PowerPoint® Lecture Presentation for

Concepts of Genetics

Ninth Edition

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Chapter 4
Extensions of Mendelian Genetics

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Chapter 4 Contents

Continued

Chapter 4 Contents

4.1 Alleles Alter Phenotypes in Different Ways

Section 4.1

- Alternative forms of a gene are called **alleles**.
- **Mutation** is the source of alleles.
- The **wild-type** allele is the one that occurs most frequently in nature and is usually, but not always, **dominant**.

Section 4.1

- Often, a mutation causes the reduction or loss of the specific wild-type function.
- Such a case is called a loss of function mutation.
- If the loss is complete, the mutation has resulted in what is called a **null allele**.

Section 4.1

- Phenotypic traits may be influenced by more than one gene and the allelic forms of each gene involved.

4.2 Geneticists Use a Variety of Symbols for Alleles

Section 4.2

- **Dominant** alleles are usually indicated either by:
  - an italic uppercase letter (*D*)
  - letters (*Wr*)

Section 4.2

- **Recessive** alleles are usually indicated either by:
  - an italic lowercase letter (*d*)
  - an italic letter or group of letters (wr)

Section 4.2

- If no dominance exists, italic uppercase letters and superscripts are used to denote alternative alleles (R^1, R^2, C^W, C^R).

4.3 Neither Allele Is Dominant in Incomplete, or Partial Dominance

Section 4.3

- In **incomplete dominance**:
  - neither trait is dominant
  - offspring from a cross between parents with contrasting traits may have an intermediate phenotype

Section 4.3

- The phenotypic ratio is identical to the genotypic ratio in cases of incomplete dominance (**Figure 4.1**).

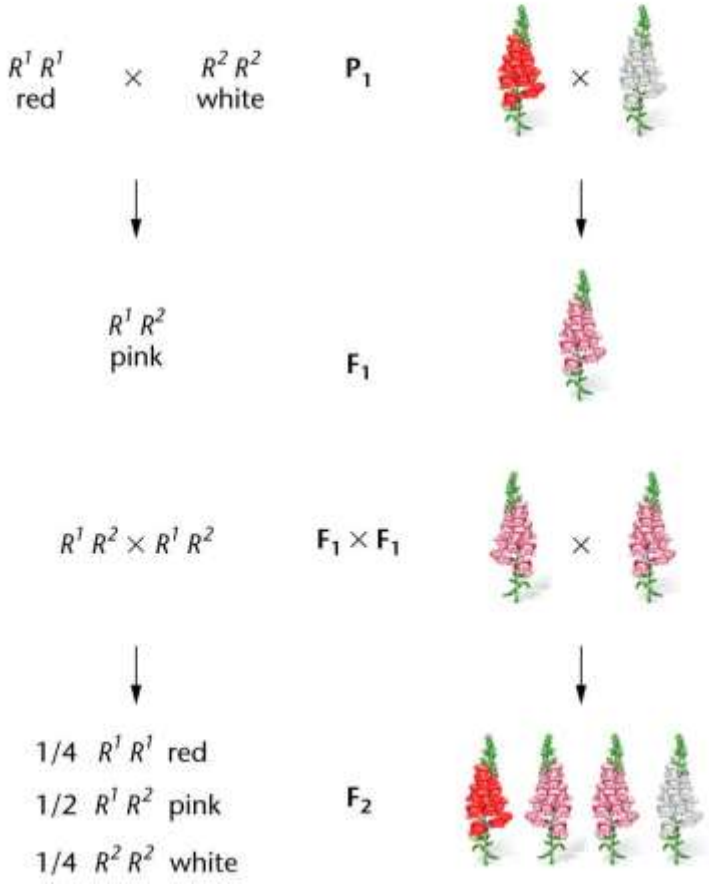
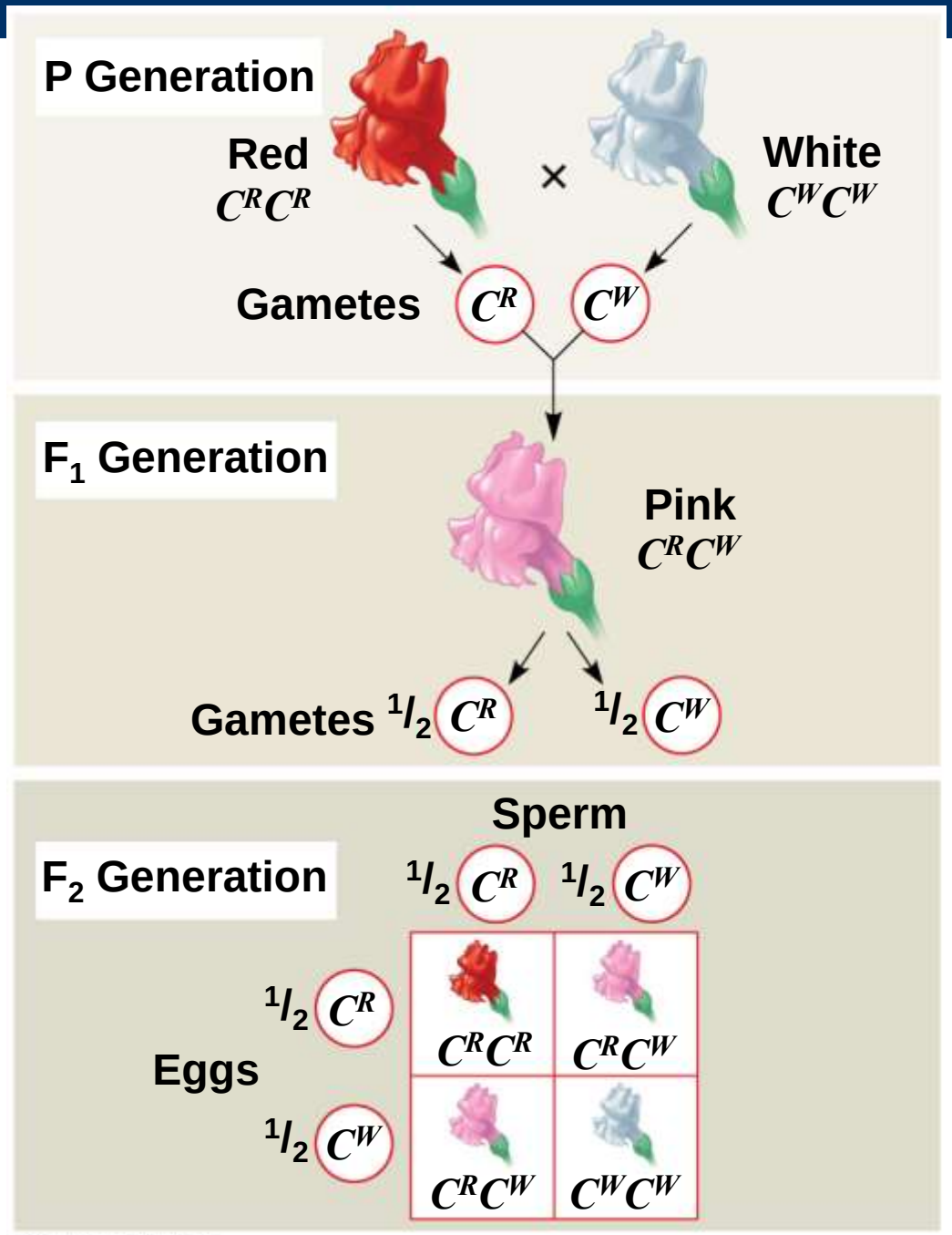


Figure 4.1

Figure 14.10-3



Section 4.3

- The **threshold effect** comes about if normal phenotypic expression occurs whenever a certain level (usually 50% or less) of gene product is attained.

4.4 In Codominance, the Influence of Both Alleles in a Heterozygote Is Clearly Evident

Section 4.4

- **Codominance** occurs when joint expression of two alleles of a gene in a heterozygote results in phenotypic detection of both gene products.
- One example is the MN blood group.

4.5 Multiple Alleles of a Gene May Exist in a Population

Section 4.5

- **Multiple alleles** (>2) can be studied only in populations, because any individual will have at most two alleles of the same gene.

Section 4.5

- The **ABO blood groups** are an example of multiple alleles.
- Each individual is A, B, AB, or O phenotype as a result of dominance of the I^A and I^B alleles to the I^O allele and codominance of the I^A and I^B alleles to each other (**Table 4.1**).

Figure 14.11

(a) The three alleles for the ABO blood groups and their carbohydrates

Allele	I^A	I^B	i
Carbohydrate	A 	B 	none

(b) Blood group genotypes and phenotypes

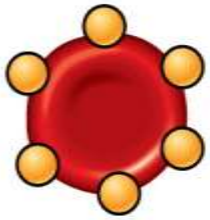
Genotype	I^AI^A or I^Ai	I^BI^B or I^Bi	I^AI^B	ii
Red blood cell appearance				
Phenotype (blood group)	A	B	AB	O

TABLE 4.1

Potential Phenotypes in the Offspring of Parents with All Possible ABO Blood Group Combinations, Assuming Heterozygosity Whenever Possible

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

Section 4.5

- The I^A allele is responsible for an enzyme that can add the terminal sugar N-acetylgalactosamine (AcGalNH) to the H substance.

Section 4.5

- The I^B allele is responsible for a modified enzyme that cannot add N-acetylgalactosamine but instead can add a terminal galactose.
- The O phenotype results from an absence of either terminal sugar (**Figure 4.2**).

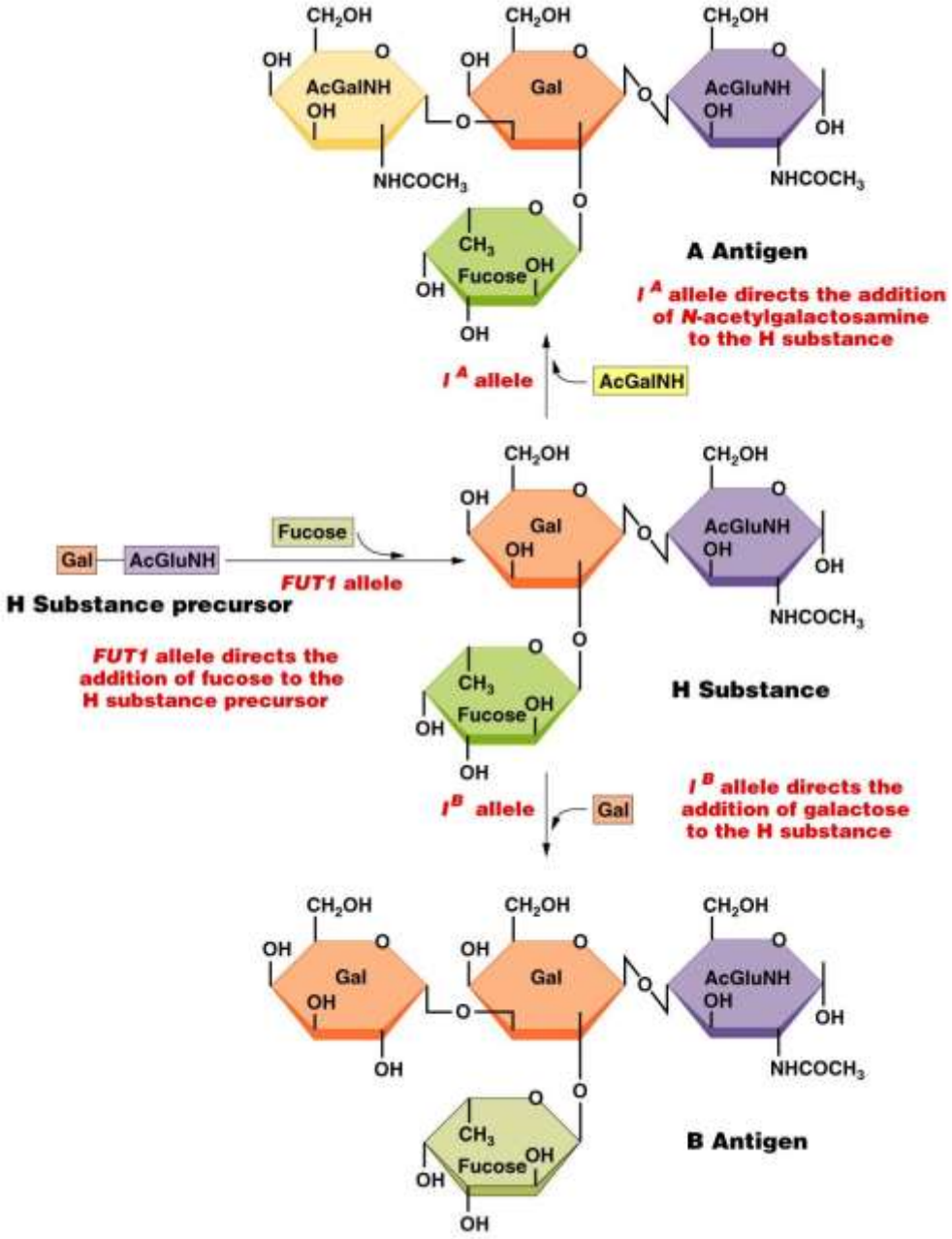
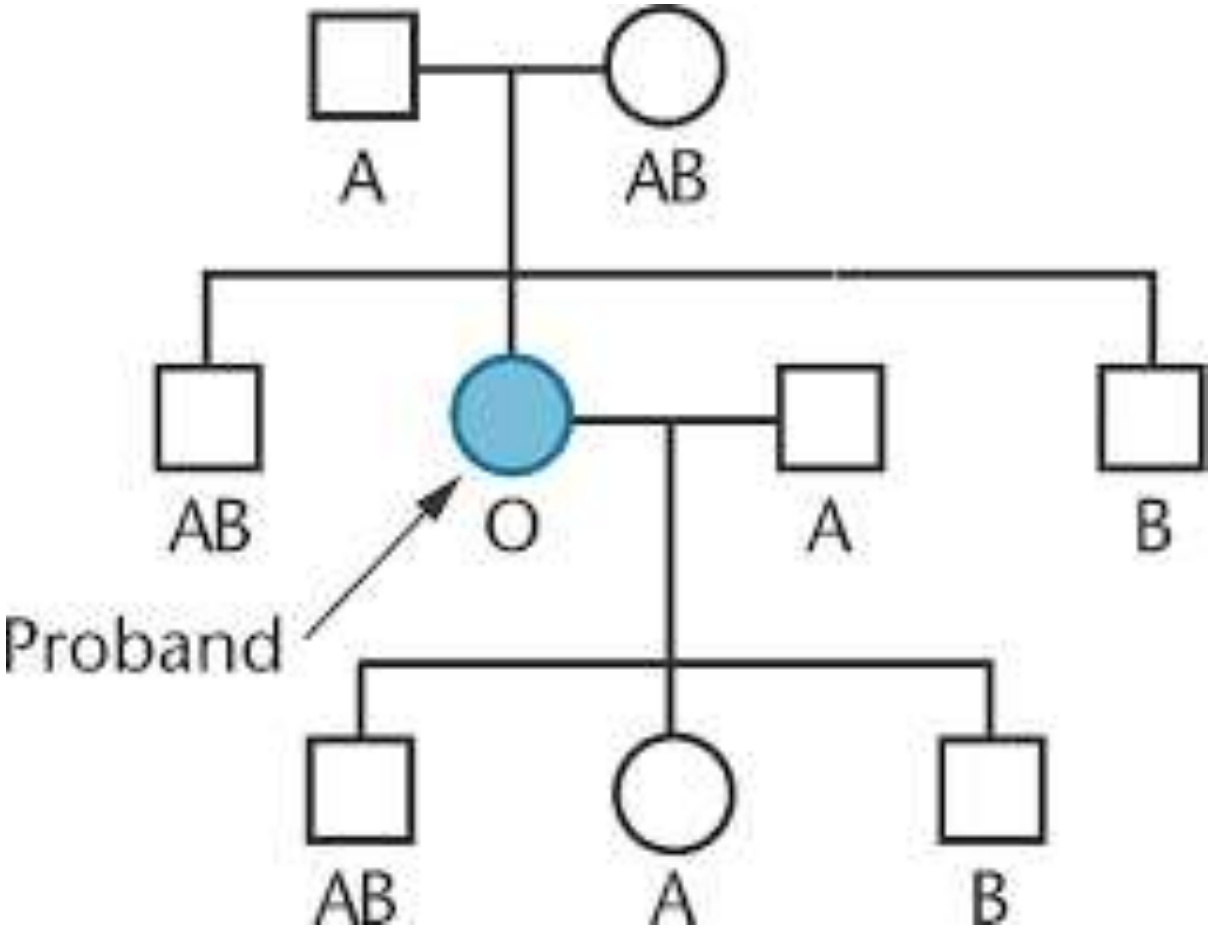


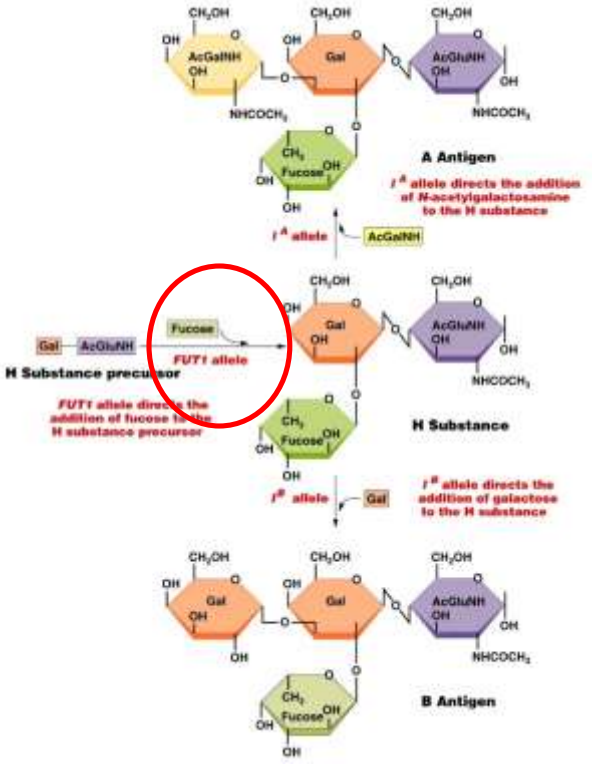
Figure 4.2

The bombay phenotype


Boy Belongs To Rare Bombay Blood Group And Suffers From Thalassaemia

There is no blood match for him





4.6 Lethal Alleles Represent Essential Genes

Section 4.6

- A loss of function mutation can sometimes be tolerated in the heterozygous state, but may behave as a **recessive lethal allele** in the homozygous state.
- In this case, homozygous recessive individuals will not survive.

Section 4.6

- The allele responsible for a lethal effect when it is homozygous can also result in a distinctive mutant phenotype when it is heterozygous.
- Such an allele is behaving as a recessive lethal, but is dominant with respect to the phenotype (**Figure 4.4**).

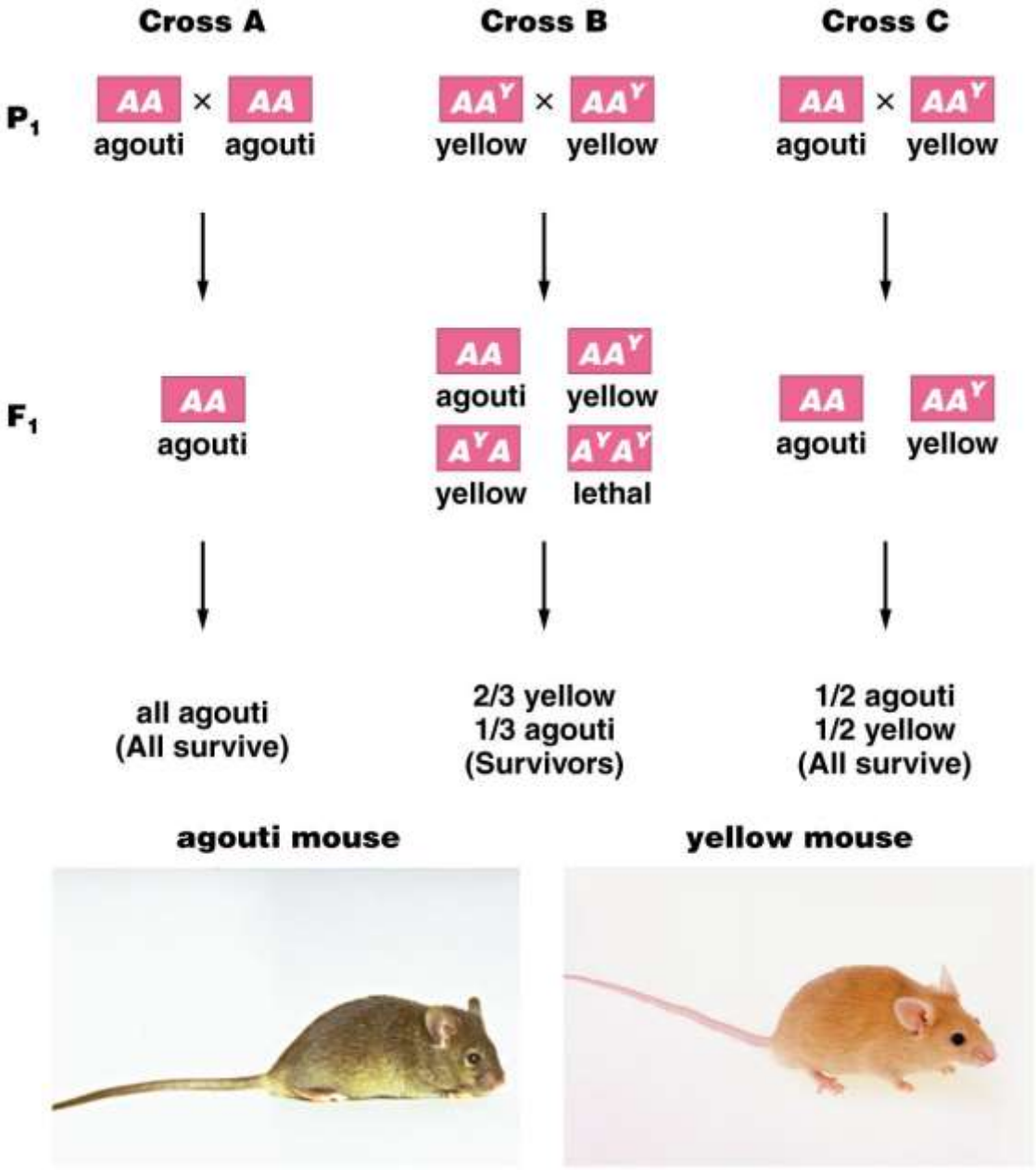


Figure 4.4

Section 4.6

- In some cases, a mutation can be a **dominant lethal allele**, in which case the heterozygote will not survive.
- Huntington disease is one example.

Section 4.6

- For dominant lethal alleles to exist, the affected individual must reproduce before dying.

4.7 Combinations of Two Gene Pairs Involving Two Modes of Inheritance Modify the 9:3:3:1 Ratio

Section 4.7

- Mendel's principle of independent assortment applies to situations in which two modes of inheritance occur simultaneously, provided that the genes controlling each character are not linked on the same chromosome.

Section 4.7

- The probability of each phenotype arising in a cross can be determined by the forked-line method or by Punnett square (**Figure 4.5**), assuming that the genes under consideration undergo independent assortment.

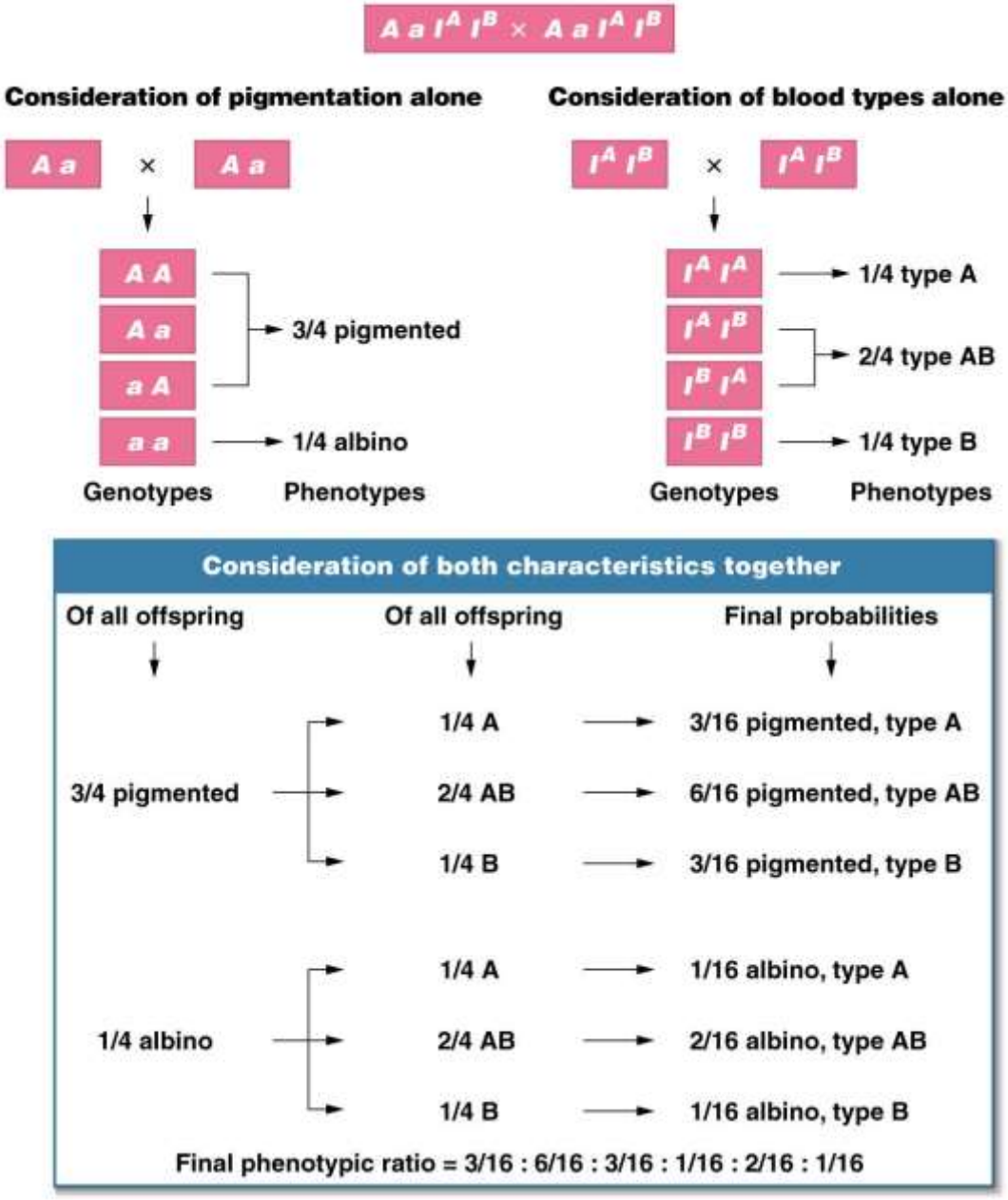


Figure 4.5

4.8 Phenotypes Are Often Affected by More Than One Gene

Section 4.8

- Many traits characterized by a distinct phenotype are affected by more than one gene.
- In gene interaction, the cellular function of numerous gene products contributes to the development of a common phenotype.

Section 4.8

- **Epistasis** occurs when:
  - one gene masks the effect of another gene, or
  - two gene pairs complement each other such that one dominant allele is required at each locus to express a certain phenotype

Section 4.8

- The Bombay phenotype for ABO blood groups is an example of epistasis in which the homozygous recessive condition at one locus masks the expression of a second locus (**Figure 4.6**).

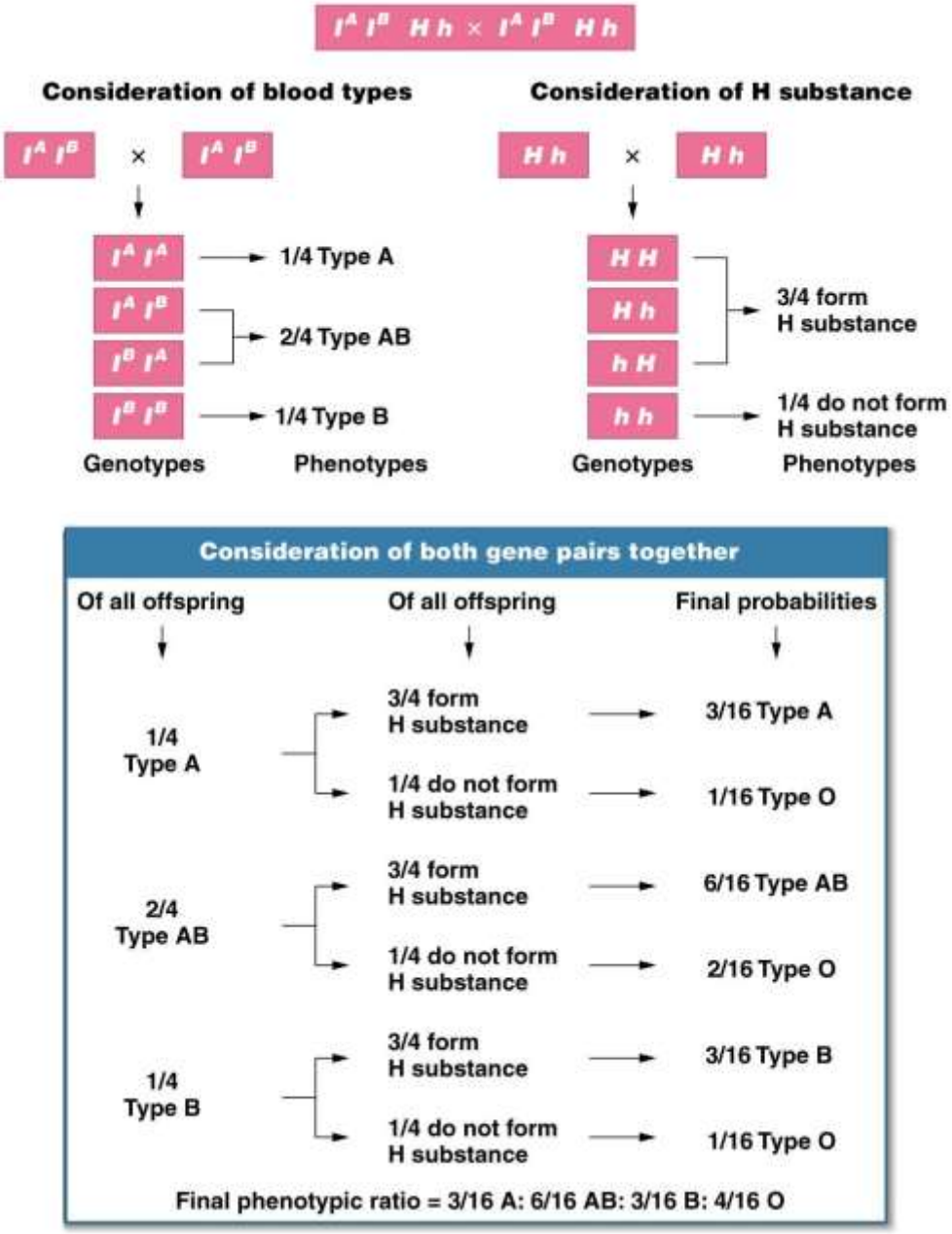


Figure 4.6

Section 4.8

- When studying a single characteristic, a ratio expressed in 16 parts (e.g., 3:6:3:4) suggests that epistasis is occurring.

Section 4.8

- Eight cases of epistasis are described in **Figure 4.8**.
- These include recessive epistasis (case 1), dominant epistasis (case 2), and complementary gene interaction (case 3).

Case	Organism	Character	F_2 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 and 3/16 red	black	jet	black	6:3:3:4

Figure 4.8

4.9 Complementation Analysis Can Determine if Two Mutations Causing a Similar Phenotype are Alleles

Section 4.9

- Two cases of mutation in *Drosophila* (**Figure 4.11**)
  - Case 1: All offspring develop normal wings
  - Case 2: All offspring fail to develop normal wings

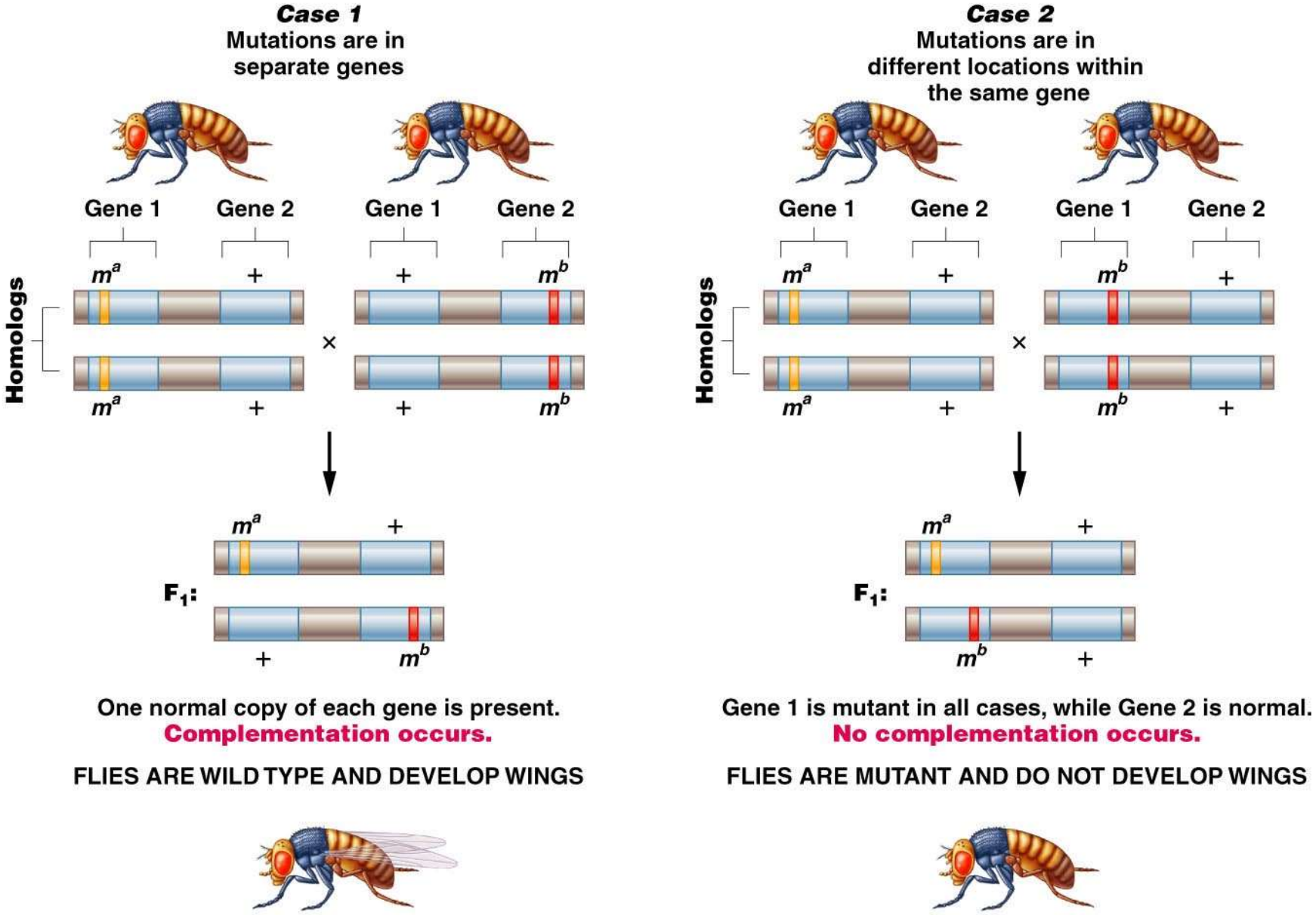


Figure 4.11

4.10 Expression of a Single Gene May Have Multiple Effects

Section 4.10

- **Pleiotropy** occurs when expression of a single gene has multiple phenotypic effects, and it is quite common.
- Examples of pleiotropy are Marfan syndrome and porphyria variegata.

4.11 X-Linkage Describes Genes on the X Chromosome

Section 4.11

- Genes present on the X chromosome exhibit unique patterns of inheritance due to the presence of only one X chromosome in males.

Section 4.11

- *Drosophila* eye color was one of the first examples of **X-linkage** described (**Figure 4.12** and **Figure 4.13**).

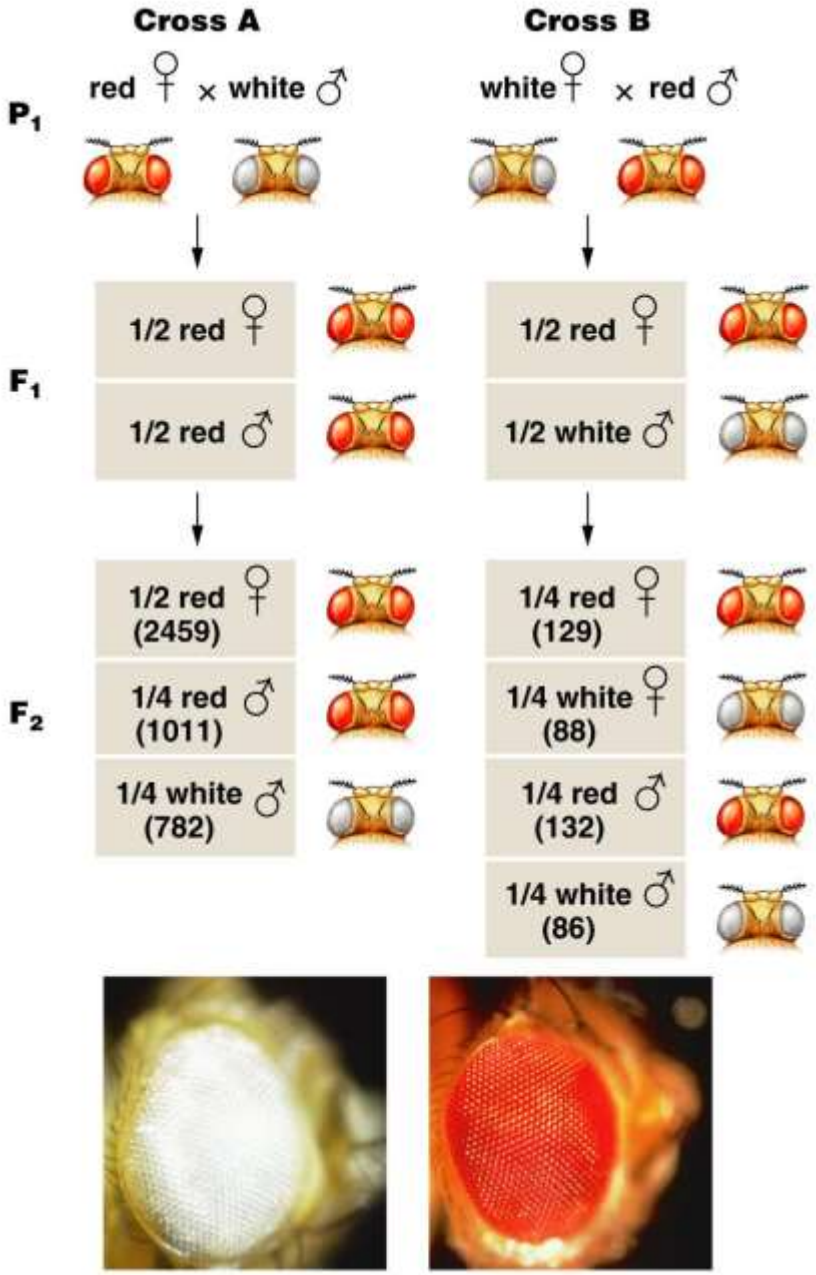


Figure 4.12

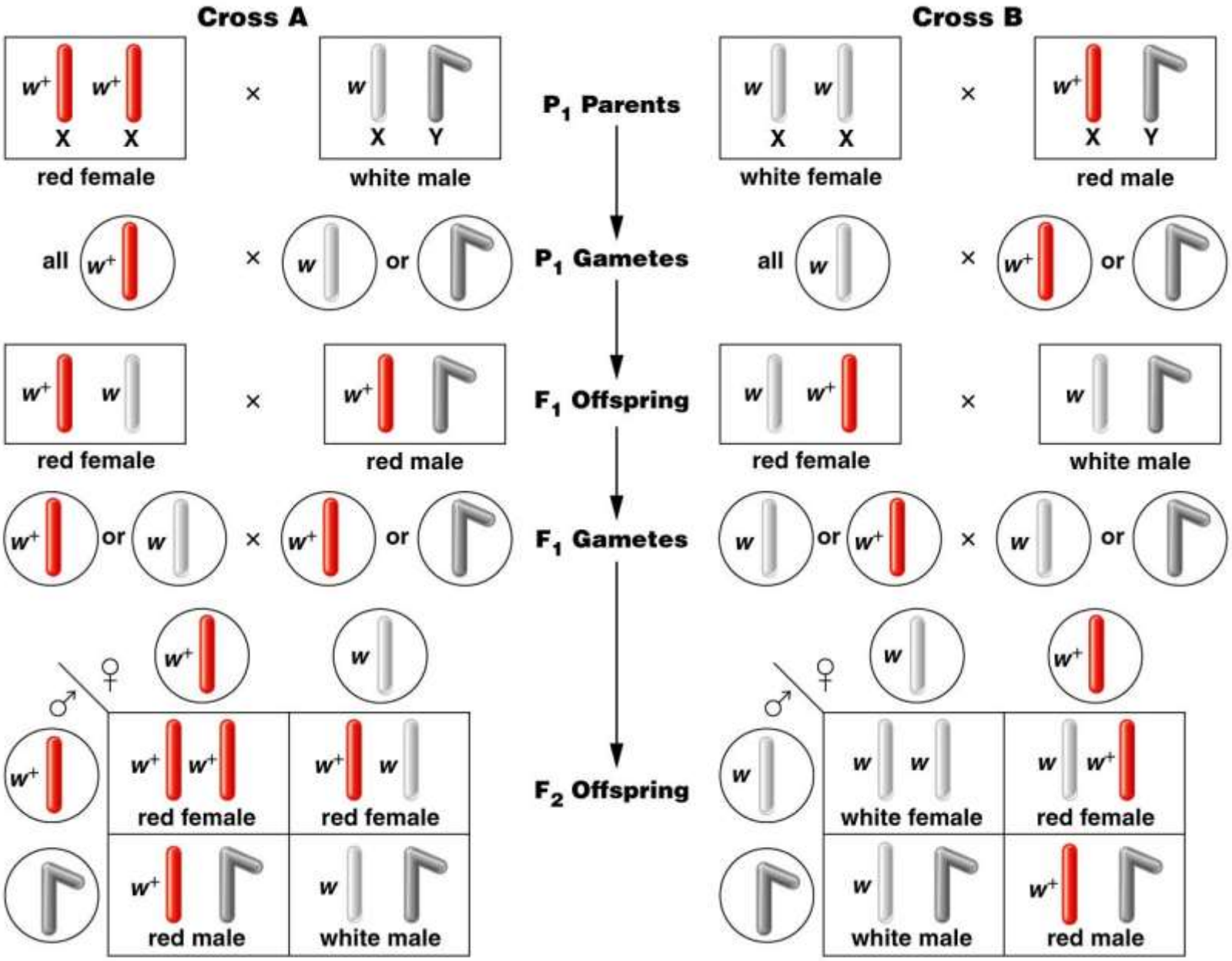


Figure 4.13

Section 4.11

- **Table 4.3** lists a number of human X-linked genes.

TABLE 4.3

Human X-Linked Traits

Condition	Characteristics
Color blindness, deutan type	Insensitivity to green light
Color blindness, protan type	Insensitivity to red light
Fabry's disease	Deficiency of galactosidase A; heart and kidney defects, early death
G-6-PD deficiency	Deficiency of glucose-6-phosphate dehydrogenase; severe anemic reaction following intake of primaquines in drugs and certain foods, including fava beans
Hemophilia A	Classic form of clotting deficiency; deficiency of clotting factor VIII
Hemophilia B	Christmas disease; deficiency of clotting factor IX
Hunter syndrome	Mucopolysaccharide storage disease resulting from iduronate sulfatase enzyme deficiency; short stature, clawlike fingers, coarse facial features, slow mental deterioration, and deafness
Ichthyosis	Deficiency of steroid sulfatase enzyme; scaly dry skin, particularly on extremities
Lesch–Nyhan syndrome	Deficiency of hypoxanthine-guanine phosphoribosyltransferase enzyme (HPRT) leading to motor and mental retardation, self-mutilation, and early death
Muscular dystrophy	Progressive, life-shortening disorder characterized by muscle degeneration and weakness; (Duchenne type) sometimes associated with mental retardation; deficiency of the protein dystrophin

Table 4.3

Section 4.11

- Lethal X-linked recessive disorders are observed only in males.
- Females can only be heterozygous carriers that do not develop the disorders.

4.12 In Sex-Limited and Sex-Influenced Inheritance, an Individual's Sex Influences the Phenotype

Section 4.12

- **Sex-limited inheritance** occurs in cases where the expression of a specific phenotype is absolutely limited to one sex.
- In **sex-influenced inheritance**, the sex of an individual influences the expression of a phenotype that is not limited to one sex or the other.

Section 4.12

- In sex-limited and sex-influenced inheritance, expression of autosomal genes responsible for a certain phenotype depends on the hormone constitution of the individual.
- Thus, one phenotype may be expressed in males and another in females.

4.13 Genotypic Background and the Environment May Alter Phenotypic Expression

Section 4.13

- Phenotypic expression of a trait may be influenced by environment as well as by genotype.

Section 4.13

- The degree of expression depends on the **penetrance** (the percentage of individuals that show at least some degree of expression) of a mutant genotype and the **expressivity** (the range of expression) of the mutant genotype.

Section 4.13

- Although it is difficult to assess the specific effect of the genetic background and the expression of a gene responsible for determining a potential phenotype, two effects of genetic background have been well characterized.

Section 4.13

- First, genetic suppression occurs when other genes affect the phenotype produced by the gene in question.
- Second, the physical location of a gene may influence its expression due to a position effect.

Section 4.13

- Mutations affected by temperature are called conditional or temperature-sensitive mutations.
- They are useful in studying mutations that affect essential processes.

Section 4.13

- Nutritional mutations may prevent the phenotype from reflecting the genotype.
- Examples include mutations in a biosynthetic pathway.

Section 4.13

- As a result of genetic anticipation, some heritable disorders exhibit a progressively earlier age of onset with an increased severity in each generation.

Section 4.13

- In cases of genomic (parental) imprinting, phenotypic expression may depend on the parental origin of the chromosome.
- Imprinting is thought to occur before or during gamete formation and may involve DNA methylation (**Figure 4.20**).

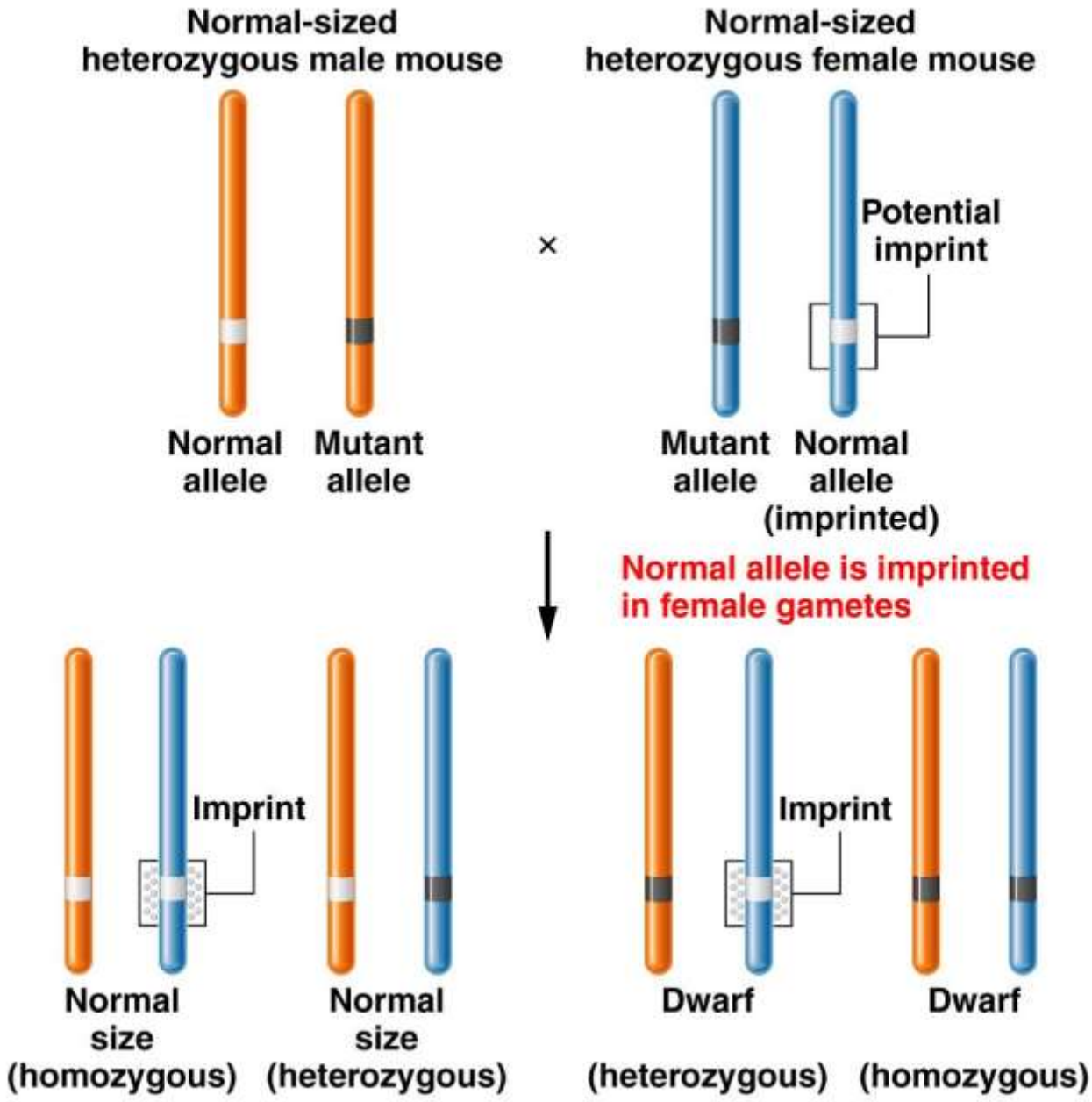


Figure 4.20