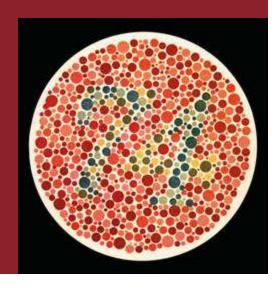


Genetics: Mendel and Beyond



10 Genetics: Mendel and Beyond

fertilization

crossing over

Put the following words in their correct location in the sentences below.

haploid	prophase I	diploid	testes	eggs
2	ovaries	sperm	46	puberty
gametes	23	metaphase I	DNA	chromosome
-	-	specifically designed		ere is genetic variation in
onspring. Sex cer	whoroas mal	e sex cells are called		and are
	, whereas mai	Mala and fo		anu are
manufactured in t	ne	Male and fe	emaie sex ceiis are	conectively caned
	The process	of	occurs in th	e reproductive organs of
both males and fe	males. This process	s reduces the chromo	some number from	n to During
this process, a spe	cial event called		occurs, where ger	netic material breaks off
				This special event that
prevents cloning of	occurs during a pha	se called	Altho	ough the process that
makes sex cells is	much like mitosis ii	n normal body cells, t	there are several d	ifferences including the
phase of	, who	ere chromosomes line	e up side by side, r	ather than in a straight
line. Also, when s	ex cells are made, o	ne cell divides to mal	ke cells, then	divides again to make
cells. In mal	es, all of these cells	become sperm, but i	n females, only on	e of these cells will
become an egg. T	he process where a	n egg and sperm com	bine is called	, and
				ganisms become sexually
active (able to pro	duce offspring) is c	alled	•	

meiosis

zygote

People have been cross-breeding plants and animals for at least 5,000 years.

By the ninteenth century, plant breeding was widespread.

At the time, breeders worked under two assumptions about how inheritance works:

- Each parent contributes equally to offspring. (Correct.) Supported by reciprocal crosses, 1770s, by Kölreuter.
- Hereditary determinants blend in the offspring. (Incorrect.) It was thought that once hereditary elements had blended they could never be separated.
- Gregor Mendel's studies refuted this.
- Before genetics were understood, people thought that animals and plants could only be made bigger by feeding them more.

Mendel was an Austrian monk.

His studies in physics and mathematics were a strong influence on his use of quantitative experimental methods.

Over seven years, he made crosses with 24,034 plants.

His new theory of inheritance was published in 1866, but was largely ignored.

Most biologists at the time were not used to thinking in mathematical terms.

Even Darwin missed the significance of Mendel's work.

Figure 10.1 Gregor Mendel and His Garden



By 1900, meiosis had been observed.

Three plant geneticists realized that chromosomes and meiosis provided a physical explanation for Mendel's results.

Mendel chose to work with the garden pea.

He could control pollination and fertilization—he could be sure of the parents of offspring.

The peas naturally self-pollinate.

Character: observable physical feature (e.g., flower color).

Trait: form of a character (e.g., purple flowers or white flowers).

A heritable trait is passed from parent to offspring.

Mendel looked for well-defined, **true- breeding** traits—the observed trait is the only one present for many generations.

True-breeding strains were isolated by inbreeding and selection.

He concentrated on seven traits.

	PARENTAL GENERATION PHENOTYPES			F ₂ GENERATI	F ₂ GENERATION PHENOTYPES		
	DOMINANT	RECESSIVE		DOMINANT	RECESSIVE	TOTAL	RATIO
	Spherical seeds ×	Wrinkled seeds		5,474	1,850	7,324	2.96:
	Yellow seeds ×	Green seeds		6,022	2,001	8,023	3.01:
(AB	Purple flowers ×	White flowers	CARS.	705	224	929	3.15:
	Inflated pods ×	Constricted pode	S	882	299	1,181	2.95:
	Green pods ×	Yellow pods		428	152	580	2.82:
A STATE OF THE STA	• Axial flowers ×	Terminal flowers	AND WEET	651	207	858	3.14:
-2102	Tall stems ×	Dwarf stems	488	787	277	1,064	2.84:

Mendel's crosses:

- Pollen from one parent was transferred to the stigma of the other parent.
 Parental generation = P.
- Resulting offspring = first filial generation or F₁.
- If F₁ plants self pollinate, produce second filial generation or F₂.

Figure 10.2 A Controlled Cross between Two Plants (Part 1)

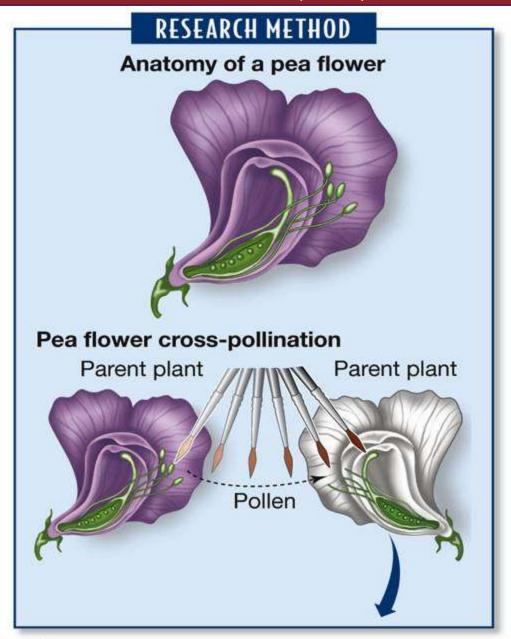
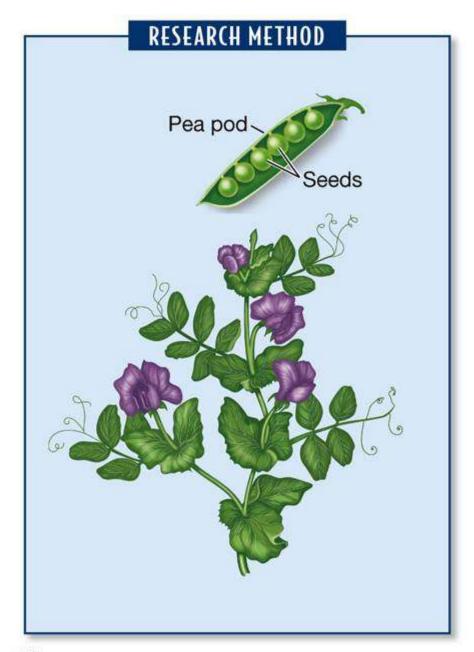


Figure 10.2 A Controlled Cross between Two Plants (Part 2)



Mendel's first experiment:

Crossed plants differing in just one trait (P).

F₁ generation are *monohybrids*.

The monohybrids were then allowed to self pollinate to form the F₂ generation: a **monohybrid cross**.

Mendel repeated this for all seven traits.

	PARENTAL GENERATION PHENOTYPES			F ₂ GENERATI	F ₂ GENERATION PHENOTYPES		
	DOMINANT	RECESSIVE		DOMINANT	RECESSIVE	TOTAL	RATIO
	Spherical seeds ×	Wrinkled seeds		5,474	1,850	7,324	2.96:
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	Inflated pods ×	Constricted pode	S	882	299	1,181	2.95:
	Green pods ×	Yellow pods		428	152	580	2.82:
A STATE OF THE STA	• Axial flowers ×	Terminal flowers	AND WEET	651	207	858	3.14:
-2102	Tall stems ×	Dwarf stems	488	787	277	1,064	2.84:

One trait of each pair disappeared in the F₁ generation and reappeared in the F₂—these traits are *recessive*.

The trait that appears in the F₁ is the *dominant* trait.

The ratio of dominant to recessive in the F_2 was about 3:1.

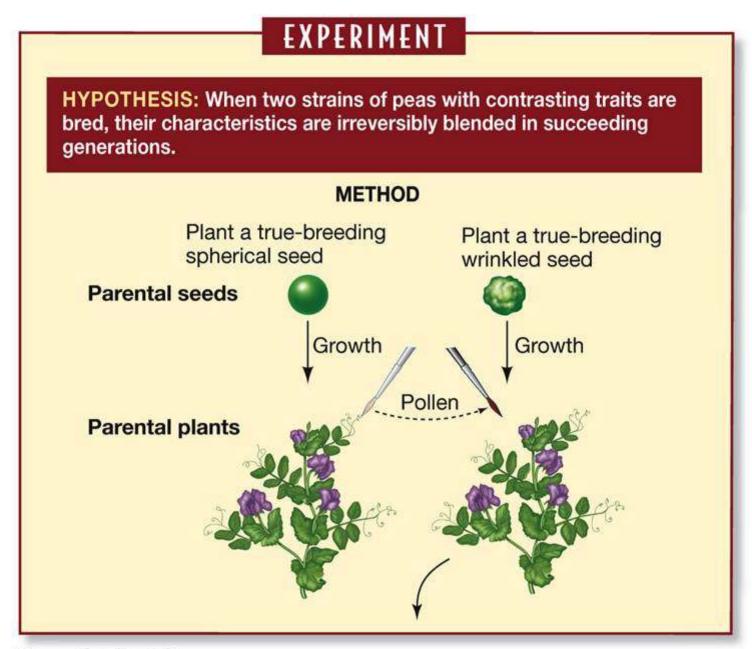
Reciprocal crosses yielded the same results: it made no difference which parent contributed pollen.

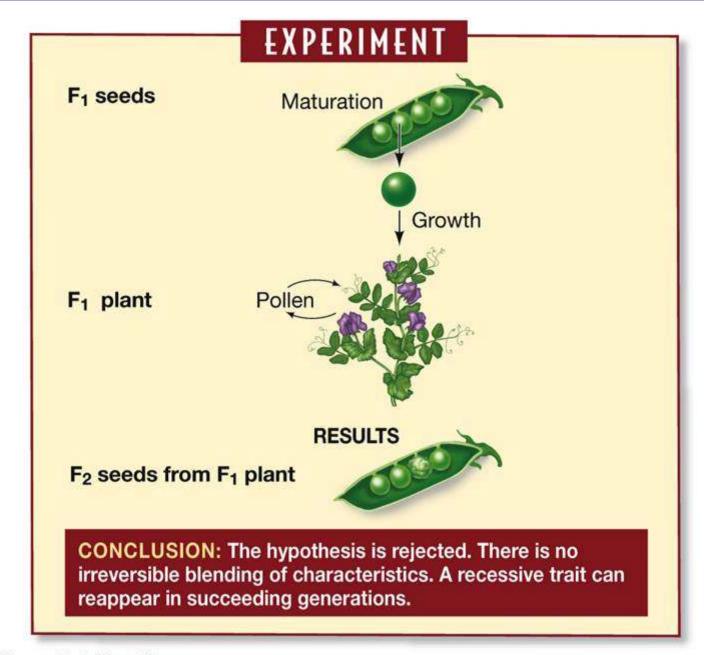
The idea that each parent contributes equally was supported.

The blending theory was not supported by Mendel's crosses.

Mendel proposed that the heritable units were *discrete particles*—the **particulate theory**.

Each plant has two particles for each character, one from each parent.





Mendel also concluded that each gamete contains only one particle (or unit), but the zygote contains two—because it is produced from the fusion of two gametes.

The "particles" are now called **genes**.

The totality of all genes in an organism is the **genome**.

The true-breeding plants in the P generation had two identical copies of the particle (gene) for each character.

Example: spherical *SS*; wrinkled *ss*Gametes from *SS* will have one *S*Gametes from *ss* will have one *s*Offspring (F₁) will be *Ss*S is dominant; *s* is not expressed in F₁

Alleles: different forms of a gene

True-breeding individuals have two copies of the same allele—they are homozygous for the allele (e.g., ss).

Heterozygous individuals have two different alleles (e.g., Ss).

Phenotype: physical appearance of an organism (e.g., spherical seeds).

Genotype: the genetic makeup (e.g., Ss).

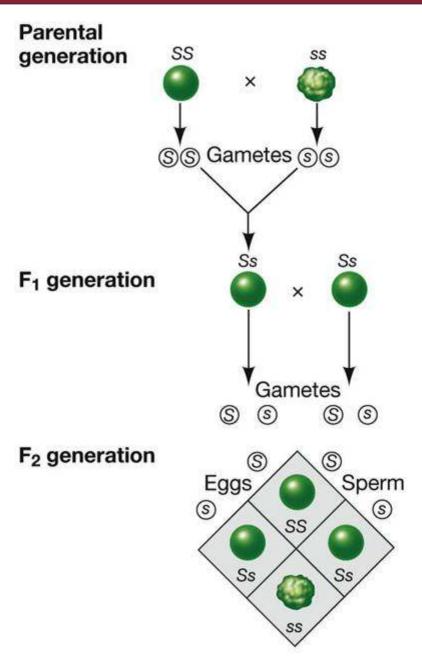
Spherical seeds can the result of two different genotypes—SS or Ss.

Mendel's first law

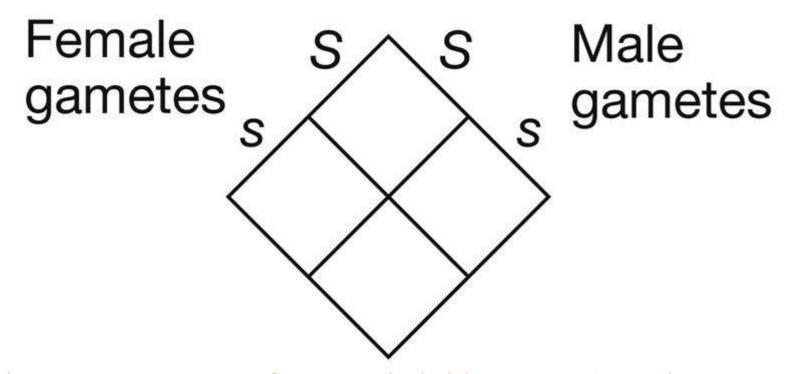
The Law of Segregation: the two copies of a gene separate when an individual makes gametes.

When the F₁ self-pollinates, there are three ways to get the dominant trait (e.g., spherical), only one way to get the recessive (wrinkled)—resulting in the 3:1 ratio.

Figure 10.4 Mendel's Explanation of Inheritance



Allele combinations can be predicted using a **Punnett square**:



Practice Punnett Squares for Monohybrid Crosses (genetics example)

A gene is a sequence on a DNA molecule that resides at a particular site on a chromosome—the **locus**—and encodes a particular character.

Genes are expressed as proteins with particular functions.

Different alleles of a gene separate during meiosis.

Figure 10.5 Meiosis Accounts for the Segregation of Alleles (Part 1)

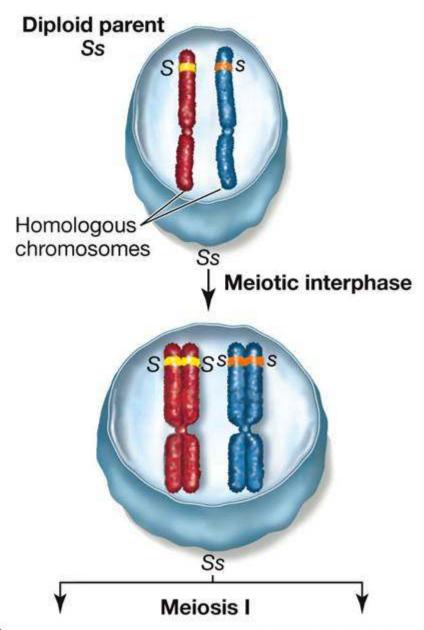
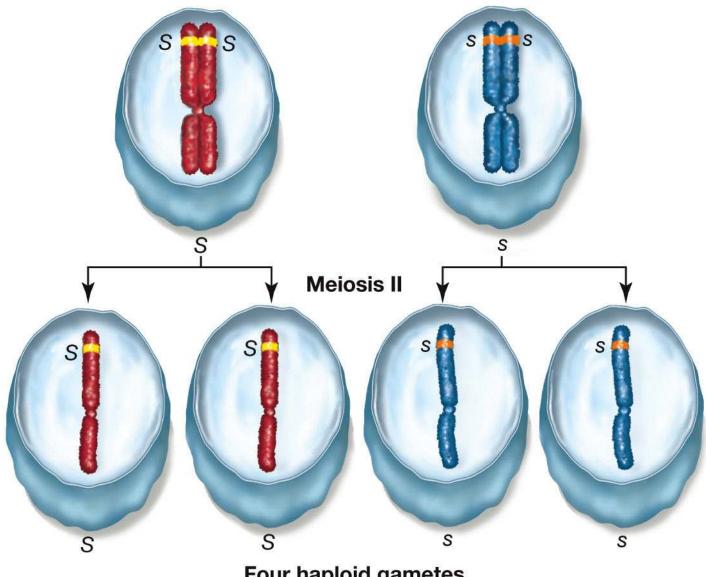


Figure 10.5 Meiosis Accounts for the Segregation of Alleles (Part 2)



Four haploid gametes

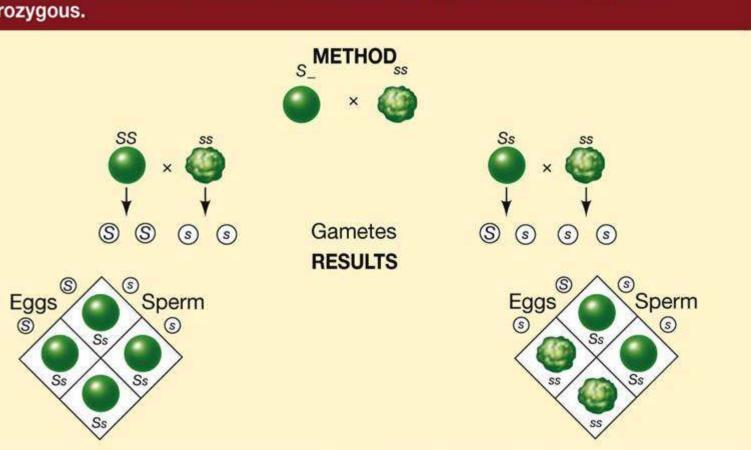
Mendel tested his hypothesis by doing test crosses:

 Determines whether an individual is homozygous or heterozygous for a trait by crossing it with the homozygous recessive.

Mendel crossed the F₁ with known homozygote's (e.g., wrinkled or ss).

EXPERIMENT

HYPOTHESIS: A test cross can reveal whether an organism is homozygous or heterozygous.



CONCLUSION: The plant being tested is homozygous.

CONCLUSION: The plant being tested is heterozygous.

Mendel's next experiment:

Crossing peas that differed in two characters—seed shape and seed color.

True-breeding parents:

SSYY—spherical yellow seeds

ssyy—wrinkled green seeds

F₁ generation is *SsYy*—all spherical yellow.

Crossing (self-pollinating) the F₁ generation is a **dihybrid cross**.

Mendel asked whether, in the gametes produced by SsYy, the traits would be linked, or segregate independently.

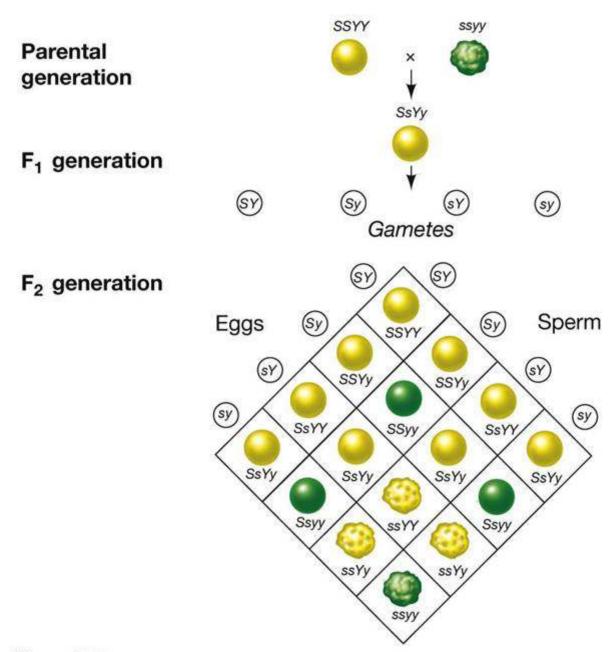
Practice Punnett Squares for Dihybrid Crosses (genetics example)

If linked, gametes would be SY or sy; F₂ would have three times more spherical yellow than wrinkled green.

If independent, gametes could be *SY*, *sy*, *Sy*, or *sY*. F₂ would have nine different genotypes; phenotypes would be in 9:3:3:1 ratio.

Results in **recombinant** phenotypes.

Figure 10.7 Independent Assortment



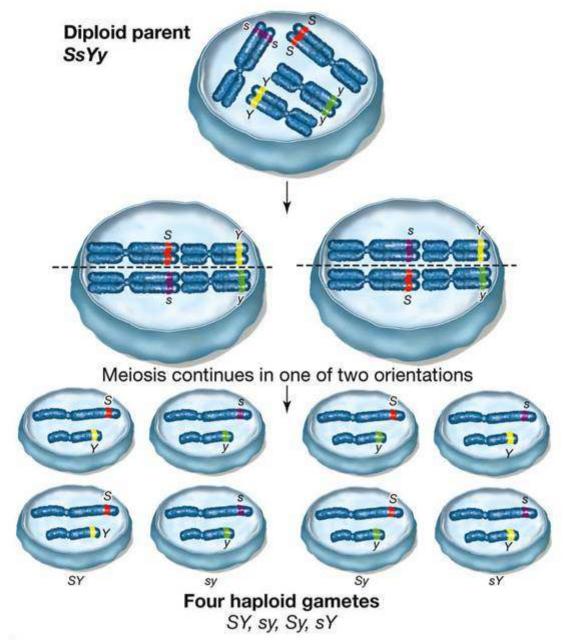
Mendel's second law

The Law of Independent Assortment:

Alleles of different genes assort independently during gamete formation.

Doesn't always apply to genes on the same chromosome; but chromosomes do segregate independently.

Figure 10.8 Meiosis Accounts for Independent Assortment of Alleles



One of Mendel's contributions to genetics was the use of mathematical analyses—the rules of statistics and probability.

Probability:

- If an event is certain to happen, probability = 1
- If an event cannot possibly happen, probability = 0
- All other events have a probability between 0 and 1

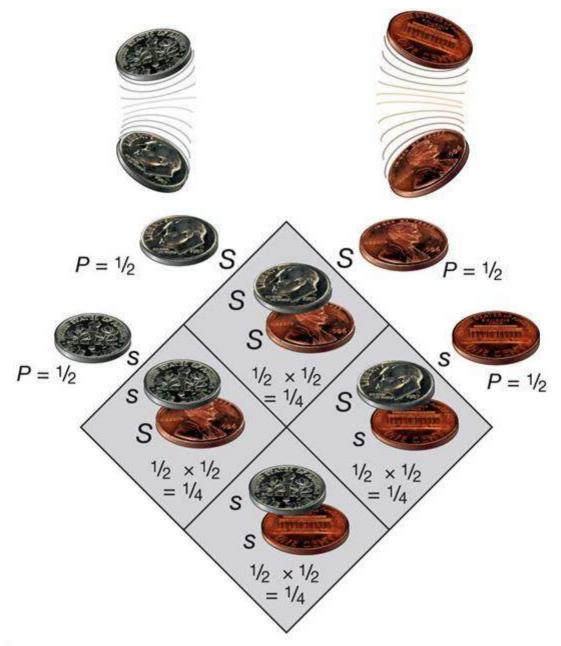
Probability of two independent events happening together: multiply the probabilities of the individual events.

Tossing two coins—probability that both will come up heads:

$$\frac{1}{2} \times \frac{1}{2} = \frac{1}{4}$$

The multiplication rule.

Figure 10.9 Using Probability Calculations in Genetics



The probability of an event that can occur in two different ways is the sum of the individual probabilities.

In F_2 , there are two ways to get a heterozygote; thus $\frac{1}{4} + \frac{1}{4} = \frac{1}{2}$

The addition rule.

Result: 1:2:1 ratio of genotypes; 3:1 ratio of phenotypes

Dihybrid crosses:

Probability that F_2 seed will be spherical is $\frac{3}{4}$ = probability of heterozygote + probability of homozygote or $\frac{1}{2}$ + $\frac{1}{4}$ = $\frac{3}{4}$

Joint probability that a seed will be spherical and yellow: $\frac{3}{4} \times \frac{3}{4} = \frac{9}{16}$

Probability calculations and Punnett squares give the same results.

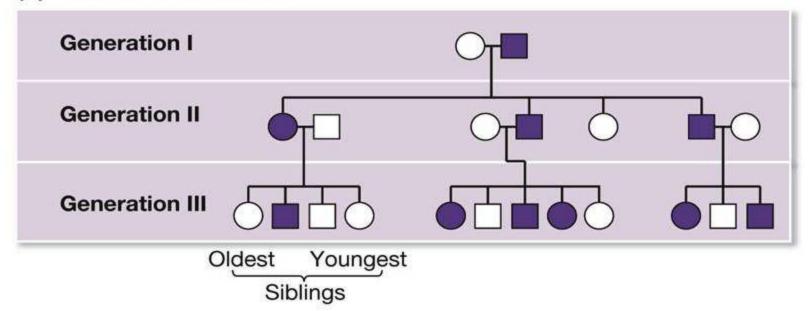
Human **pedigrees** can show Mendel's laws.

Humans have few offspring; pedigrees do not show the clear proportions that the pea plants showed.

Geneticists use pedigrees to determine whether a rare allele is dominant or recessive.

Figure 10.10 Pedigree Analysis and Inheritance (Part 1)

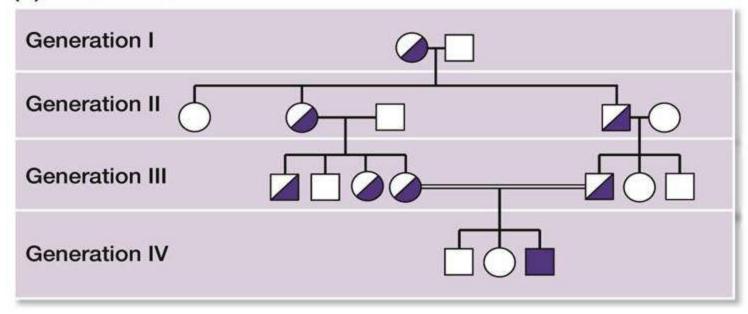
(A) Dominant inheritance



Female	Unaffected	Affected	Heterozygote (unaffected phenotype)
Male			
Mating	\bigcirc		
Mating between relatives	0-0		

Figure 10.10 Pedigree Analysis and Inheritance (Part 2)

(B) Recessive inheritance



Female	Unaffected	Affected	Heterozygote (unaffected phenotype)
Male			
Mating	\bigcirc		
Mating between relatives	0=□		

When there is a rare recessive phenotype in a family, there is usually marriage of relatives.

If a recessive allele is rare in the general population, it is unlikely that two people that marry will both carry it unless they are related (e.g., cousins).

10.2 How Do Alleles Interact?

Different alleles arise through **mutation**: rare, stable, inherited changes in the genetic material.

Wild type: allele present in most of the population. Other alleles are *mutant* alleles.

Locus with wild-type allele present less than 99% of the time is **polymorphic**.

10.2 How Do Alleles Interact?

A given gene may have more than two alleles.

Example: coat color in rabbits

Multiple alleles increase the number of possible phenotypes.

Can you do Genetics?

- In humans, tongue rolling is a dominant trait, those with the recessive condition cannot roll their tongues. Bob can roll his tongue, but his mother could not. He is married to Sally, who cannot roll her tongue. What is the probability that their first born child will not be able to roll his tongue?
- An organism with the genotype AaBbCCdd is crossed with one that is AAbbccDd. What are the odds of having an offspring with the genotype AabbCcDd. In this problem, do not use punnet squares. Solve the problem using math and statistics and make sure you show all work.

Figure 10.11 Inheritance of Coat Color in Rabbits

Possible genotypes	CC, Cc ^{ch} , Cc ^h , Cc	CchCch	cchch, cchc	chch, chc	СС
Phenotype	Dark gray	Chinchilla	Light gray	Point restricted	Albino









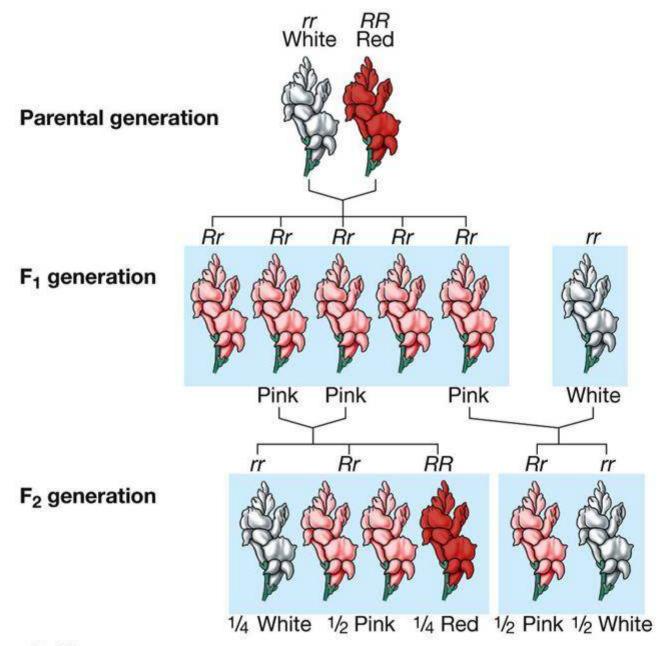


10.2 How Do Alleles Interact?

Some alleles are neither dominant nor recessive—heterozygote has an intermediate phenotype: **incomplete dominance**.

Example: snapdragons

Figure 10.12 Incomplete Dominance Follows Mendel's Laws



10.2 How Do Alleles Interact?

Codominance: two alleles at one locus produce phenotypes that are both present in the heterozygote.

Example: ABO blood group system—three alleles at one locus.

Figure 10.13 ABO Blood Reactions Are Important in Transfusions

Blood type	type mad		Reaction to added antibodies		
of cells	Genotype	body	Anti-A	Anti-B	
Α	IAIA or IAiO	Anti-B	* * * *		
В	I ^B I ^B or I ^B i [○]	Anti-A		***	
AB	 A B	Neither anti-A nor anti-B	* 8 8 8 8 8 8 8 8 8 8 8 8 8 8 8 8 8 8 8	****	
0	i°i°	Both anti-A and anti-B			

10.2 How Do Alleles Interact?

A single allele can have multiple effects: **pleiotropic**.

Example: allele for coloration pattern in Siamese cats; the same allele results in crossed eyes—both result from the same protein.

10.3 How Do Genes Interact?

Epistasis: phenotypic expression of one gene is influenced by another gene.

Example: coat color in Labrador retrievers.

Allele **B** (black) dominant to **b** (brown)

Allele **E** (pigment deposition) is dominant to **e** (no pigment deposition—yellow)

Figure 10.14 Genes May Interact Epistatically



(A) Black labrador (B_E_)



(B) Chocolate labrador (bbE_)



(C) Yellow labrador (_ _ee)

LIFE 8e, Figure 10.14

LIFE: THE SCIENCE OF BIOLOGY, Eighth Edition @ 2007 Sinauer Associates, Inc. and W. H. Freeman & Co.

10.3 How Do Genes Interact?

Inbreeding: mating among close relatives; can result in offspring of low quality.

Close relatives tend to have the same recessive alleles.

10.3 How Do Genes Interact?

A cross between two different truebreeding homozygotes can result in offspring with stronger, larger phenotypes: *hybrid vigor* or **heterosis**.

First discovered with corn by G.H. Shull.

Figure 10.15 Hybrid Vigor in Corn



Parent

Parent

Hybrid offspring

LIFE 8e, Figure 10.15

10.3 How Do Genes Interact?

Environment also affects phenotype.

Light, temperature, nutrition, etc., can affect expression of the genotype.

Siamese cats and certain rabbit breeds enzyme that produces dark fur is inactive at higher temperatures.

Figure 10.16 The Environment Influences Gene Expression



10.3 How Do Genes Interact?

Effects of genes and environment:

- Penetrance: proportion of individuals with a certain genotype that show the phenotype.
- Expressivity: degree to which genotype is expressed in an individual.

10.3 How Do Genes Interact?

Mendel's characters were discrete and qualitative.

For more complex characters, phenotypes vary continuously over a range—quantitative variation, or continuous.

Quantitative variation is usually due to both genes and environment.

Figure 10.17 Quantitative Variation



10.3 How Do Genes Interact?

Genes that determine these complex characters: quantitative trait loci.

Identifying these loci can help improve crop yields, understand disease susceptibility and behavior, etc.

Much genetic research has been done with the fruit fly *Drosophila melanogaster*.

Beginning in 1909 in Thomas Hunt Morgan's lab at Columbia—the "fly room."

Some crosses performed with *Drosophila* did not yield expected ratios according to the law of independent assortment.

Some genes were inherited together; the two loci were on the same chromosome, or linked.

All the loci on a chromosome form a linkage group.

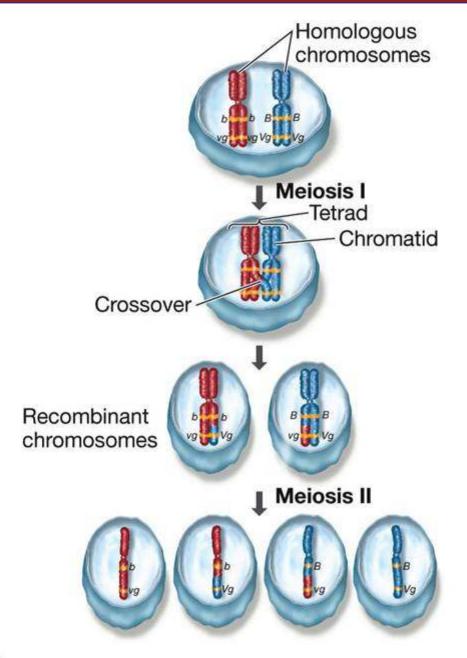
EXPERIMENT HYPOTHESIS: Alleles for different characteristics always assort independently. METHOD Parent BbVgvg bbvgvg Wild type ď RESULTS F₁ Genotypes BbVqvq bbVqvq bbvgvg Bbvqvq Wild type Black vestigial Gray vestigial Black normal 575 575 575 575 Expected phenotypes Observed phenotypes 965 944 206 185 Parental Recombinant phenotypes phenotypes CONCLUSION: The hypothesis is rejected. These two genes do not assort independently, but are linked (on the same chromosome).

Absolute linkage is rare.

Genes may recombine during prophase I of meiosis by crossing over.

Chromosomes exchange corresponding segments. The exchange involves two chromatids in the tetrad; both chromatids become recombinant.

Figure 10.19 Crossing Over Results in Genetic Recombination

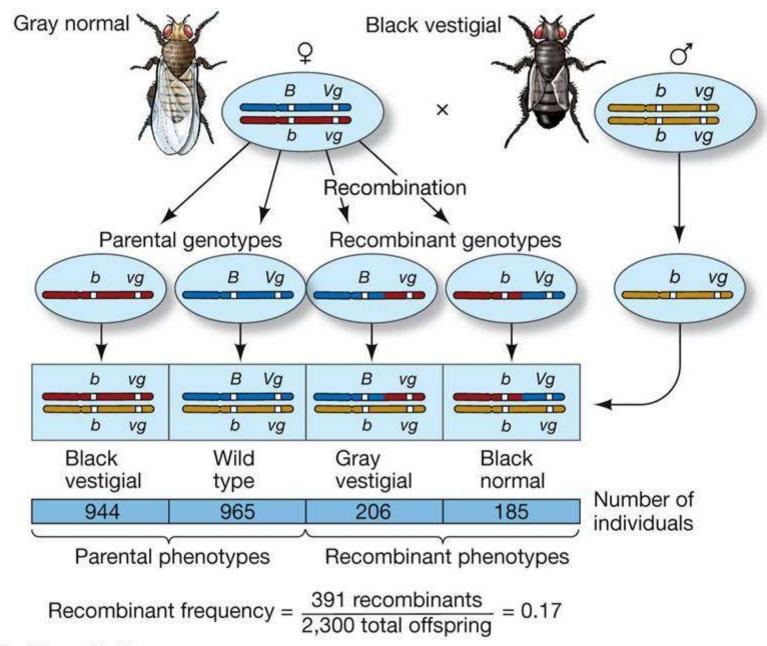


Recombinant offspring phenotypes (non-parental) appear in **recombinant frequencies**:

Divide number of recombinant offspring by total number of offspring.

Recombinant frequencies are greater for loci that are farther apart.

Figure 10.20 Recombinant Frequencies



Recombinant frequencies can be used to make **genetic maps** showing the arrangement of genes along a chromosome.

Distance between genes = map unit = recombinant frequency of 0.01.

Map unit also called a centimorgan (cM)

Figure 10.21 Steps toward a Genetic Map

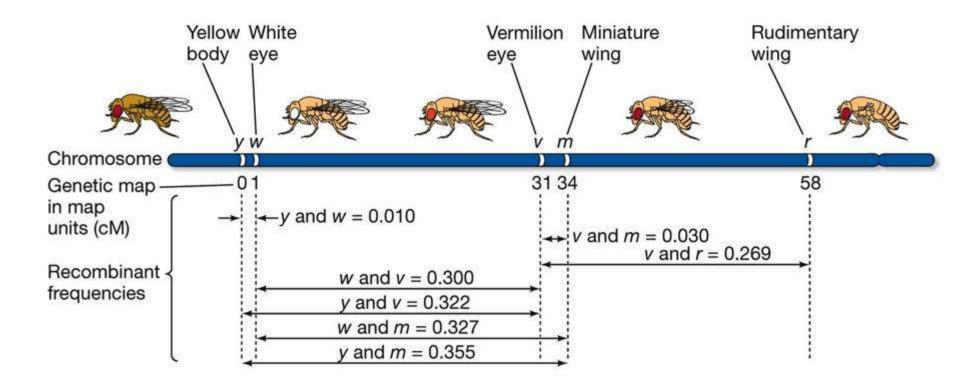


Figure 10.22 Map These Genes (Part 1)

			a of the indivi possible sequ			
	а		b		С	
	а				c b	
	b	а			С	
with a g	enotype A	AaBb. We tes	bb, and obta st cross these types of the fi	e AaBb indi	vidual	s
			b, 50 <i>Aabb</i> , a (recombin		Ç.	

Figure 10.22 Map These Genes (Part 2)

2 How far apart are the a and b genes?

What is the recombinant frequency? Which are the recombinant types, and which are the parental types?

Recombinant frequency (a to b) = (50 + 50)/1,000 = 0.1So the map distance is

Map distance = $100 \times \text{recombinant frequency} = 100 \times 0.1 = 10 \text{ cM}$

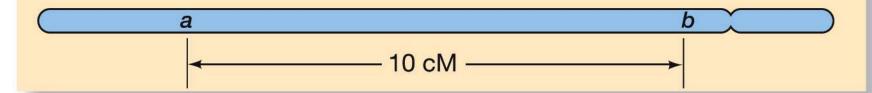


Figure 10.22 Map These Genes (Part 3)

3 How far apart are the a and c genes?

Now we make a cross $AACC \times aacc$, obtain an F_1 generation, and test cross it, obtaining

460 AaCc, 460 aacc, 40 Aacc, and 40 aaCc

Recombinant frequency (a to c) = (40 + 40)/1,000 = 0.08

Map distance = $100 \times \text{recombinant frequency} = 100 \times 0.08 = 8 \text{ cM}$

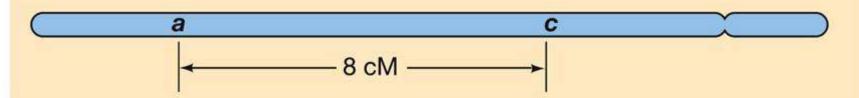


Figure 10.22 Map These Genes (Part 4)

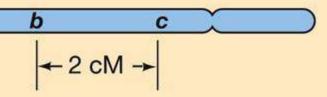
4 How far apart are the b and c genes?

We make a cross $BBCC \times bbcc$, obtain an F_1 generation, and test cross it, obtaining

490 BbCc, 490 bbcc, 10 Bbcc, and 10 bbCc

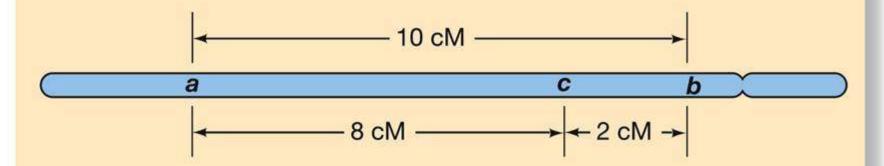
Recombinant frequency (b to c) = (10 + 10)/1,000 = 0.02

Map distance = $100 \times \text{recombinant frequency} = 100 \times 0.02 = 2 \text{ cM}$



Which of the three genes is between the other two?

Because a and b are the farthest apart, c must be between them.



These numbers add up perfectly. In most real cases, they will not add up perfectly because of multiple crossovers.

Sex determination varies among species.

Corn: each adult produces both male and female gametes—*monoecious* ("one house").

Some plants and most animals are dioecious ("two houses")—male and female gametes produced by different individuals.

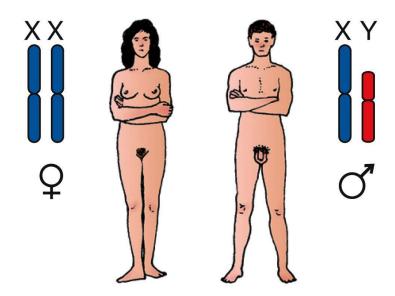
In most dioecious organisms, sex is determined by differences in the chromosomes.

Many animals have a pair of sex chromosomes; all others are autosomes.

Mammals:

Female has two X chromosomes.

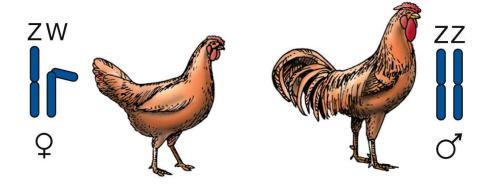
Male has one X and one Y.



Birds:

Females have one Z and one W.

Males have two Z chromosomes.



Nondisjunction in meiosis:

Pair of sister chromosomes fail to separate in meiosis I, or

Pair of sister chromatids fail to separate in meiosis II.

Result is an euploidy.

If there is nondisjunction of sex chromosomes, possible outcomes:

XO—the individual has only one sex chromosome—*Turner syndrome*.

XXY—Klinefelter syndrome.

The SRY gene—(sex-determining region on the Y chromosome) encodes a protein involved in primary sex determination.

If SRY protein is present, the embryo develops testes.

If there is no SRY, the embryo develops ovaries.

A gene on the X chromosome, *DAX1*, produces an anti-testis factor.

SRY in males inhibits the DAX1 maleness inhibitor.

Secondary sex determination results in outward characteristics of each sex—not determined directly by presence or absence of Y chromosome.

Determined by genes scattered on all the chromosomes that control hormones.

Genes on sex chromosomes don't follow Mendelian patterns.

The Y chromosome carries few genes; the X carries many.

Thus, males have only one copy of these genes—hemizygous.

Sex-linked inheritance—governed by loci on the sex chromosomes.

Example: eye color in *Drosophila*

Figure 10.23 Eye Color Is a Sex-Linked Trait in *Drosophila* (Part 1)

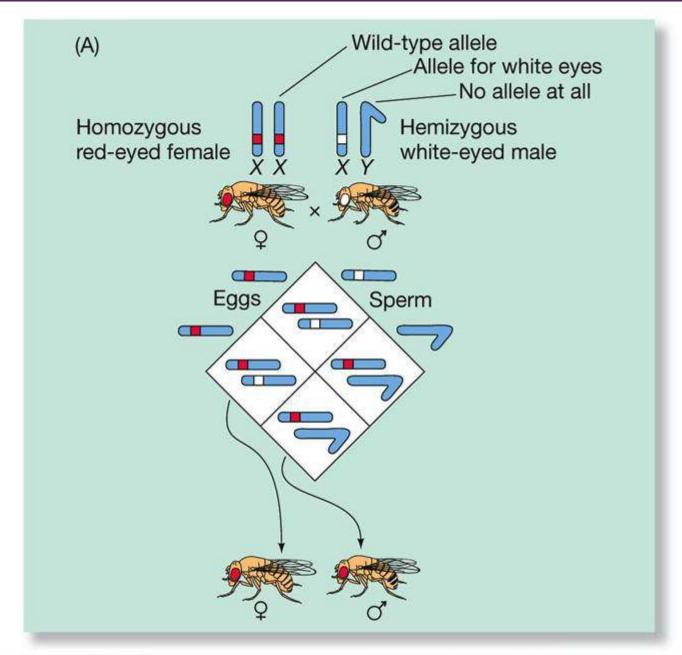
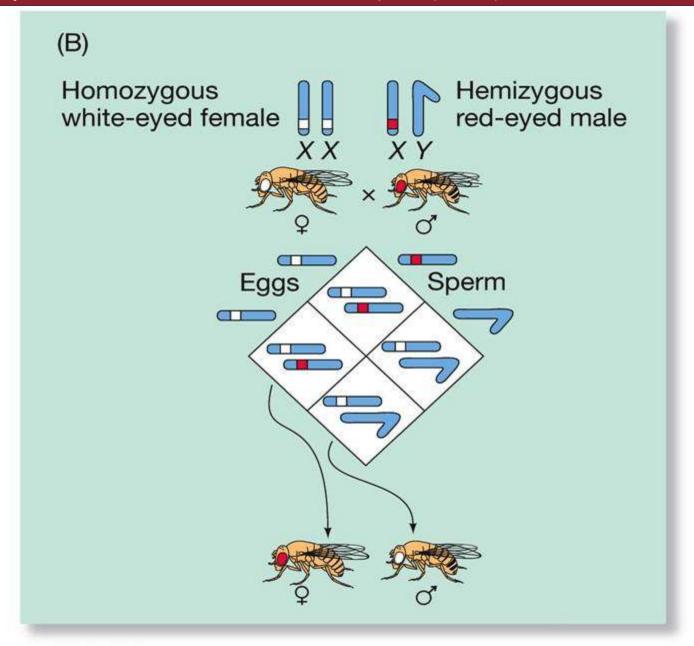


Figure 10.23 Eye Color Is a Sex-Linked Trait in *Drosophila* (Part 2)

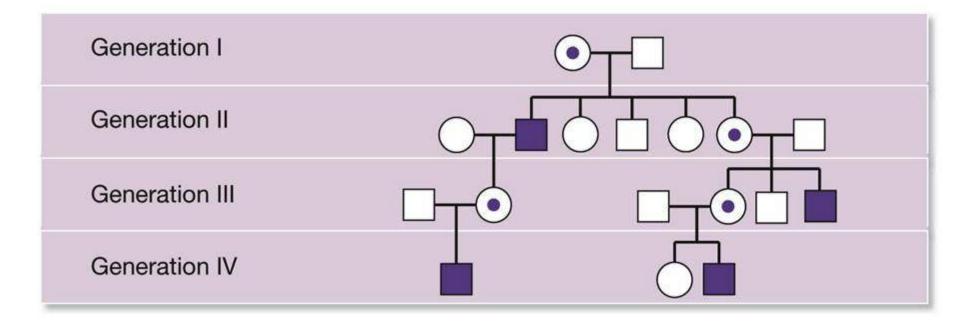


X-linked recessive phenotypes:

- Appear much more often in males than females
- Daughters who are heterozygous are carriers
- Mutant phenotype can skip a generation if it passes from a male to his daughter

Figure 10.24 Red-Green Color Blindness Is a Sex-Linked Trait in Humans

• Female who carries gene for phenotype of interest on one X chromosome



10.5 What Are the Effects of Genes Outside the Nucleus?

Mitochondria and plastids contain small numbers of genes—important in organelle assembly and function.

Mitochondria and plastids are inherited from the mother.

There may be hundreds of mitochondria or plastids in a cell.

10.5 What Are the Effects of Genes Outside the Nucleus?

Organelle genes tend to mutate faster than nuclear genes—multiple alleles.

Some plastid gene mutations affect chlorophyll synthesis, resulting in a white phenotype.

Mitochondria gene mutations may affect ATP production, especially in tissues with high energy requirements.