

10: Genetics: Mendel and Beyond

The Foundation of Genetics

- Five thousand years ago or earlier, people were using applied genetics in the form of plant and animal breeding.
- The foundation for the science of genetics was laid in 1866, when Gregor Mendel used varieties of peas to conduct experiments on inheritance.
- Mendel's research was ignored until the turn of the twentieth century.

Plant breeders showed that both parents contribute equally to inheritance

- Plants have some desirable characteristics for genetic studies.
 - Plants can be grown in large quantities.
 - They produce large numbers of offspring.
 - Many have both male and female reproductive organs in the same plant, which allows self-fertilization.
 - The generation time is relatively short.
 - It is easy to control which individuals mate.
- Josef Gottlieb Kölreuter made a few observations that Mendel later found useful.
 - Kölreuter's research focused on the relative contribution of males and females to the genetics of plants.
 - Reciprocal crosses helped prove that both male and female parents contribute equally to the characteristics inherited by offspring.
 - An example is a cross of a male white plant with a female pink plant, and the reciprocal cross of a male pink with a female white plant.
 - The resulting progeny have the same appearance.
 - Before the acceptance of Mendel's research, the concept of blending was favored. An example of this form of inheritance would be purple progeny resulting from red and blue parents. As a consequence of blending, the purple color would be forever blended and could not be separated.

Mendel's discoveries were overlooked for decades

- Gregor Mendel was a monk with scientific training in mathematics, physics, and biology.
- Mendel presented his nine-year-long project orally in 1865 and in writing in 1866. His data challenged the blending concept, but he was ahead of his time.
- His theory was ignored, perhaps because his biological peers were not accustomed to reviewing mathematical data. Even Darwin, whose evolutionary theory rests on genetic variation among individuals, failed to understand Mendel's point and relied on the blending concept.
- In 1900, Hugo de Vries, Karl Correns, and Erich von Tschermak each independently published papers on the quantitative outcomes from crosses. Each cited Mendel's then rediscovered 1866 paper. By 1900, meiosis had been described and Mendel's ideas were finally accepted.

Mendel's Experiments and the Laws of Inheritance

Mendel devised a careful research plan

- Mendel chose garden peas as his subjects as they are easily grown and their pollination is easily controlled.
- He controlled pollination by manually moving pollen between plants. (See Figure 10.1.)
- He could also allow the plants to self-pollinate.

- Mendel examined varieties of peas for heritable characters and traits for his study.
 - A character is a feature, such as flower color. (See Table 10.1.)
 - A trait is a particular form of a character, such as white flowers.
 - "Heritable" means the trait can be passed from parent to progeny.
- Mendel looked for characters that had well-defined alternative traits and that were true breeding.
 - A trait is true breeding when it is the only trait that occurs through many generations of breeding individuals that share that trait.
 - A true-breeding white-flowered plant would have only white-flowered progeny when crossed with others in its strain.
- True-breeding plants, when used for crossing with other plants that have an alternative trait, are called the parental generation, designated P.
- The progeny from the cross of the P parents are called the first filial generation, designated F₁.
- When F₁ individuals are crossed to each other or self-fertilized, their progeny are designated F₂.
- Mendel's well-organized plan allowed him to observe and record the traits of each generation in sufficient quantity to explain through analysis the relative proportions of the kinds of progeny.
- His paper is recognized today as a model of clarity.

Mendel's Experiment 1 examined a monohybrid cross

- Mendel crossed true-breeding plants that differed for a given character.
 - A monohybrid cross involves one (mono) character and different (hybrid) traits.
 - Pollen from true-breeding pea plants with wrinkled seeds (one trait) was placed on stigmas of true-breeding plants with spherical seeds (another trait). (See Figure 10.2.)
 - The F₁ seeds were all spherical; the wrinkled trait failed to appear at all.
 - Because the spherical trait completely masks the wrinkled trait when true-breeding plants are crossed, the spherical trait is called *dominant*, and the wrinkled trait is called *recessive*.
 - The F₁ plants were allowed to self-pollinate.
 - This step was the monohybrid cross.
 - *This is also called an F₁ cross.*
 - *Self-pollination is sometimes called selfing.*
 - The progeny, called F₂, were examined: 5,474 were spherical and 1,850 were wrinkled. (See Figure 10.3.)
- Mendel proposed that the units responsible for inheritance were discrete particles.
 - They existed within an organism in pairs, separated during gamete formation, and retained their integrity.
 - This is called the particulate theory, which is in sharp contrast to the blending theory.
- Each pea has two units of inheritance for each character. (See Figure 10.4.)
- During production of gametes, only one of the pair members for a given character passes to the gamete.
- When fertilization occurs, the zygote gets one from each parent, restoring the pair.
- Mendel's units of inheritance are now called genes.
- Different forms of a gene are called alleles.
 - Each allele is given a symbol.

- In the case of wrinkled seeds, *S* might represent smooth and *s* wrinkled. By convention, uppercase *S* represents the dominant; lowercase *s* represents the recessive.
- True-breeding individuals would have two copies of the same allele.
 - Wrinkled would be *ss*. (two copies of same allele = *homozygous*)
 - Smooth true-breeding would be *SS*. (two copies of same allele = *homozygous*)
- Some smooth-seeded plants could be *Ss*, although they would *not* be true-breeding.
- Individuals that are smooth, but had a wrinkled parent, are *heterozygous*: *Ss*.
- When an organism is studied for three different genes and has the alleles *AABbCC*, it is homozygous for *A* and *C* genes but heterozygous for the *B* gene.
- The physical appearance of an organism is its *phenotype*. Wrinkled-seed would be a phenotype.
- The actual composition of the organism's alleles for a gene is its *genotype*: *Ss* is a genotype.
- *Organisms have many different genes—some have thousands, and complex organisms have 10 times that number. Most of these genes are yet to be described in terms of the DNA sequence or the amino acid sequence of the gene product.*

Mendel's first law says that alleles segregate

- When an individual produces gametes, alleles separate, so each gamete receives one member of the pair of alleles.
- This is Mendel's first law, the law of segregation.
- When fertilization occurs, pairs are reestablished by receiving one copy from each parent.
- The Punnett square is a simple box-like device that helps us to consider all genetic combinations and can provide clarity by showing the expected frequencies of genotypes. (Figure 10.4 includes a simple Punnett square.)
 - The *S* and *s* symbols represent the single allele each gamete receives.
 - Fertilization provides the two alleles for the new individual, one from the male (sperm) and one from the female (egg).
- The Punnett square shows that the genotypes and associated ratios for a monohybrid cross are 1 *SS*:2 *Ss*:1 *ss*.
- Any progeny with an *S* would have the dominant (smooth) phenotype, so the phenotypic ratio is 3 smooth to 1 wrinkled.
- Now it is known that a gene is a portion of the chromosomal DNA that resides at a particular site, called a *locus* (plural is *loci*).
- The gene codes for a particular function.
- Mendel arrived at the law of segregation with no knowledge of meiosis or chromosomes. The mechanism of chromosome separation in meiosis I today explains his law of segregation. (See Figure 10.5.)

Mendel verified his hypothesis by performing a test cross

- A test cross can determine the genotype (*heterozygous* or *homozygous*) of an individual with a dominant trait. (See Figure 10.6.)
- It involves crossing the individual to a true-breeding recessive (homozygous recessive).
 - If the unknown is heterozygous, approximately half the progeny will have the dominant trait and half the recessive trait.
 - If the unknown is homozygous dominant, all the progeny will have the dominant trait.

Mendel's second law says that alleles of different genes assort independently

- The second law describes the outcome of dihybrid (two character) crosses, or hybrid crosses involving additional characters.
- A dihybrid is an individual that is a double heterozygote (e.g., with the genotype $SsYy$).
- Mendel's second law states that the Ss alleles assort into gametes independently of the Yy alleles.
- The dihybrid, $SsYy$, produces gametes that have one allele of each gene. (See Figure 10.7.)
- Four different gametes are possible and will be produced in equal proportions: SY , Sy , sY , and sy .
- Random fertilization of gametes yields the outcome visible in the Punnett square of Figure 10.7. Note its 4×4 table construction to accommodate 16 possible phenotypes.
- Filling in the table and adding the like cells reveals a 9:3:3:1 ratio of the four possible phenotypes (smooth yellow, smooth green, wrinkled yellow, and wrinkled green).
- The law of independent assortment states that alleles of different genes assort independently of one another during gamete formation. (See Figure 10.8.)
 - In fact, this law is not always true.
 - The law of independent assortment is accurate for genes that are on separate chromosomes, but not necessarily for genes that are on the same chromosome.
 - *Genes that are close to each other on the same chromosome tend to stay together, but crossing over during meiosis may separate them.*
 - *The closer together on the same chromosome genes are, the more they tend to stay together.*

Punnett squares or probability calculations: A choice of methods

- Multiplying probabilities:
 - If two coins, a penny and a dime, are tossed, each acts independently of the other.
 - The probability of both landing on heads would be $1/2 \times 1/2 = 1/4$.
 - To find the probability that independent events will *both* happen, the general rule is to multiply the probabilities of the individual events. (See Figure 10.9.)
- Monohybrid cross probabilities:
 - In the example of smooth and wrinkled seeds, heterozygotes produce S and s gametes.
 - The probability of a gamete being S is $1/2$.
 - The probability of that an F_2 plant will be SS is $1/2 \times 1/2 = 1/4$.
- Adding probabilities:
 - The probability of an event that can occur in two or more ways is the sum of the probabilities for each way in which the event can occur.
 - For example, the genotype Ss can result from s in the egg and S in the sperm, or from S in the egg and s in the sperm. Thus the probability of heterozygotes in the F_2 generation of a monohybrid cross is $1/4 + 1/4 = 1/2$.
- The dihybrid cross:
 - To calculate the probabilities of the outcomes of dihybrid crosses, simply multiply the outcomes from each of the individual monohybrid components.
 - For example, the probability of the $SSYy$ genotype can be calculated as follows: An F_1 (dihybrid) cross of $SsYy$ generates $1/4 SS$, $1/2 Ss$, $1/4 ss$, and $1/4 YY$, $1/2 Yy$, $1/4 yy$. The probability of the $SSYy$ genotype is the probability of the SS genotype, which is $1/4$, times the probability of the Yy genotype, which is $1/2$. This would be $1/8$ ($1/4 \times 1/2$).

Mendel's laws can be observed in human pedigrees

- Patterns for over 2,500 inherited human characteristics have so far been determined.
- Humans cannot be studied using planned crosses, so human geneticists rely on pedigrees, which show phenotype segregation in several generations of related individuals. (See Figure 10.11.)
- Since humans have such small numbers of offspring, human pedigrees do not show clear proportions.
- The actual number of affected versus unaffected offspring is impossible to predict for a certain couple because outcomes for small samples fail to follow closely the expected outcomes.
- If neither parent has a given phenotype, but it shows up in progeny, the trait is recessive and the parents are heterozygous. The chance of other children getting the trait is 1/4.
- Half of the children from such a cross will be carriers (heterozygous for the trait).
- The probability of a carrier (heterozygote) for a rare allele unknowingly marrying another unrelated carrier is quite low.
- See Figure 10.10 for a pedigree analysis of a dominant allele. For a dominant allele,
 - *Every* affected person has an affected parent.
 - About half of the offspring of an affected person are also affected (assuming only one parent is affected).
 - The phenotype occurs equally in both sexes.
- See Figure 10.11 for a pedigree analysis of a recessive allele. For a rare recessive trait
 - Affected people usually have parents who are both not affected.
 - One-quarter of the children, on average, of unaffected parents would be affected.
 - The phenotype occurs equally in both sexes.
- Marriage between close relatives results in a higher likelihood that both parents will be carriers of a rare allele and produce affected children.
- The major use of pedigree analysis is in clinical evaluation and counseling of patients with inherited abnormalities.

Alleles and Their Interactions

- Differences in alleles of genes are slight differences in the DNA sequence at the same locus, which result in slightly different protein products.
- Some alleles are not simply dominant or recessive. There may be many alleles for a single character or a single allele may have multiple phenotypic effects.

New alleles arise by mutation

- Different alleles exist because any gene is subject to *mutation*, or change, to a stable, heritable new form.
- Alleles can randomly mutate to become a different allele depending on DNA sequence changes.
- *Wild type* is a term used for the most common allele in the population.
- Other alleles, often called mutant alleles, may produce a phenotype different from that of the wild-type allele.
- A genetic locus is called *polymorphic* if the wild-type allele has a frequency less than 99% in a population (that is, if more than 1% of the alleles at that locus are mutant alleles).

Many genes have multiple alleles

- A population might have more than two alleles for a given gene.
- *The ABO blood types are an example of multiple alleles.*

- In rabbits, coat color is determined by one gene with four different alleles. Five different colors result from the combinations of these alleles. (See Figure 10.12.)
- Even if more than two alleles exist in a population, any given individual can have no more than two of them: one from the mother and one from the father.

Dominance is usually not complete

- Heterozygotes may show an intermediate phenotype.
- For example, red-flowered snapdragons when crossed with white will generate pink-flowered plants. (See Figure 10.13.)
- This phenotype might seem to support the blending theory. (The blending theory predicts pink F₁ progeny.)
- The F₂ progeny, however, demonstrate Mendelian genetics. When the F₁ pink individuals self-fertilize, the F₂ progeny have a phenotypic ratio of 1 red:2 pink:1 white. (The blending theory predicts all pink F₂ progeny.)
- This mode of inheritance is called incomplete dominance.
- The phenotypic outcomes for snapdragon flower color and incomplete dominance in general can be explained biochemically.
 - One allele of the gene codes for an enzyme that functions in the production of the red color.
 - The enzyme coded by the other allele is not functional in pigment production.
 - The red-flowered plants have two functional copies of the gene and produce enough enzyme to make red flowers.
 - The pink-flowered plants have one functional allele, just enough to create a pink color.
 - Neither allele in white-flowered plants is capable of making the functional enzyme, so the resulting flowers are white.
- Mendel's laws are not compromised here, as is shown in Figure 10.13. He just happened to find in peas examples of complete dominance only.

In codominance, both alleles are expressed

- In codominance, two different alleles for a gene produce two different phenotypes in the heterozygotes. The AB of the human ABO blood group system is an example.
- The alleles for blood type are I^A , I^B and I^O . They all occupy one locus.
- These alleles determine which antigens (proteins) are present on the surface of red blood cells. (See Figure 10.14. The Instructor's Resource CD-ROM includes a photograph illustrating the ABO blood reactions.)
- These antigens react with proteins called antibodies in the serum of certain individuals. The result is red blood cell agglutination, or clumping, which may be fatal for those individuals.
 - Individuals with two I^A , or with I^A and I^O , are type A.
 - Individuals with two I^B , or with I^B and I^O , are type B.
 - Individuals with I^A and I^B are type AB.
 - Individuals with I^O and I^O are type O.
- Both I^A and I^B are expressed when present, and both produce an antigen.
 - This is why they are called codominant.
- I^O is a recessive trait and is the absence of either the A or B antigen.

Some alleles have multiple phenotypic effects

- Some single alleles have more than one distinguishable phenotypic effect.
 - This is called pleiotropy.

- An example is the coloration pattern and crossed eyes of Siamese cats, which are both caused by the same allele. These unrelated characters are caused by the same protein produced by the same allele.

Gene Interactions

Some genes alter the effects of other genes

- Epistasis occurs when the alleles of one gene cover up or alter the expression of alleles of another gene.
- An example is coat color in mice. (See Figure 10.15.)
 - The *B* allele determines a banded pattern, called agouti.
 - The recessive *b* allele results in unbanded hairs.
 - The genotypes *BB* or *Bb* are agouti. The genotype *bb* is solid colored (black).
 - Another locus determines if any coloration occurs. The genotypes *AA* and *Aa* have color and *aa* are albino. The *aa* genotype blocks *all* pigment production.
 - The mice heterozygous for both genes are agouti.
 - The F_2 phenotypic ratio is 9 agouti:3 black:4 white.
 - The corresponding genotypes are 9 agouti (1 *BBAA* + 2 *BbAA* + 4 *BbAa*):3 black (1 *bbAA* + 2 *bbAa*):4 albino (1 *BBaa* + 2 *Bbaa* + 1 *bbaa*).
- *As another example of epistasis, imagine that when a true-breeding white-flowered plant is crossed to a true-breeding purple-flowered plant, the F_1 are all purple.*
- *When the purple F_1 plants are self-crossed, 9 purple for every 7 white are observed in the F_2 progeny.*
 - *This ratio is different from what would be expected if purple were simply dominant to white.*
 - *The ratio provides a clue to the relationship of two different genes (A and B) necessary to create the purple pigment in this plant.*
 - *Suppose the gene at locus A has two alleles: A, which is dominant and codes for enzyme A, and a, which is recessive and codes for a nonfunctional enzyme.*
 - *Suppose also that the gene at locus B has two alleles that follow the same pattern: B is dominant and codes for enzyme B, and b is recessive and codes for a nonfunctional enzyme.*
 - *The following biochemical pathway for production of purple pigment could explain the ratio in the dihybrid cross:*

colorless precursor	enzyme A □	colorless intermediate	enzyme B □	purple pigment
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- *Dominant alleles are necessary at both the A and B loci to produce purple pigment. Both enzyme reactions, A and B, must take place. Such genes are called complementary genes.*
- *A Punnett square for the dihybrid cross, with Xs drawn through the boxes of offspring that cannot produce pigment, shows clearly the 9 purple:7 white ratio.*

Hybrid vigor results from new gene combinations and interactions

- For centuries, it has been known that when two homozygous strains of plants or animals are crossed, the offspring are phenotypically stronger, larger, and more vigorous than either parent.
- In the early twentieth century, G. H. Shull crossed two varieties of corn, and the yield went from 20 to 80 bushels per acre. (See Figure 10.16.)
- This is called either hybrid vigor or heterosis. Hybridization is now a common agricultural practice to increase production in plants.

- A hypothesis called overdominance proposes that the heterozygous condition in certain important genes is superior to either homozygote.

Polygenes mediate quantitative inheritance

- Individual heritable characters are often found to be controlled by groups of several genes, called polygenes.
- Each allele intensifies or diminishes the phenotype.
- Variation is continuous or quantitative (“adding up”).
- Examples of continuous characters are height, skin color, and possibly intelligence. (See Figure 10.17.)

The environment affects gene action

- Genotype and environment interact to determine the phenotype of an organism.
- Variables such as light, temperature, and nutrition can affect the translation of genotype into phenotype.
- For example, the darkness of the fur on extremities of a Siamese cat is affected by the temperature of that region. Darkened extremities normally have a lower temperature than the rest of the body. The coloration can be manipulated experimentally.
- The proportion of individuals in a group with a given genotype that express the corresponding phenotype can sometimes be measured, and the measure is called *penetrance*.
- The *expressivity* of the genotype is the degree to which it is expressed in an individual.
 - *An example is hereditary hemochromatosis.*
 - *This disease causes abnormally high levels of iron to accumulate in the liver and other organs of affected people.*
 - *Some people with the disease accumulate toxic levels of iron, and others accumulate levels just above normal.*
- The influence of environment on genotype or phenotype can be studied with identical twins, especially when they are separated from birth and reared apart in substantially different environments.

Genes and Chromosomes

- How do we determine the order and distance between the genes that are located on the same chromosome?
- A system was first developed in Thomas Hunt Morgan's fly lab in 1909.
- The biological model used was the fruit fly, *Drosophila melanogaster*, which is still used today in chromosomal studies.

Linked genes are on the same chromosome

- Mendel's second law, independent assortment, failed to be universal.
- One early exception was found in *Drosophila* when crossing flies that were hybrids for two particular alleles (body color and wing size) with flies that were recessive for both alleles (a test cross).
- The results were not the expected 1:1:1:1, but instead, two of the genotypes occurred at a frequency higher than the other two. (See Figure 10.19.)
- These results make sense if the two loci are on the same chromosome, and thus their inheritance is linked. All the loci on a given chromosome make up a linkage group. Absolute or total linkage of all loci is extremely rare, however.
- *Drosophila have just 3 pairs of autosomal chromosomes and one pair of sex chromosomes. The normal fly is diploid and has 8 chromosomes.*

- The fruit fly has thousands of genes on just 4 pairs of chromosomes. Therefore, many exist together on the same chromosome.

Genes can be exchanged between chromatids

- See Figure 9.16 for a review of the recombination of chromosomal segments.
- When two homologous chromosomes physically exchange corresponding segments during prophase I of meiosis, geneticists call it crossing over.
- Recombinations occur at chiasmata. (See Figure 10.21.)
- If just a few exchanges occur, genes that are closer together tend to stay together.
- The farther apart on the same chromosome genes are, the more likely they will separate during recombination.
- The two extremes are independent assortment and complete or absolute linkage. (See Figure 10.20.)
- The progeny resulting from crossing over appear in repeatable proportions, called the recombinant frequency. (See Figure 10.22.)
- Greater recombination frequencies are observed for genes that are farther apart on the chromosomes because a chiasma is more likely to cut between genes that are far apart than genes that are closer together.

Geneticists make maps of eukaryotic chromosomes

- Alfred Sturtevant, an undergraduate student working in Morgan's fly room, resolved the puzzling question of the deviation of results from the expected ratios by suggesting that as the distance between two genes on a chromosome increases, so does the likelihood that they will separate and recombine.
- The closer loci are on a chromosome, the less likely they will separate and recombine in meiosis.
- A map unit is a recombination frequency of 0.01 (or a 1% recombination). It is also referred to as a centimorgan (cM). (See Figures 10.23 and 10.24.)

Sex Determination and Sex-Linked Inheritance

- Sometimes parental origin of a chromosome does matter.
- Reciprocal crosses give identical results when organisms are diploid.
- *Many organisms have homologous pairs of all chromosomes except for those that determine sex. The homologous pairs are called autosomes; the unpaired X and Y chromosomes are called sex chromosomes.*

Sex is determined in different ways in different species

- In corn, every diploid adult has both male and female reproductive structures. The same is true for peas.
- This type of organism is called monoecious (“one house”).
- Other plants and animals, which have individuals that are one or the other sex, are called dioecious (“two houses”).
- In most dioecious organisms, sex is determined by differences in the chromosomes.
- In honeybees, eggs either are fertilized with a sperm and become diploid females, or they are not fertilized and become haploid males, called *drones*.
- Female grasshoppers have two X chromosomes, and males have just one. The sperm determines the sex of the zygote. If a sperm without an X fertilizes an egg, the zygote becomes a male grasshopper.
- Humans have different sex chromosomes, X and Y. Males have X and Y; females have X and X.
- Sex of the offspring is determined by the sperm.

- Females do not have a Y chromosome, so all normal female gametes (eggs) have one X chromosome.
- Males have one X chromosome and one Y chromosome, so half of the male gametes (sperm) have an X chromosome and the other half have a Y chromosome.
- If a sperm with an X chromosome reaches the egg, the resulting offspring will be female (XX). If a sperm with a Y chromosome reaches the egg, the resulting offspring will be male (XY).
- In humans, maleness is determined by the presence of the Y chromosome.
- Those with XO condition, Turner syndrome, a somewhat rare chromosomal anomaly, are female.
- Those with Klinefelter syndrome, another anomaly, are male and have the XXY condition.

The X and Y chromosomes have different functions

- The gene that determines maleness was identified by studying people with chromosomal abnormalities.
- Some XY females were found.
- Some XX males were found.
- The XY females had a piece missing from the Y, and the XX males had a piece of a Y on one X.
- The fragment missing from the Y chromosome in XY females or present on the X chromosome in XX males contained the maleness-determining gene.
- The gene was named *SRY* (for *sex-determining region* on the *Y* chromosome).
- The *SRY* gene codes for a functional protein involved in primary sex determination.
- If this protein is present, testes develop; if not, ovaries develop.
- A gene on the X chromosome called *DAXI* produces an anti-testis factor. The *SRY* gene product in a male inhibits the gene *DAXI*, and no maleness inhibitor is made.
- Secondary sex traits like breast development, body hair, and voice are influenced by hormones such as testosterone and estrogen.
- *Drosophila* chromosomes follow the same pattern as humans, but the mechanism is different.
- The males are XY, and females XX.
- The ratio of X chromosomes to the autosomal sets determines sex.
- Two X chromosomes for each diploid set yield females.
- One X for each diploid set yields males. (XO is sterile; XY is fertile).
- Birds, moths, and butterflies have XX males and XY females. These are called ZZ males and ZW females to help prevent confusion.
- In these organisms, the egg rather than the sperm determines the sex of the offspring.

Genes on sex chromosomes are inherited in special ways

- The Y carries very few genes (about 20 are known), but the X carries a great variety of characters.
- Females with XX are diploid for X-linked genes; males with XY are haploid. This partial haploid condition of sex chromosomes for males is called hemizygous.
- This generates a special type of inheritance called sex-linked inheritance.
- See Figure 10.25 for a description of Morgan's early research on eye color inheritance in *Drosophila*.

Human beings display many sex-linked characters

- The human X chromosome carries thousands of genes.

- *The probability for a male of having an X-linked genetic disease caused by a mutant recessive allele is much higher than it is for a female.*
- *Barring inbreeding, the probability of a woman having a recessive X-linked genetic disease is the square of frequency of the disease-causing allele. (That is the probability that she would inherit the gene from her mother times the probability that she would inherit it from her father.)*
- *Because men have only one X chromosome, and they express what they get whether it is dominant or recessive, the probability for a man of having an X-linked recessive genetic disease is simply the frequency of the allele for the disease in the population.*
- *The number of lethal or severely detrimental genes on the X is kept low by the hemizygous state of the males.*
- Pedigree analysis of X-linked recessive phenotypes reveal certain patterns:
- The phenotype appears much more often in males than in females.
- A male with the mutation can pass it on only to his daughters, through an X-bearing sperm; his sons get his Y chromosome, which does not carry the trait.
- Daughters who receive one mutant X are heterozygous carriers. They pass the allele to approximately half of their sons and daughters.
- The mutant phenotype can skip a generation if the mutation is passed from a male to his daughter and then to her son.
- The most common forms of muscular dystrophy and hemophilia, as well as red-green color blindness, are a few X-linked human phenotypes. (See Figure 10.26.)
- *Contrary to popular opinion, male pattern baldness is not X-linked. This is a sex-influenced trait that is probably subject to hormonal influence. Males require only one gene for baldness to appear; female baldness requires the presence of two genes.*

Cytoplasmic Inheritance

- Mendelian genetics is the genetics of the nucleus, yet other cytoplasmic organelles carry genetic material.
- Mitochondria, chloroplasts, and other plastids possess a small amount of DNA.
- Humans have about 600,000 genes in the nucleus and 37 genes in mitochondria.
- Plastid genomes are five times larger than those of mitochondria.
- The genome is the total configuration of genetic material. Mitochondria and plastids are passed on by the mother only, as the egg contains abundant cytoplasm and organelles. The mitochondria in sperm do not take part in gamete union.
- Some chloroplasts are white, not green, because of a mutation in their DNA.
- Mitochondrial mutations may also be linked to human genetic diseases.

The Instructor's Resource CD-ROM includes photographs showing the ABO blood reactions, a banded polytene chromosome from *Drosophila*, and the Mendelian ratios of the bay scallop.