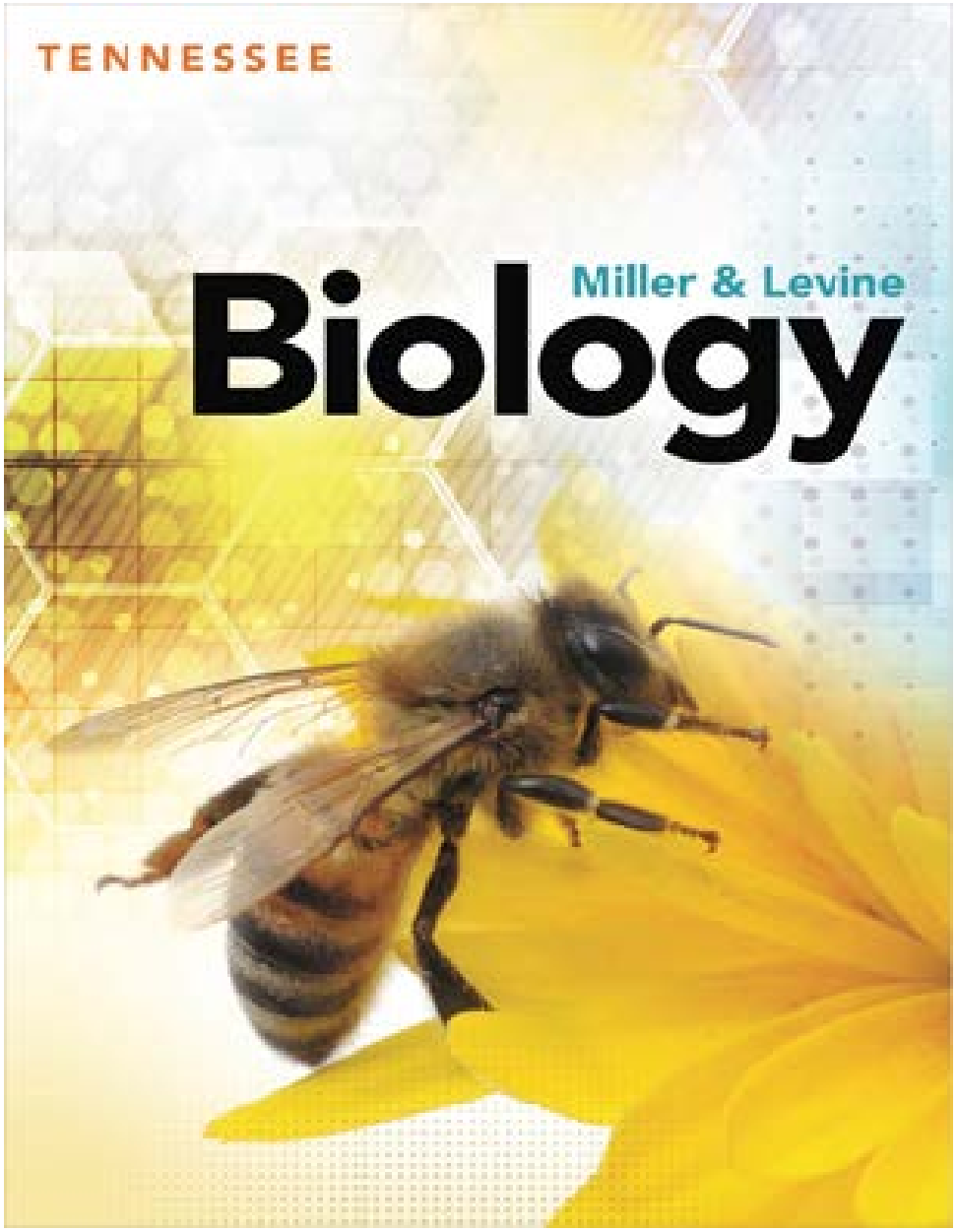


Lesson Overview

15.1 Human Chromosomes

THINK ABOUT IT

If you had to pick an ideal organism for the study of genetics, would you choose one that produced lots of offspring, was easy to grow in the lab, and had a short life span that allowed you to do several crosses per month?

THINK ABOUT IT

You certainly would not choose an organism that produced very few offspring, had a long life span, and could not be grown in a lab. Yet, when we study human genetics, this is exactly the sort of organism we deal with.

Given all of these difficulties, it may seem a wonder that we know as much about human genetics as we do.

Karyotypes

- What is a karyotype?

Karyotypes

- What is a karyotype?

- A karyotype shows the complete diploid set of chromosomes grouped together in pairs, arranged in order of decreasing size.

Karyotypes

To find what makes us uniquely human, we have to explore the human genome.

A **genome** is the full set of genetic information that an organism carries in its DNA.

A study of any genome starts with chromosomes, the bundles of DNA and protein found in the nuclei of eukaryotic cells.

Karyotypes

To see human chromosomes clearly, cell biologists photograph cells in mitosis, when the chromosomes are fully condensed and easy to view.

Karyotypes

Scientists then cut out the chromosomes from the photographs and arrange them in a picture known as a **karyotype**. It shows the complete diploid set of chromosomes grouped together in pairs, arranged in order of decreasing size.

A karyotype from a typical human cell, which contains 46 chromosomes, is arranged in 23 pairs.

Sex Chromosomes

Two of the 46 chromosomes in the human genome are known as **sex chromosomes, because they determine an individual's sex.**

Females have two copies of the X chromosome.

Males have one X chromosome and one Y chromosome.

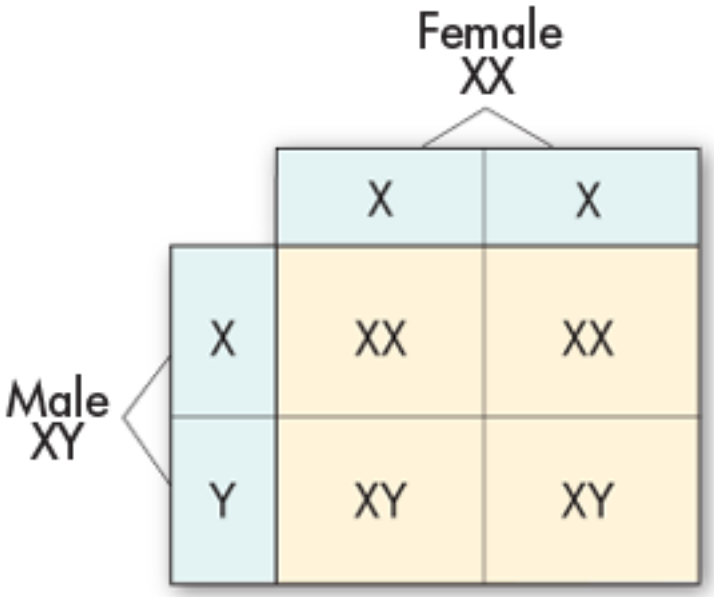
Sex Chromosomes

This Punnett square illustrates why males and females are born in a roughly 50 : 50 ratio.

All human egg cells carry a single X chromosome (23,X).

However, half of all sperm cells carry an X chromosome (23,X) and half carry a Y chromosome (23,Y).

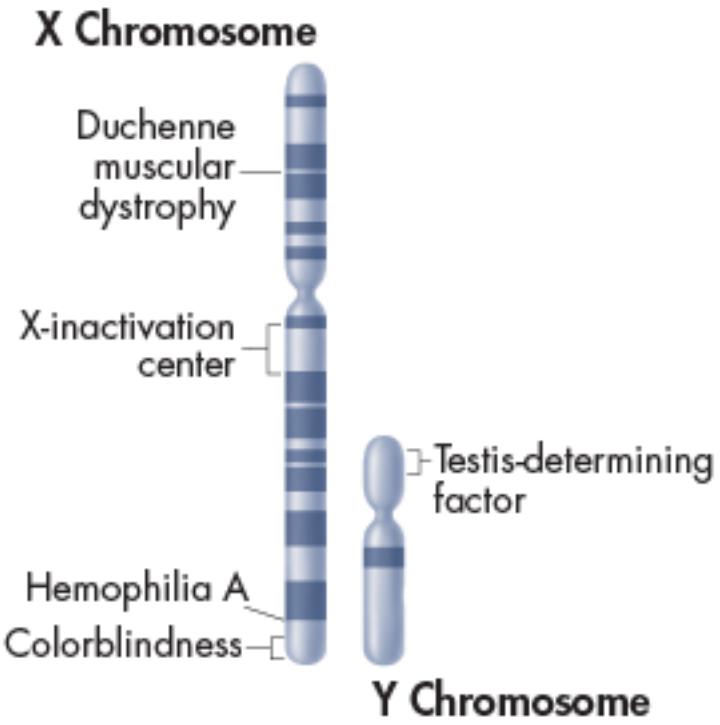
This ensures that just about half the zygotes will be males and half will be females.



Sex Chromosomes

More than 1200 genes are found on the X chromosome, some of which are shown.

The human Y chromosome is much smaller than the X chromosome and contains only about 140 genes, most of which are associated with male sex determination and sperm development.



Autosomal Chromosomes

The remaining 44 human chromosomes are known as autosomal chromosomes, or **autosomes.**

The complete human genome consists of 46 chromosomes, including 44 autosomes and 2 sex chromosomes.

To quickly summarize the total number of chromosomes present in a human cell, biologists write 46,XX for females and 46,XY for males.

Transmission of Human Traits

- What patterns of inheritance do human traits follow?

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- Many human traits follow a pattern of simple dominance.

Transmission of Human Traits

- What patterns of inheritance do human traits follow?
- The alleles for many human genes display codominant inheritance.

Transmission of Human Traits

- What patterns of inheritance do human traits follow?

- Because the X and Y chromosomes determine sex, the genes located on them show a pattern of inheritance called sex-linked.

Dominant and Recessive Alleles

Many human traits follow a pattern of simple dominance.

For example, a gene known as *MC1R* helps determine skin and hair color.

Some of *MC1R's* recessive alleles produce red hair. An individual with red hair usually has two sets of these recessive alleles, inheriting a copy from each parent.

Dominant alleles for the *MC1R* gene help produce darker hair colors.

Dominant and Recessive Alleles

Another trait that displays simple dominance is the Rhesus, or Rh blood group.

The allele for Rh factor comes in two forms: Rh^+ and Rh^-.

Rh^+ is dominant, so an individual with both alleles (Rh^+/Rh^-) is said to have Rh positive blood.

Rh negative blood is found in individuals with two recessive alleles (Rh^+/Rh^-).

Codominant and Multiple Alleles

The alleles for many human genes display codominant inheritance.

One example is the ABO blood group, determined by a gene with three alleles: I^A, I^B, *and i.*

Codominant and Multiple Alleles

This table shows the relationship between genotype and phenotype for the ABO blood group.

It also shows which blood types can safely be transfused into people with other blood types.

If a patient has AB-negative blood, it means the individual has I^A and I^B alleles from the ABO gene and two Rh^- alleles from the Rh gene.

Blood Groups

Phenotype (Blood Type)	Genotype	Antigen on Red Blood Cell	Safe Transfusions	
			To	From
A	I^AI^A or I^Ai	A	A, AB	A, O
B	I^BI^B or I^Bi	B	B, AB	B, O
AB	I^AI^B	A and B	AB	A, B, AB,
O	ii	None	A, B, AB,	O

Codominant and Multiple Alleles

If a patient has AB-negative blood, it means the individual has I^A and I^B alleles from the ABO gene and two Rh^- alleles from the Rh gene.

Blood Groups

Phenotype (Blood Type)	Genotype	Antigen on Red Blood Cell	Safe Transfusions	
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A	I^AI^A or I^Ai	A	A, AB	A, O
B	I^BI^B or I^Bi	B	B, AB	B, O
AB	I^AI^B	A and B	AB	A, B, AB,
O	ii	None	A, B, AB,	O

Codominant and Multiple Alleles

Alleles I^A and I^B are codominant. They produce molecules known as antigens on the surface of red blood cells.

Individuals with alleles I^A and I^B produce both A and B antigens, making them blood type AB.

Blood Groups

Phenotype (Blood Type)	Genotype	Antigen on Red Blood Cell	Safe Transfusions	
			To	From
A	I^AI^A or I^Ai	A	A, AB	A, O
B	I^BI^B or I^Bi	B	B, AB	B, O
AB	I^AI^B	A and B	AB	A, B, AB,
O	ii	None	A, B, AB,	O

Codominant and Multiple Alleles

The *i* allele is recessive.

Individuals with alleles I^AI^A or I^Ai produce only the A antigen, making them blood type A.

Those with I^BI^B or I^Bi alleles are type B.

Those homozygous for the *i* allele (ii) produce no antigen and are said to have blood type O.

Sex-Linked Inheritance

The genes located on the X and Y chromosomes show a pattern of inheritance called sex-linked.

A **sex-linked gene** is a gene located on a sex chromosome.

Genes on the Y chromosome are found only in males and are passed directly from father to son.

Genes located on the X chromosome are found in both sexes, but the fact that men have just one X chromosome leads to some interesting consequences.

Sex-Linked Inheritance

For example, humans have three genes responsible for color vision, all located on the X chromosome.

In males, a defective allele for any of these genes results in colorblindness, an inability to distinguish certain colors. The most common form, red-green colorblindness, occurs in about 1 in 12 males.

Among females, however, colorblindness affects only about 1 in 200. In order for a recessive allele, like colorblindness, to be expressed in females, it must be present in two copies—one on each of the X chromosomes.

The recessive phenotype of a sex-linked genetic disorder tends to be much more common among males than among females.

X-Chromosome Inactivation

If just one X chromosome is enough for cells in males, how does the cell "adjust" to the extra X chromosome in female cells?

In female cells, most of the genes in one of the X chromosomes are randomly switched off, forming a dense region in the nucleus known as a Barr body.

Barr bodies are generally not found in males because their single X chromosome is still active.

X-Chromosome Inactivation

X-chromosome inactivation also happens in other mammals.

In cats, a gene that controls the color of coat spots is located on the X chromosome.

X-Chromosome Inactivation

One X chromosome may have an allele for orange spots and the other X chromosome may have an allele for black spots.

In cells in some parts of the body, one X chromosome is switched off. In other parts of the body, the other X chromosome is switched off. As a result, the cat's fur has a mixture of orange and black spots.

X-Chromosome Inactivation

Male cats, which have just one X chromosome, can have spots of only one color.

If a cat's fur has three colors—white with orange and black spots, for example—you can almost be certain that the cat is female.

Human Pedigrees

- How can pedigrees be used to analyze human inheritance?

Human Pedigrees

- How can pedigrees be used to analyze human inheritance?

- The information gained from pedigree analysis makes it possible to determine the nature of genes and alleles associated with inherited human traits.

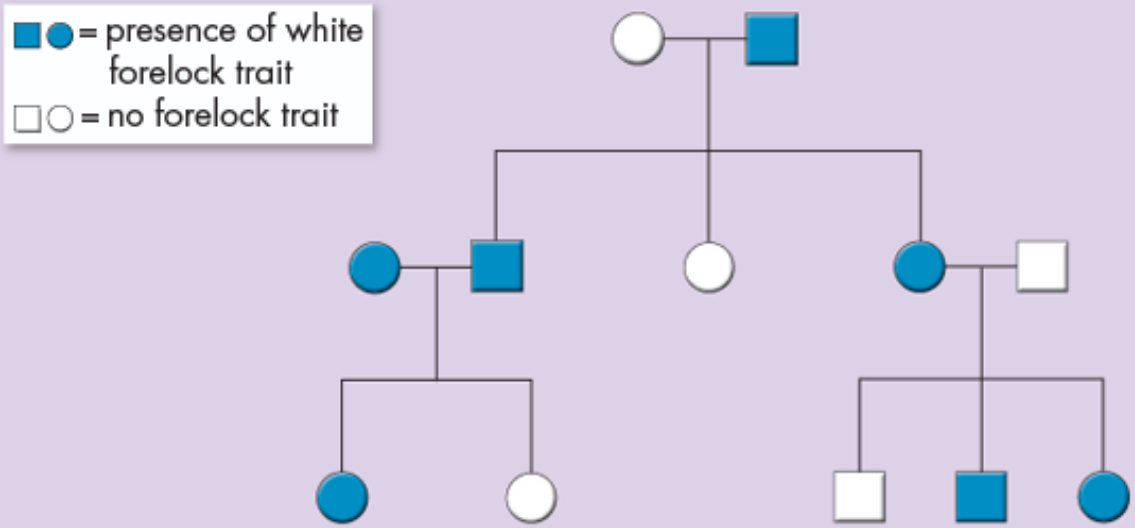
Human Pedigrees

To analyze the pattern of inheritance followed by a particular trait, you can use a chart, called a **pedigree**, which shows the relationships within a family.

A pedigree shows the presence or absence of a trait according to the relationships between parents, siblings, and offspring.

Human Pedigrees

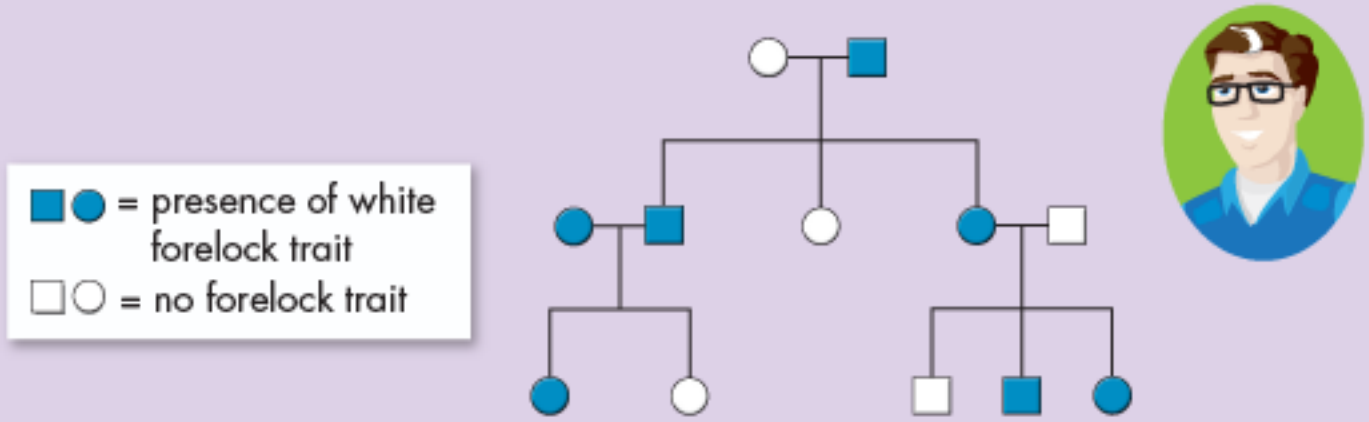
This diagram shows what the symbols in a pedigree represent.



Human Pedigrees

This pedigree shows how one human trait—a white lock of hair just above the forehead—passes through three generations of a family.

The allele for the white forelock trait is dominant.

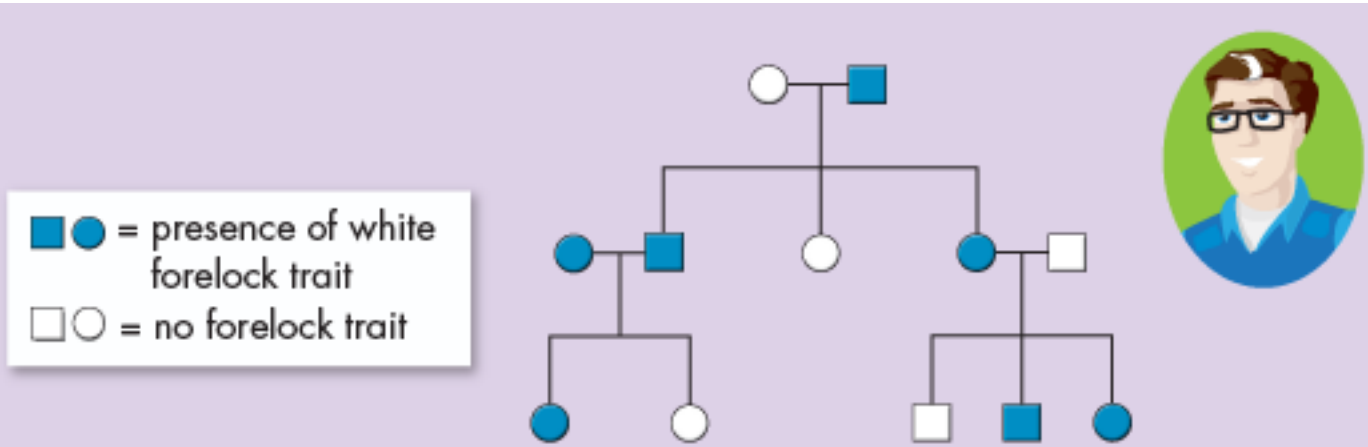


Human Pedigrees

At the top of the chart is a grandfather who had the white forelock trait.

Two of his three children inherited the trait.

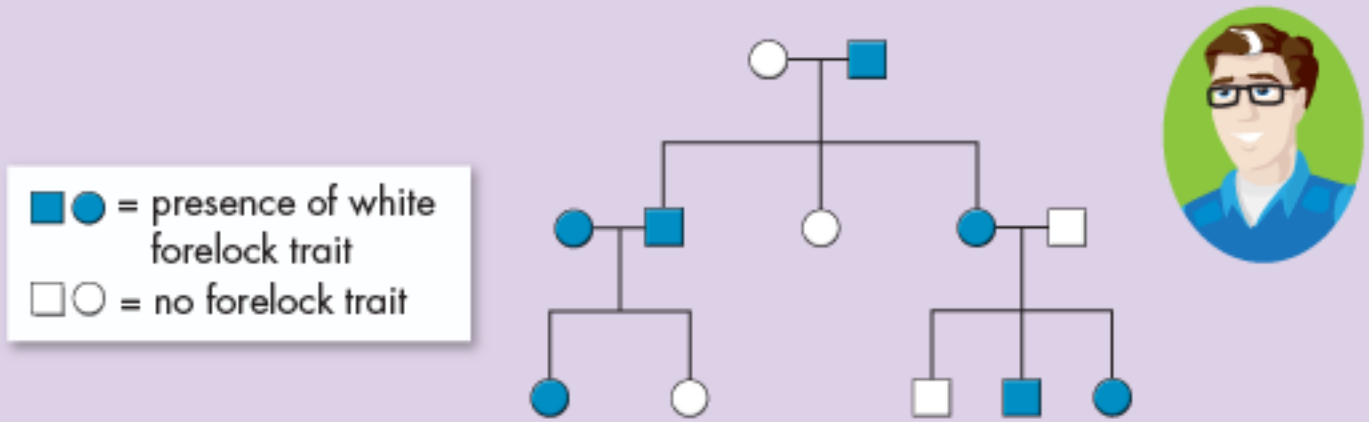
Three grandchildren have the trait, but two do not.



Human Pedigrees

Because the white forelock trait is dominant, all the family members in the pedigree lacking this trait must have homozygous recessive alleles.

One of the grandfather's children lacks the white forelock trait, so the grandfather must be heterozygous for this trait.



Human Pedigrees

The information gained from pedigree analysis makes it possible to determine the nature of genes and alleles associated with inherited human traits.

Based on a pedigree, you can often determine if an allele for a trait is dominant or recessive, autosomal or sex-linked.