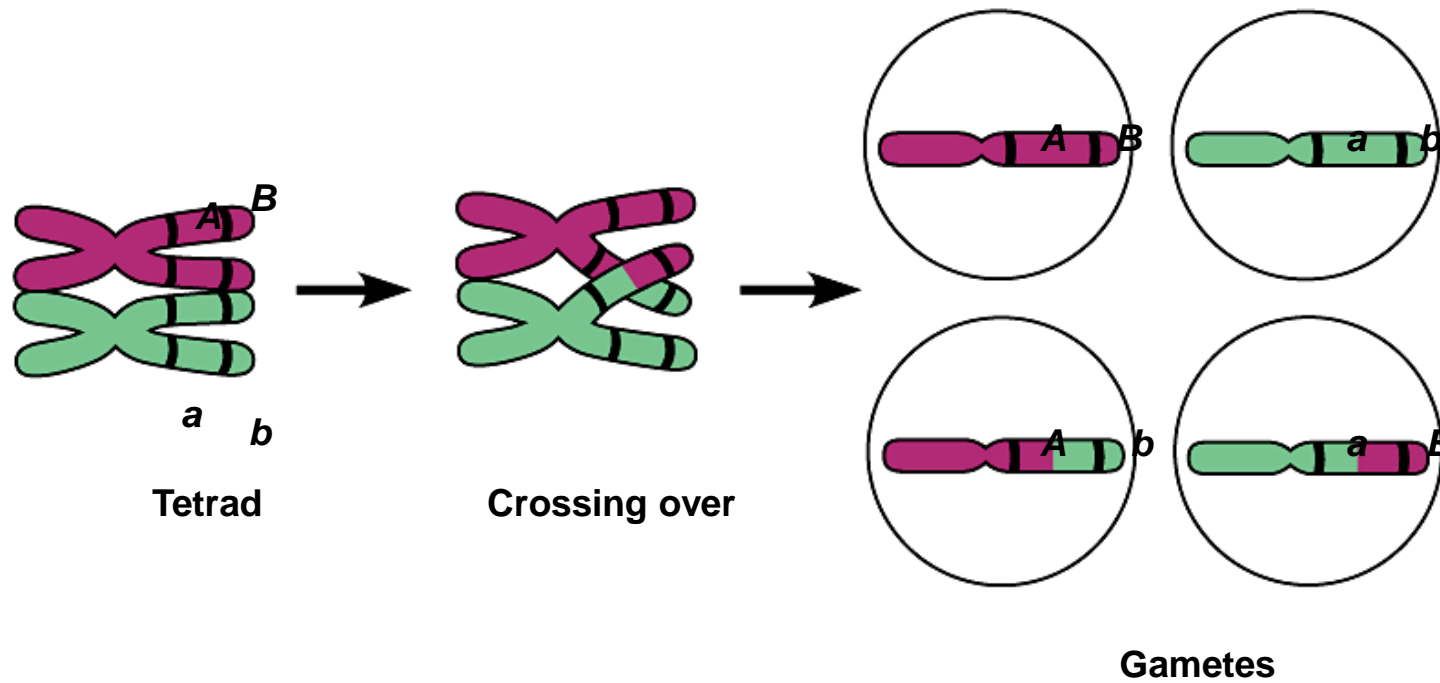


Figure 9.17



- Genes on the same chromosome tend to be inherited together = **linked genes**
- Crossing over produces gametes with recombinant chromosomes

Geneticists use crossover data to map genes

- Crossing over is more likely to occur between genes that are farther apart
 - Recombination frequencies

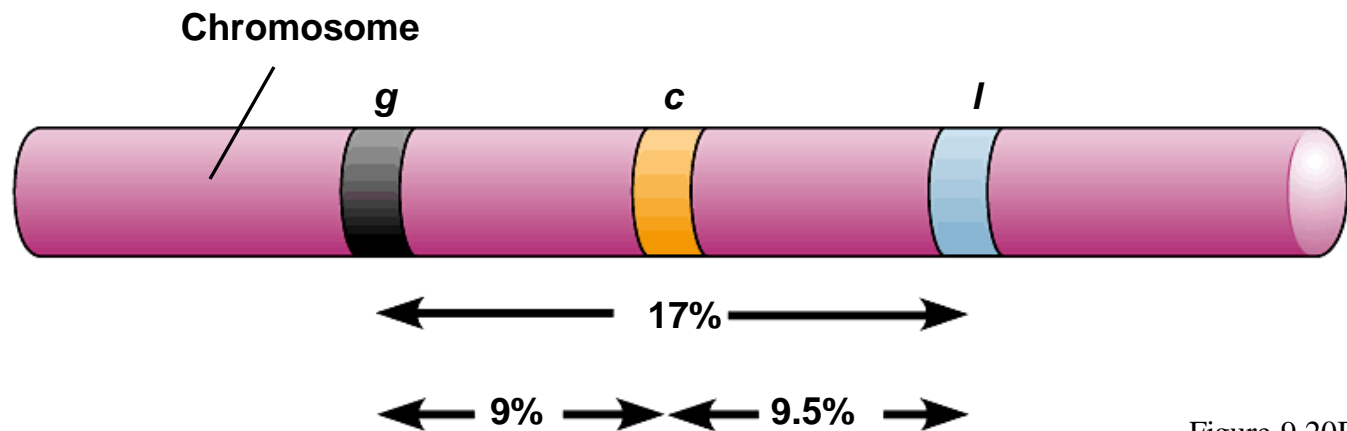
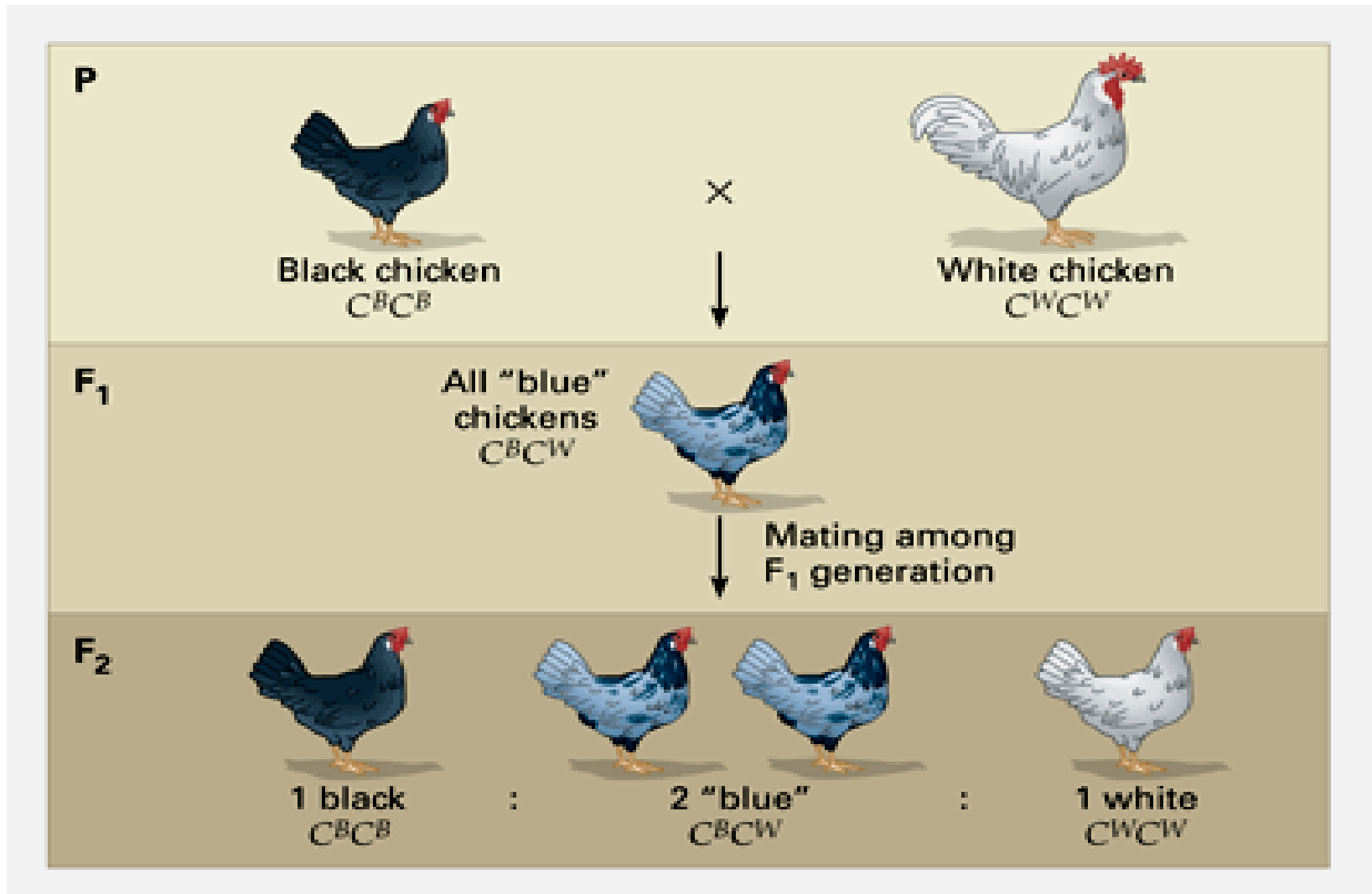


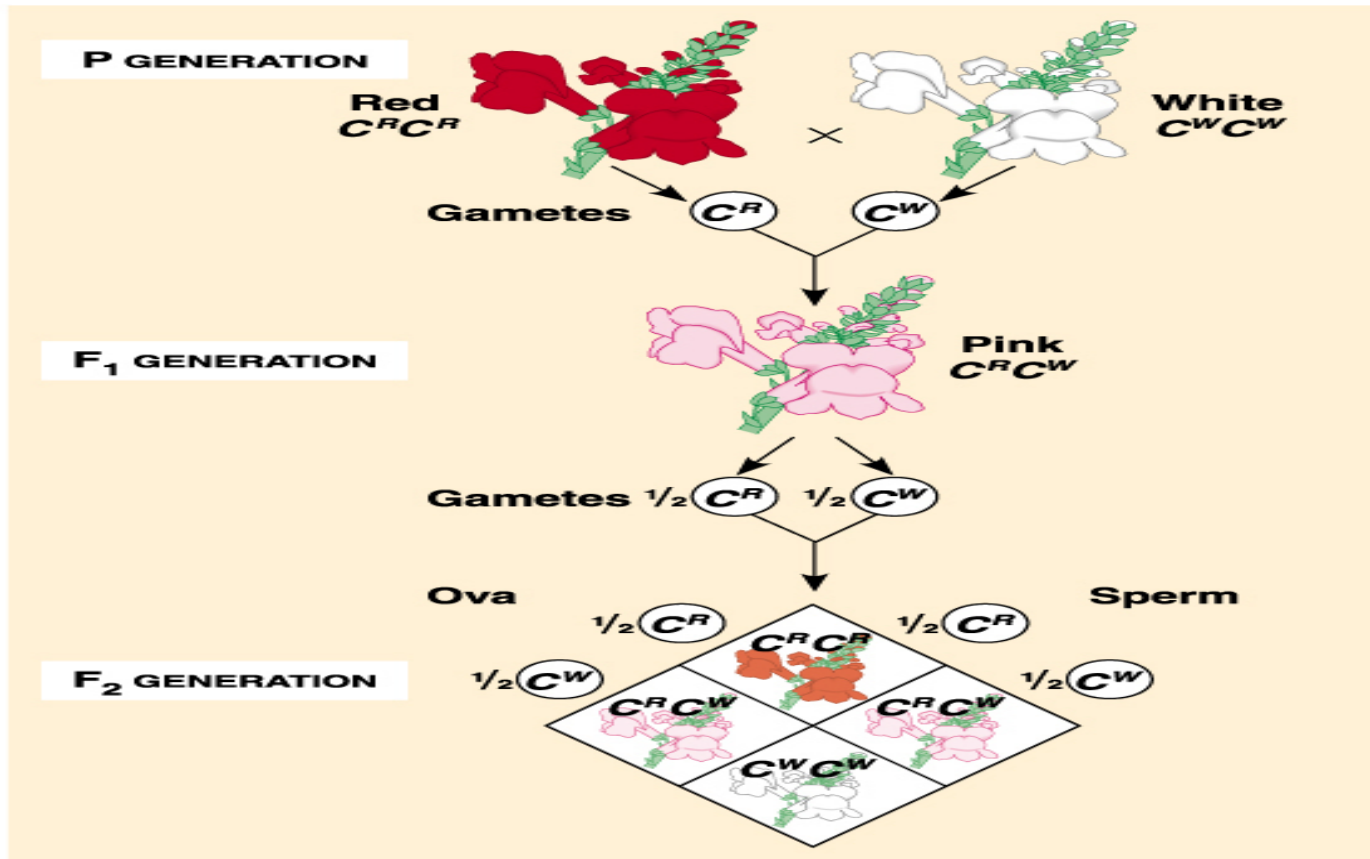
Figure 9.20B

Variation in Patterns of Inheritance

Intermediate Inheritance (blending): inheritance in which heterozygotes have a phenotype intermediate between the phenotypes of the two homozygotes



Incomplete Dominance

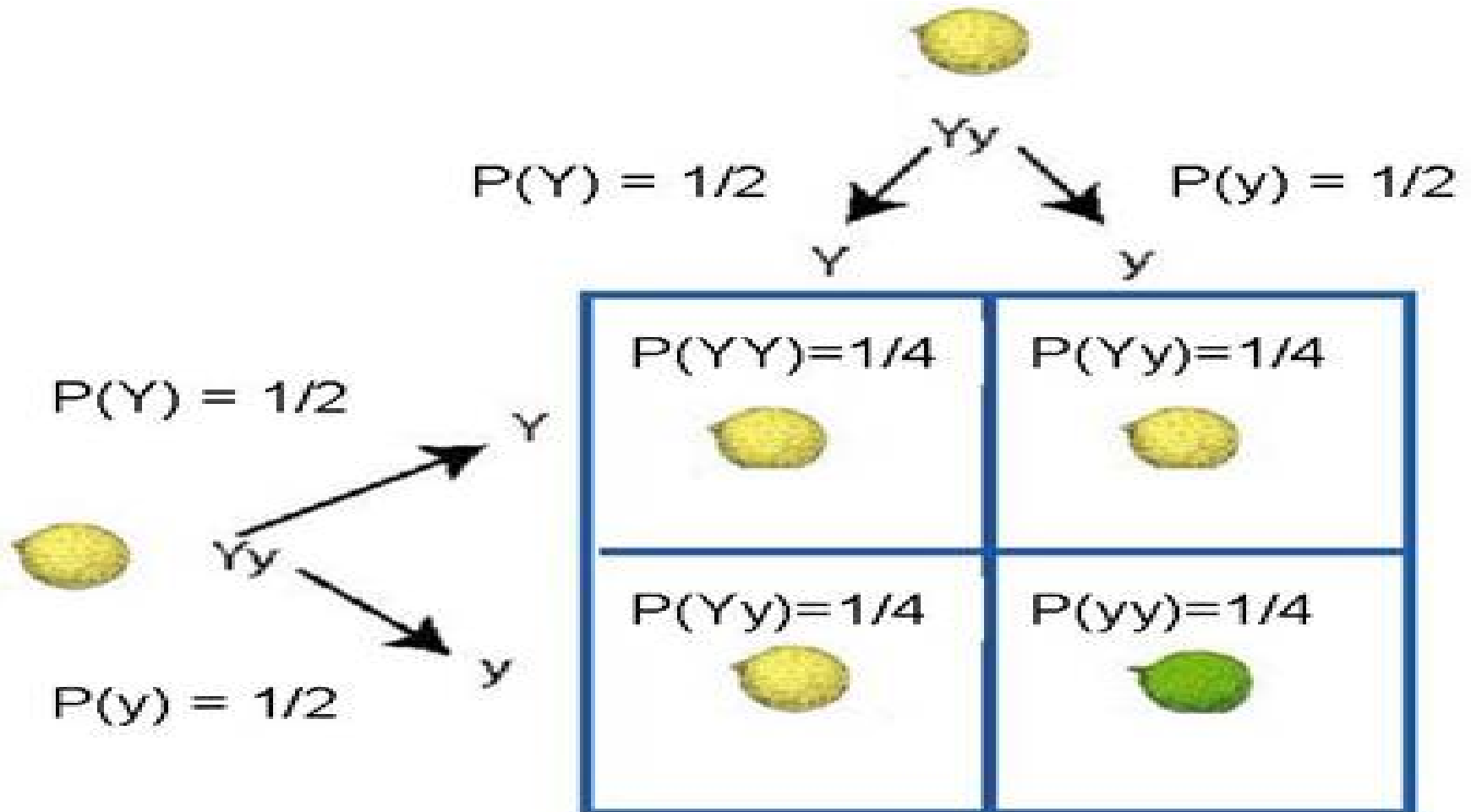


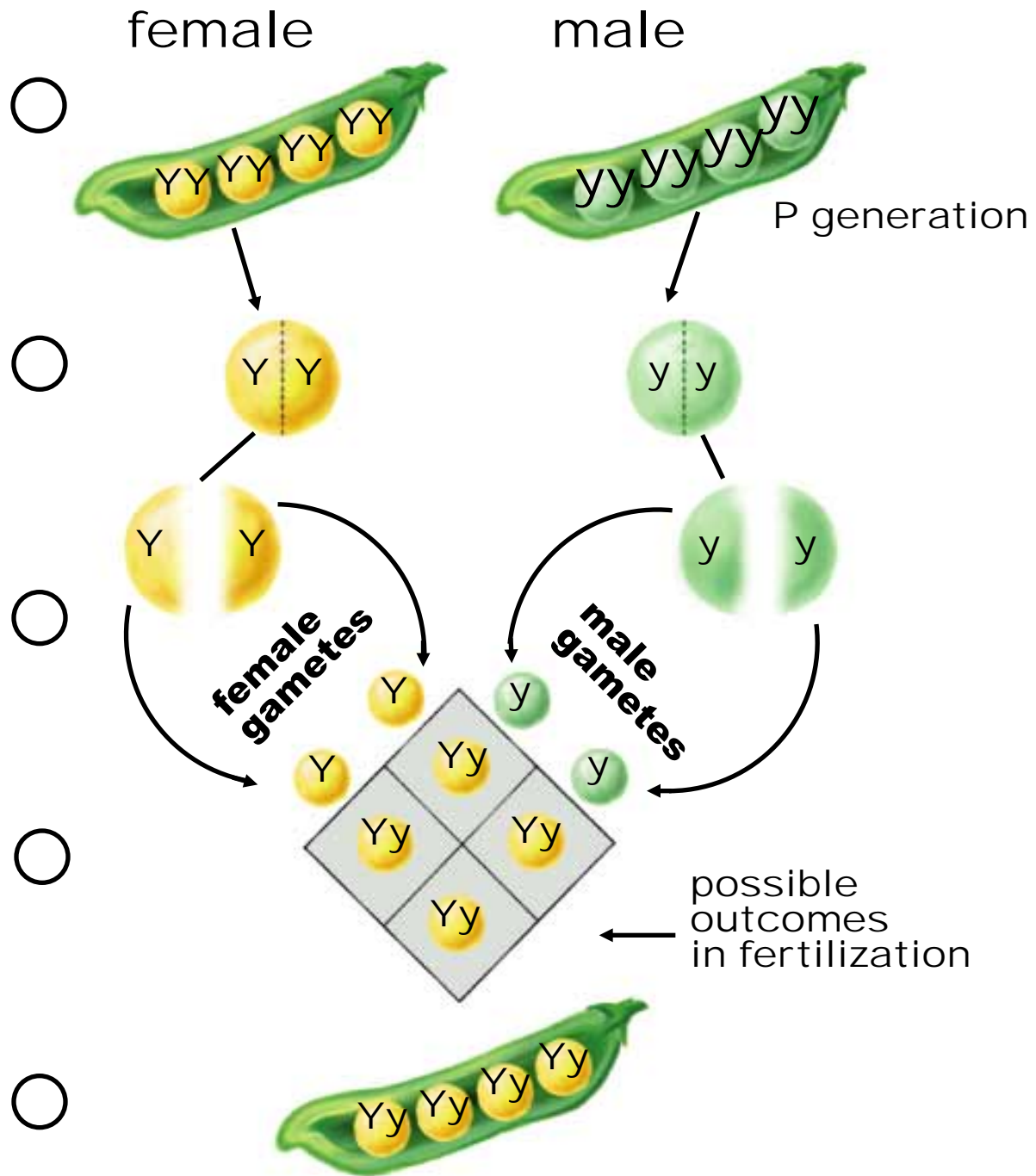
Codominance

- Two alleles affect the phenotype in separate and distinguishable ways.
- Neither allele can mask the other and both are expressed in the offspring and not in an “intermediate” form.
- Example: red flowers that are crossed with white flowers that yield red and white flowers.



Monohybrid Cross





How Does it Work?

Father's Genes

M
O
T
H
E
R
S
G
E
N
E
S

	A	a
A	AA	Aa
a	aA	aa

















T


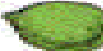


T

t

t

Dihybrid Cross

		♂ gametes			
		$\begin{matrix} RY \\ \frac{1}{4} \end{matrix}$	$\begin{matrix} Ry \\ \frac{1}{4} \end{matrix}$	$\begin{matrix} ry \\ \frac{1}{4} \end{matrix}$	$\begin{matrix} rY \\ \frac{1}{4} \end{matrix}$
♀ gametes	$\begin{matrix} RY \\ \frac{1}{4} \end{matrix}$	$\begin{matrix} RRYY \\ \frac{1}{16} \end{matrix}$ 	$\begin{matrix} RR Yy \\ \frac{1}{16} \end{matrix}$ 	$\begin{matrix} Rr Yy \\ \frac{1}{16} \end{matrix}$ 	$\begin{matrix} Rr YY \\ \frac{1}{16} \end{matrix}$ 
	$\begin{matrix} Ry \\ \frac{1}{4} \end{matrix}$	$\begin{matrix} RR Yy \\ \frac{1}{16} \end{matrix}$ 	$\begin{matrix} RR yy \\ \frac{1}{16} \end{matrix}$ 	$\begin{matrix} Rr yy \\ \frac{1}{16} \end{matrix}$ 	$\begin{matrix} Rr Yy \\ \frac{1}{16} \end{matrix}$ 
	$\begin{matrix} ry \\ \frac{1}{4} \end{matrix}$	$\begin{matrix} Rr Yy \\ \frac{1}{16} \end{matrix}$ 	$\begin{matrix} Rr yy \\ \frac{1}{16} \end{matrix}$ 	$\begin{matrix} rr yy \\ \frac{1}{16} \end{matrix}$ 	$\begin{matrix} rr Yy \\ \frac{1}{16} \end{matrix}$ 
	$\begin{matrix} rY \\ \frac{1}{4} \end{matrix}$	$\begin{matrix} Rr YY \\ \frac{1}{16} \end{matrix}$ 	$\begin{matrix} Rr Yy \\ \frac{1}{16} \end{matrix}$ 	$\begin{matrix} rr Yy \\ \frac{1}{16} \end{matrix}$ 	$\begin{matrix} rr YY \\ \frac{1}{16} \end{matrix}$ 

9  : 3  : 3  : 1 

 Round, yellow

 Wrinkled, yellow

 Round, green

 Wrinkled, green

Step 1: Make a Key....

- G = yellow
- g = green
- W = round
- w = wrinkled

Step 2: Give the genotypes of the parents.

..

- *Homozygous **round** green X **wrinkled**
homozygous yellow plant*
- **P1 = WWgg X wwGG**

Step 3:
Determine the Gametes . . .

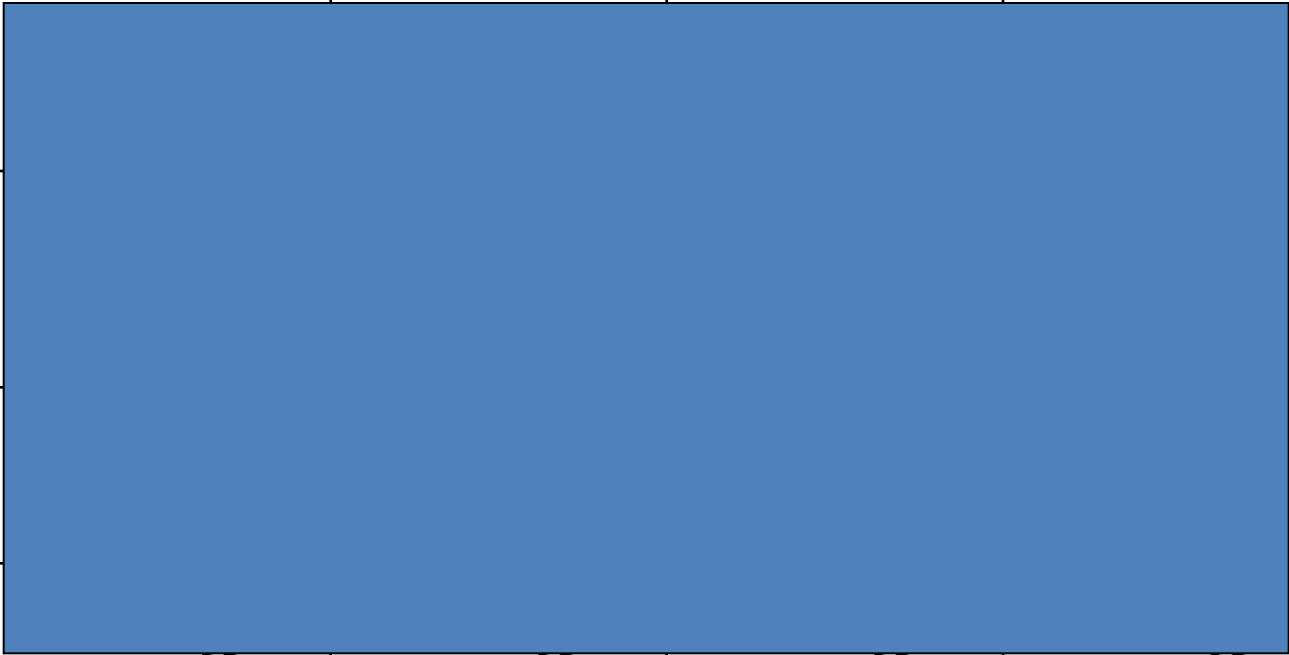
- **WWgg X wwGG**

WWgg = Wg, Wg, Wg, Wg

wwGG = wG, wG, wG, wG

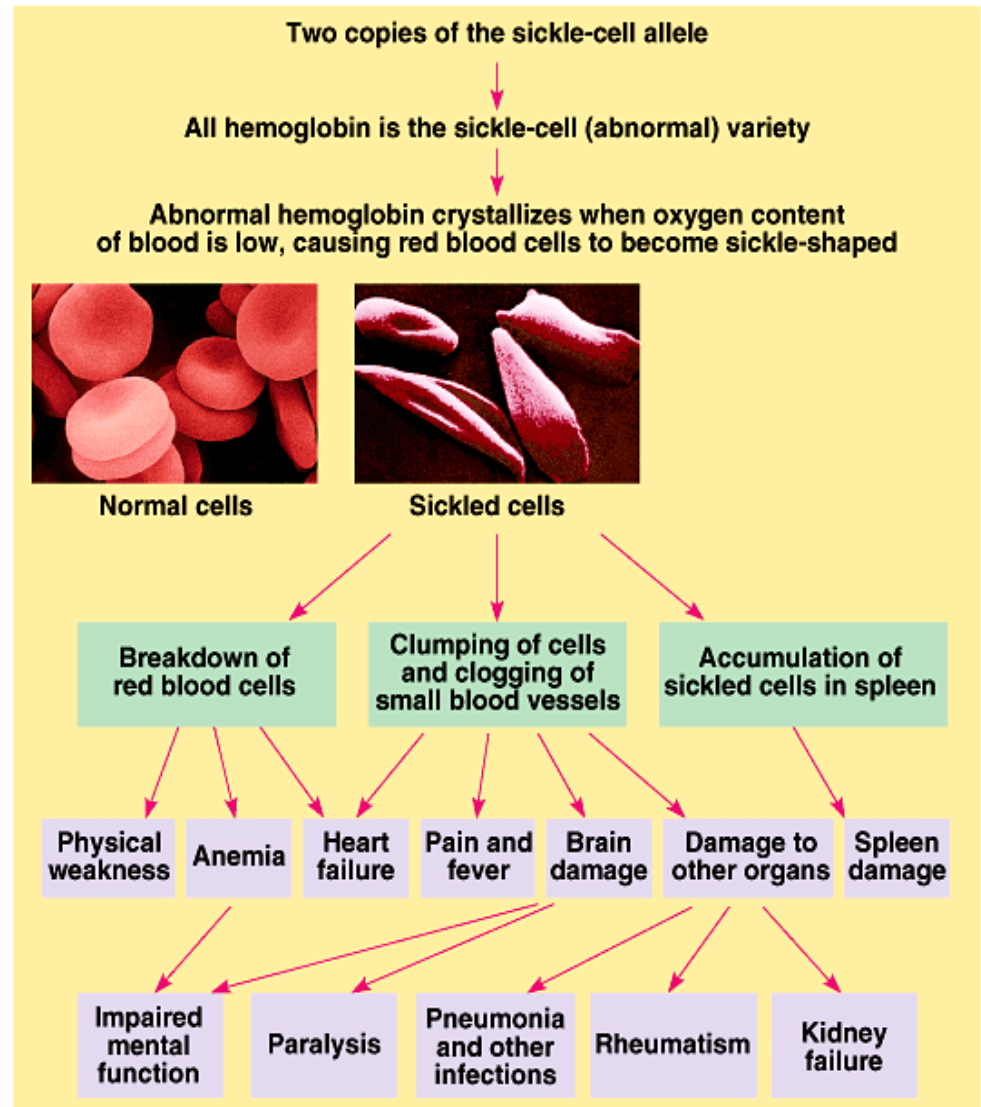
HINT: Foil to get the gametes

Step 4:
Fill in the Punnett square . . .

	Wg	Wg	Wg	Wg
wG				
wG				
wG				
wG				

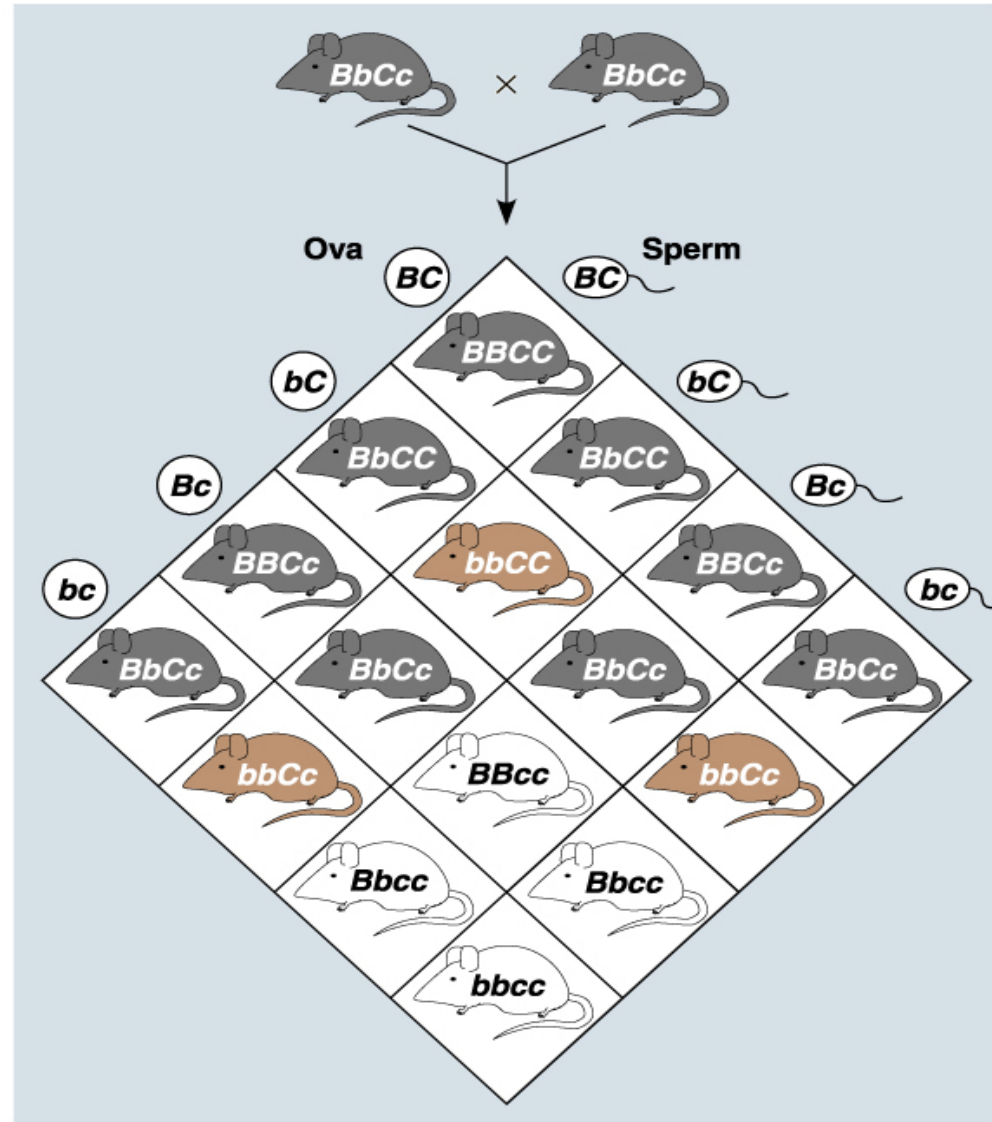
Pleiotropy

- **Most genes have multiple phenotypic effects. The ability of a gene to affect an organism in many ways is called pleiotropy.**



Epistasis

- Epistasis occurs when a gene at one locus alters or influences the expression of a gene at a second loci. In this example, C is for color and the dominate allele must be present for pigment (color) to be expressed.



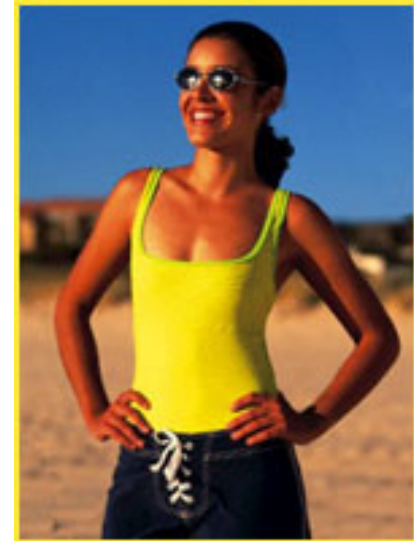
The Importance of the Environment

The environmental influences the expression of the genotype so the phenotype is altered.

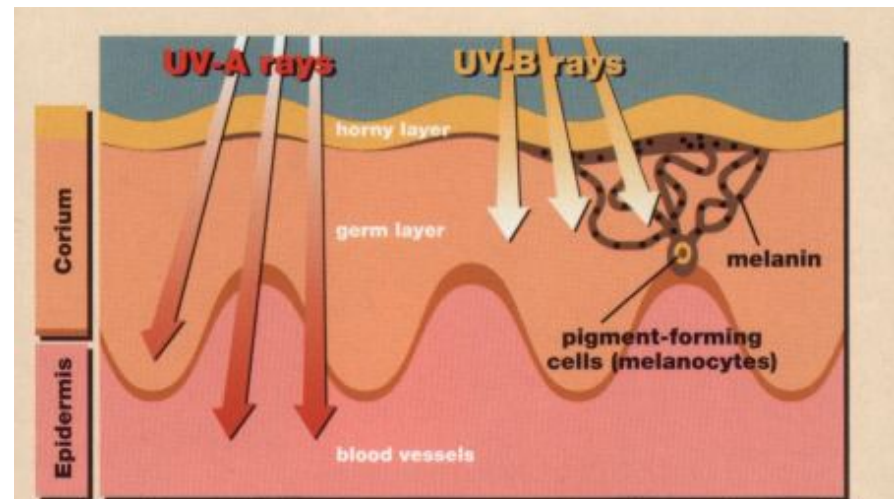
Hydrangea flowers of the same genetic variety range in color from blue-violet to pink, depending on the acidity of the soil.



Benjamin
Cummings



Multifactorial; many factors, both genetic and environmental, collectively influence phenotype in examples such as skin tanning



Chromosome Theory of Inheritance

Improved microscopy techniques, understand cell processes and genetic studies converged during the late 1800's and early 1900's.

It was discovered that Mendelian inheritance has its physical basis in the behavior of chromosomes during sexual life cycles.



Walter S. Sutton

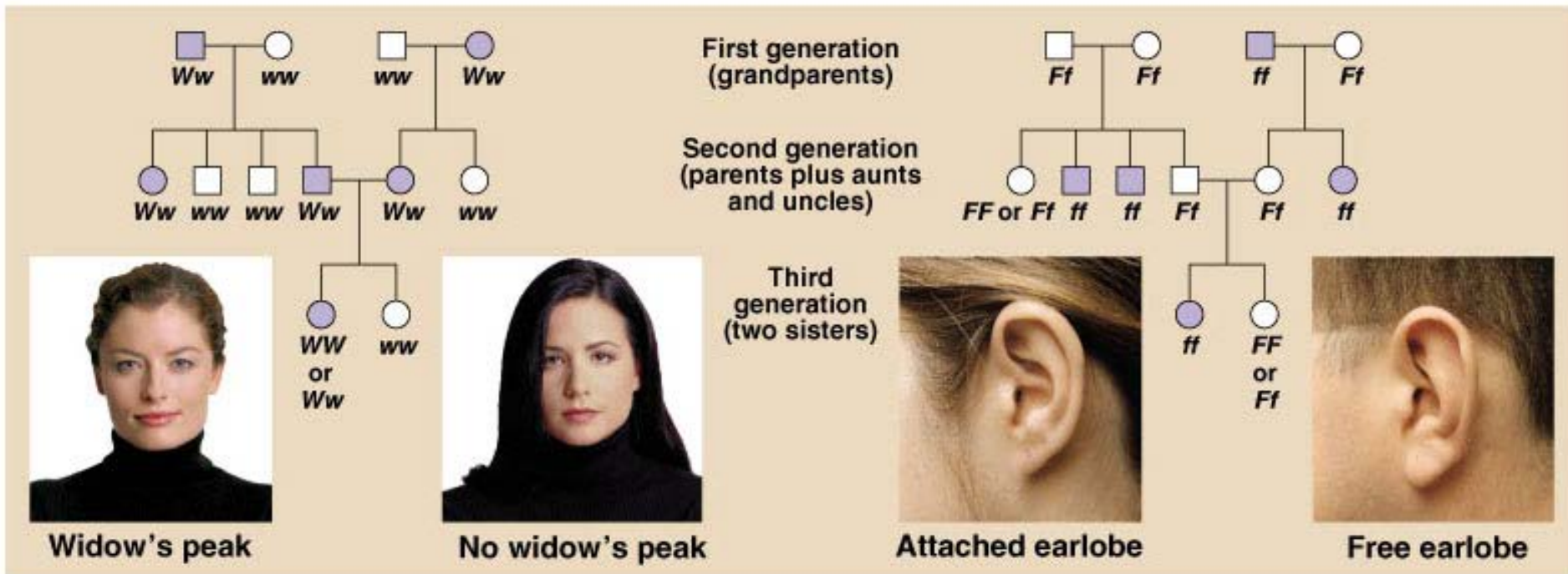


Theodor Boveri



Hugo de Vries

Pedigree analysis reveals Mendelian patterns in human inheritance

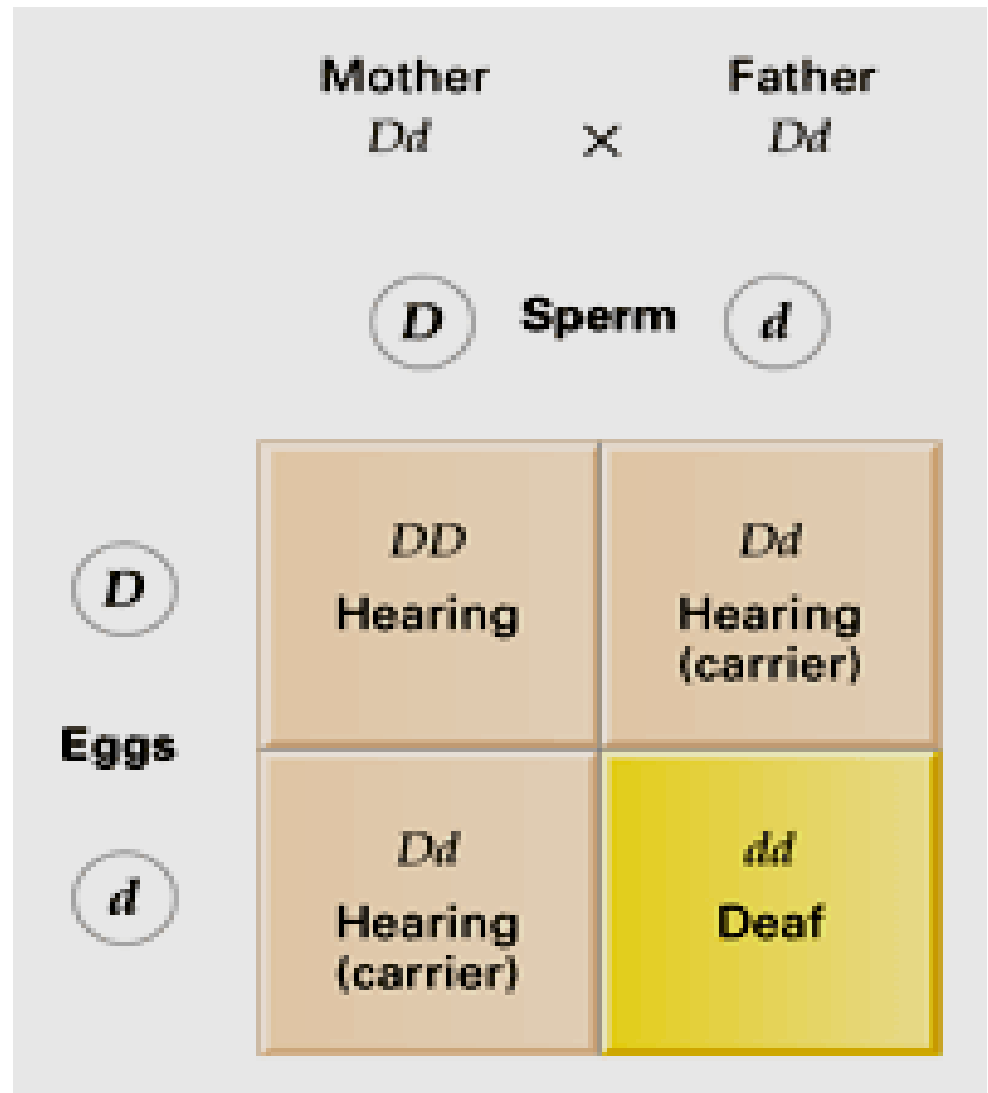


In these family trees, squares symbolize males and circles represent females. A horizontal line connecting a male and female (--) indicates a mating, with offspring listed below in their order of birth, from left to right. Shaded symbols stand for individuals with the trait being traced.

Disorders Inherited as Recessive Traits

Over a thousand human genetic disorders are known to have Mendelian inheritance patterns. Each of these disorders is inherited as a dominant or recessive trait controlled by a single gene. Most human genetic disorders are recessive.

A particular form of deafness is inherited as a recessive trait.

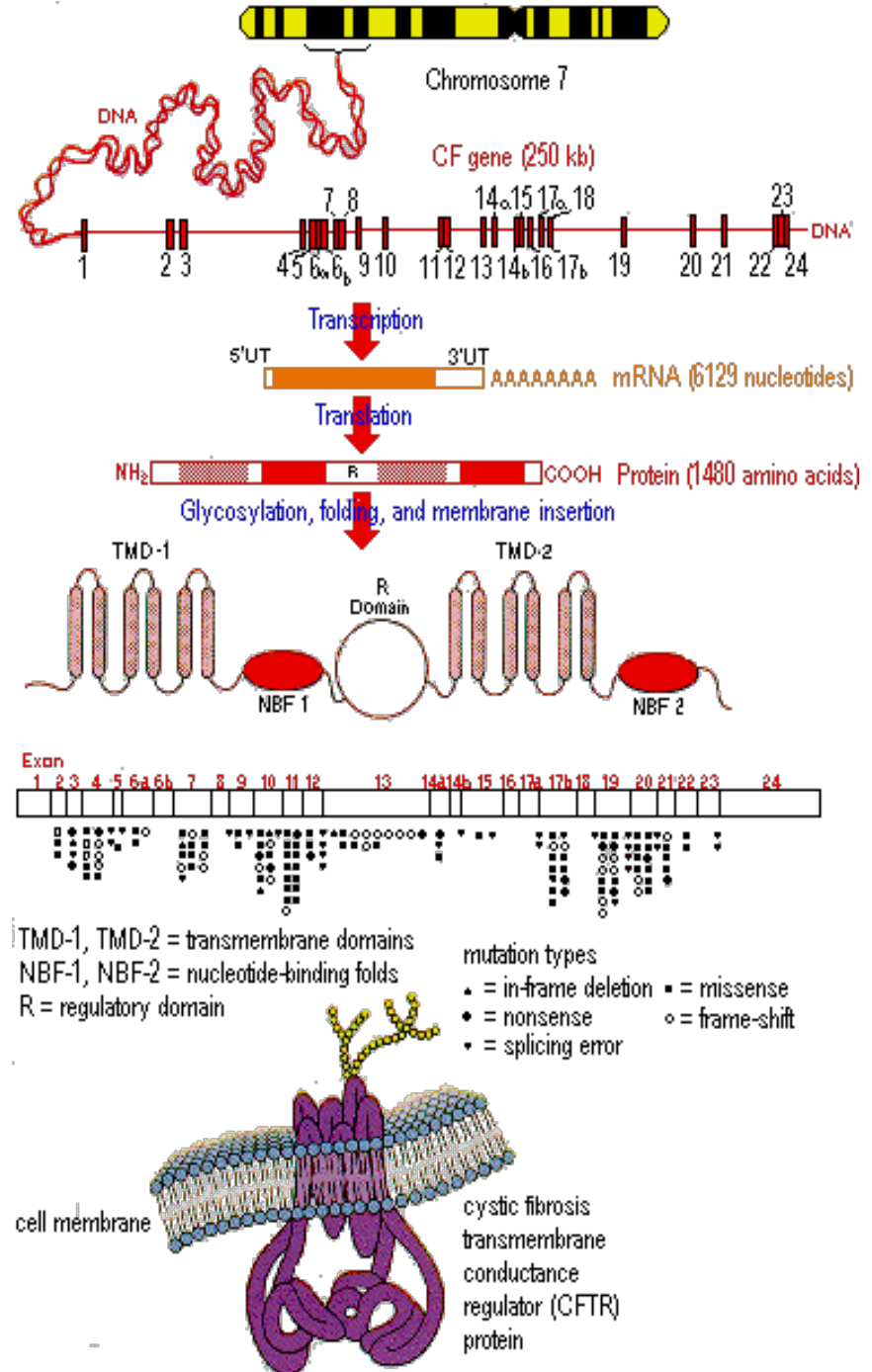


Many human disorders follow Mendelian patterns of inheritance

Cystic fibrosis, which strikes one out of every 2,500 whites of European descent but is much rarer in other groups. One out of 25 whites (4%) is a carrier.

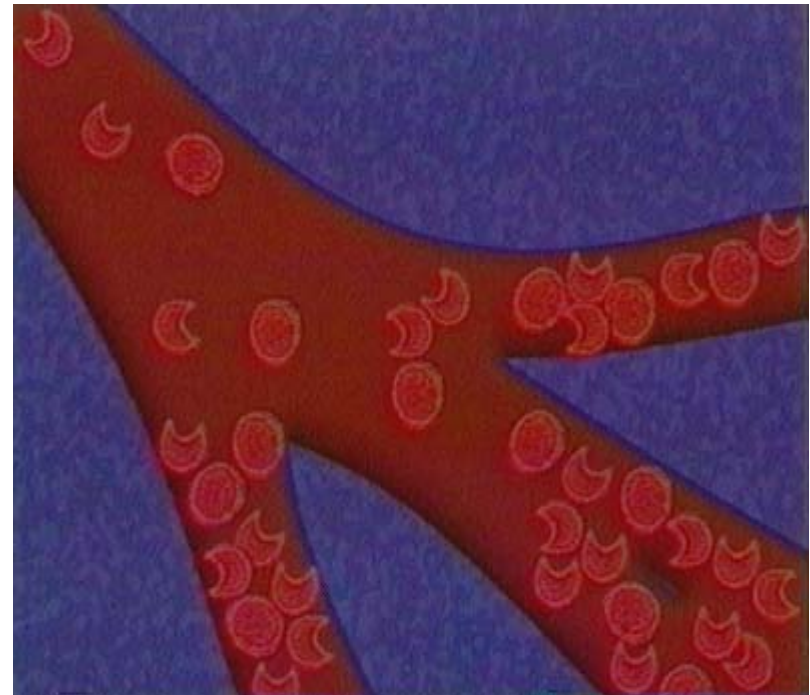
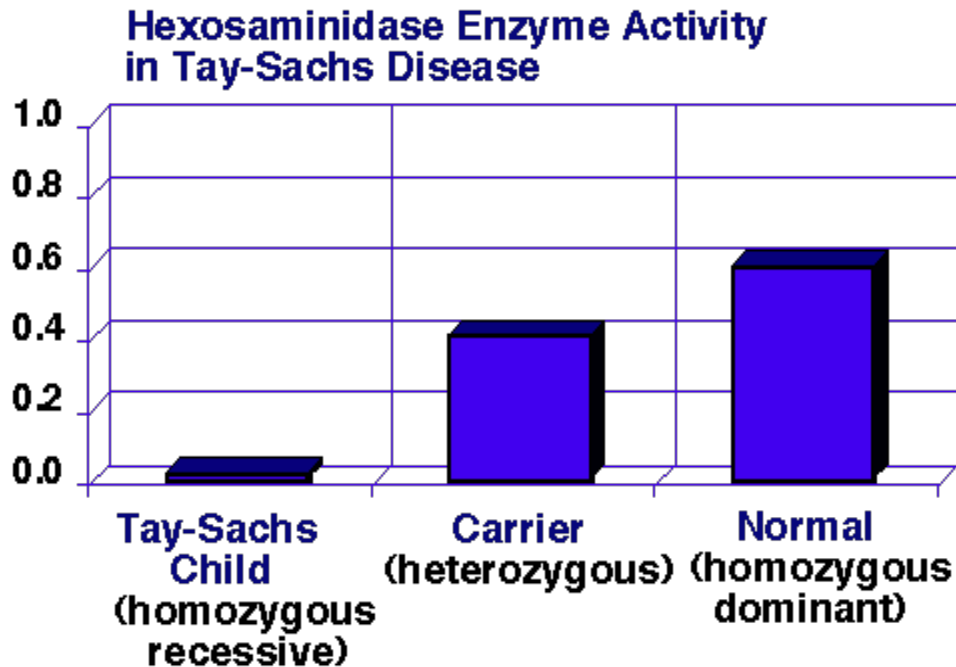
The normal allele for this gene codes for a membrane protein that functions in chloride ion transport between certain cells and the extracellular fluid. These chloride channels are defective or absent.

The result is an abnormally high concentration of extracellular chloride, which causes the mucus that coats certain cells to become thicker and stickier than normal.



Tay-Sachs disease is caused by a dysfunctional enzyme that fails to break down brain lipids of a certain class. Its proportionately high incidence of Tay-Sachs disease among Ashkenazic Jews, Jewish people whose ancestors lived in central Europe

Sickle-cell disease, which affects one out of 400 African Americans. Sickle-cell disease is caused by the substitution of a single amino acid in the hemoglobin protein of red blood cells



Dominantly Inherited Disorders

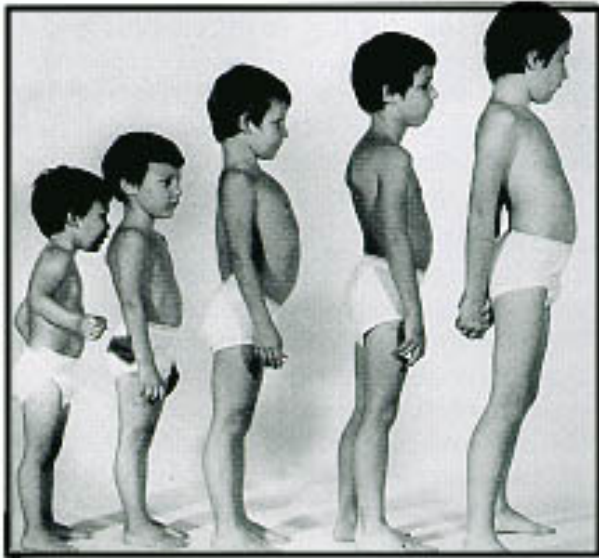
Achondroplasia, a form of dwarfism with an incidence of one case among every 10,000 people. Heterozygous individuals have the dwarf phenotype.

Huntington's disease, a degenerative disease of the nervous system, is caused by a lethal dominant allele that has no obvious phenotypic effect until the individual is about 35 to 45 years old.

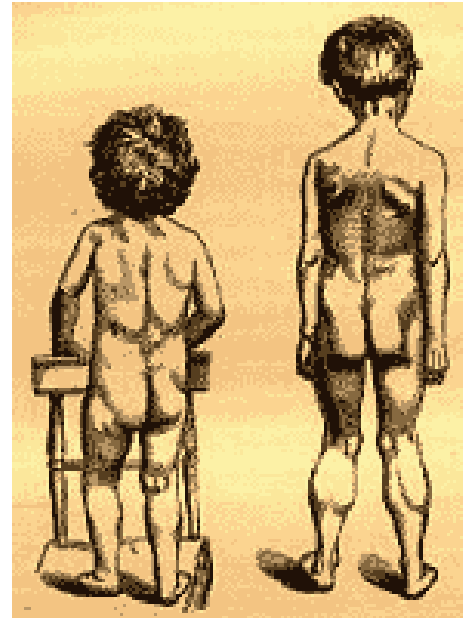


Sex-Linked Disorders in Humans

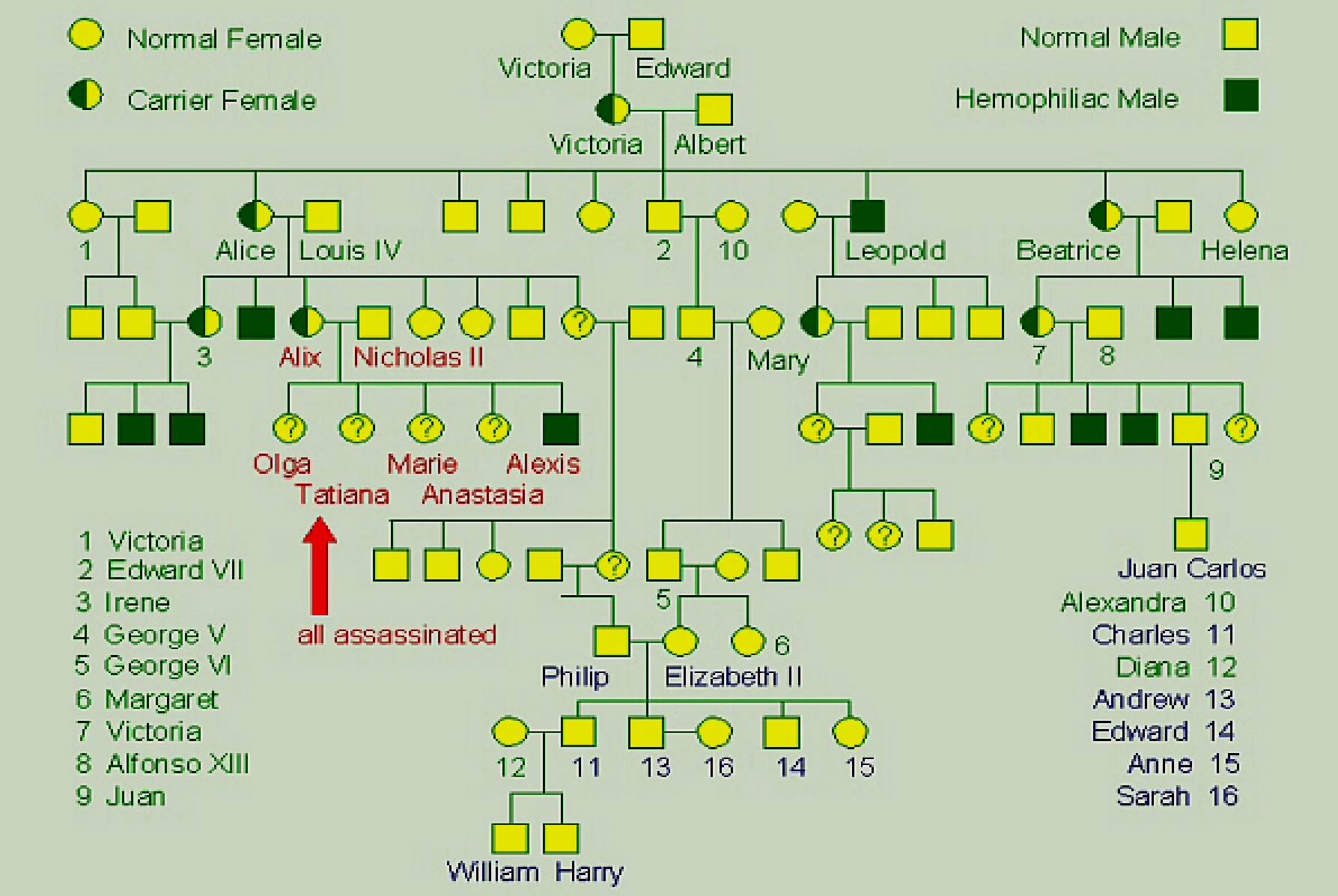
Duchenne muscular dystrophy, affects about one out of every 3,500 males born in the United States. People with Duchenne muscular dystrophy rarely live past their early 20s. The disease is characterized by a progressive weakening of the muscles and loss of coordination. Researchers have traced the disorder to the absence of a key muscle protein called dystrophin and have tracked the gene for this protein to a specific locus on the X chromosome.



Posture changes during progression of Duchenne muscular dystrophy.

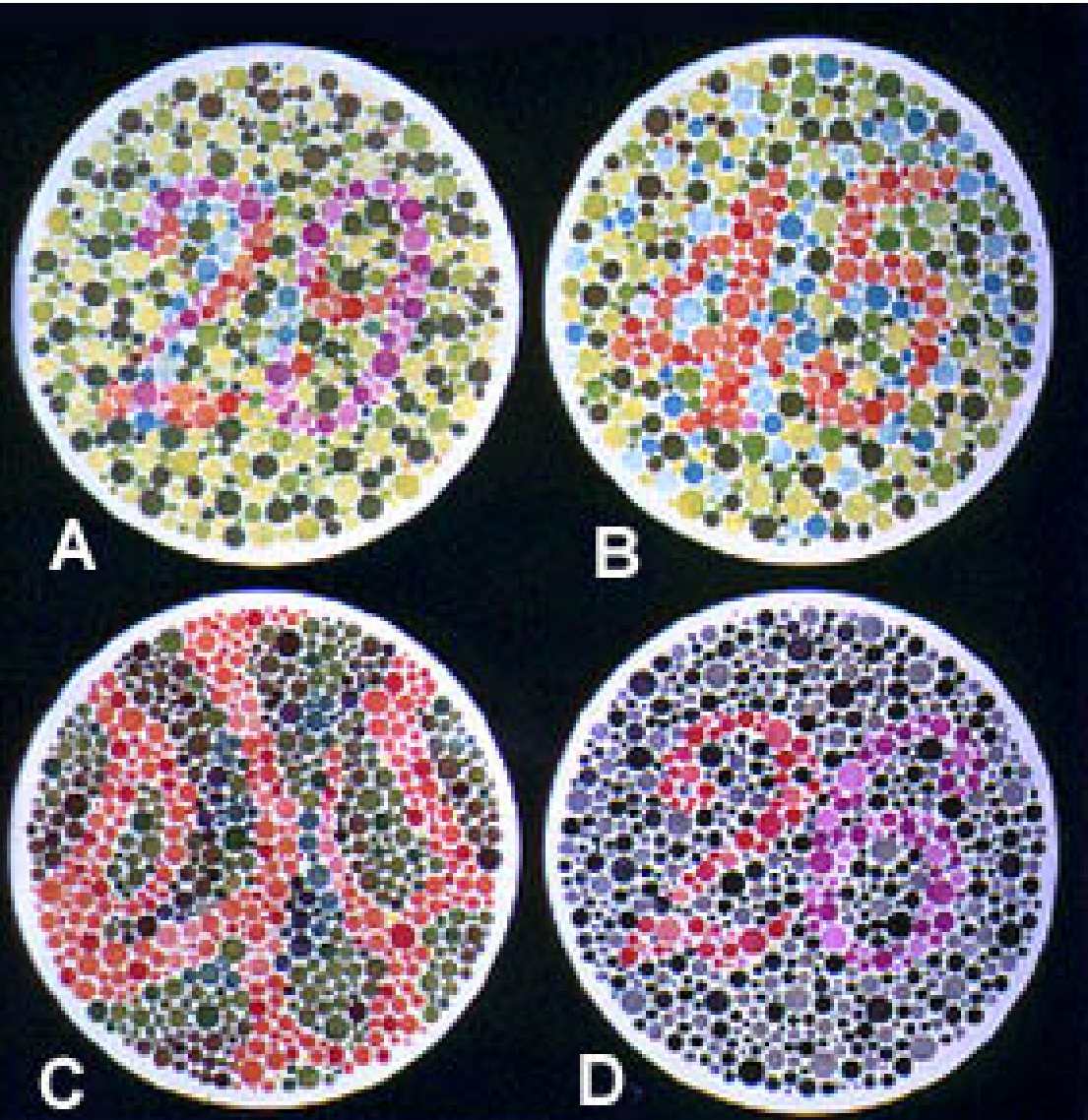


Hemophilia is a sex-linked recessive trait defined by the absence of one or more of the proteins required for blood clotting.



Color Blindness In Humans: An X-Linked Trait

Numbers That You Should See If You Are In One Of The Following Four Categories: [Some Letter Choices Show No Visible Numbers]



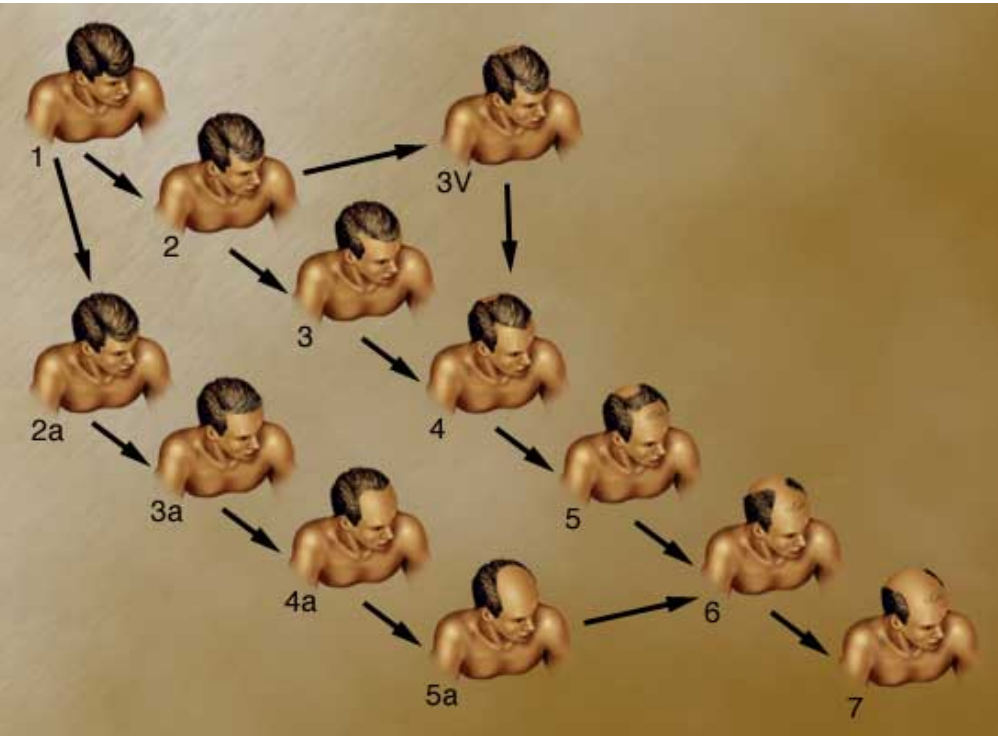
Sex-Linked Traits:

- 1. Normal Color Vision:
A: 29, B: 45, C: --, D: 26
- 2. Red-Green Color-Blind:
A: 70, B: --, C: 5, D: --
- 3. Red Color-blind:
A: 70, B: --, C: 5, D: 6
- 4. Green Color-Blind:
A: 70, B: --, C: 5, D: 2

Pattern Baldness In Humans: A Sex Influenced Trait

Baldness is an autosomal trait and is apparently influenced by sex hormones after people reach 30 years of age or older.

In men the gene is dominant, while in women it is recessive. A man needs only one allele (B) for the baldness trait to be expressed, while a bald woman must be homozygous for the trait (BB).



What are the probabilities for the children for a bald man and woman with no history of baldness in the family?