SECTION

7.4 Human Genetics and Pedigrees

KEY CONCEPT A combination of methods is used to study human genetics.

Human genetics follows the patterns seen in other organisms.

Fruit flies and pea plants may seem simple. And they are certainly different than humans. But human genetics follows the same patterns of heredity. The process of meiosis happens the same way in humans as in fruit flies. Humans, like other organisms that reproduce sexually, have the same relationships between alleles: dominant-recessive interactions, polygenic traits, and sex-linked genes, among others.

Although many traits are complex, single-gene traits are helpful in understanding human genetics. A downward-pointed hairline—called a widow's peak—is a single-gene trait with a dominant-recessive inheritance pattern. Many human genetic disorders are, too. Much of what is known about human genetics comes from studying genetic disorders.

In what ways are human genetics similar to fruit fly or pea plant genetics? _____

_____

_____

Females can carry sex-linked disorders.

Recall from Section 7.1 that some genetic disorders are caused by genes on autosomes. Both males and females can be carriers of a recessive autosomal disorder. That is, they can have one recessive allele but have no symptoms of the disorder.

In contrast, only females can be carriers of a sex-linked disorder. Recall that the X chromosome has far more genes than the Y chromosome, including some that cause genetic disorders. A sex-linked disorder usually refers to a gene on the X chromosome.

Females have an XX genotype. Because they have two X chromosomes, they can be heterozygous and have one recessive allele, but not have symptoms of the disorder. A male has an XY genotype. A male who has an allele for a disorder located on the X chromosome will not have a second, normal allele to mask it.

Why can't males be carriers of sex-linked disorders? _____

_____

VOCABULARY

The term *carrier* means "a person who transports something." In genetics, a carrier is a person who "transports" a recessive allele but does not express the recessive phenotype.

A pedigree is a chart for tracing genes in a family.

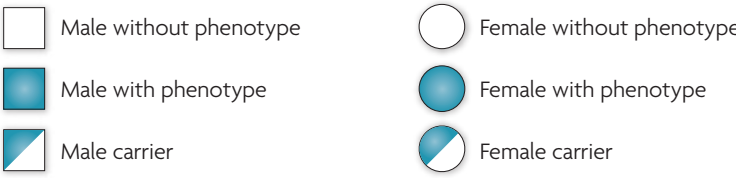
A **pedigree** chart can help trace the phenotypes and genotypes in a family to determine the chance that a child might have a certain genetic disorder. The genotypes can often be figured out using enough family phenotypes. Using phenotypes to figure out the possible genotypes in a family is like solving a puzzle. You have to use logic* and clues to narrow the possibilities for each person's genotype.

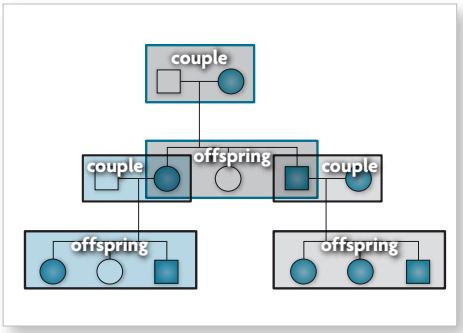
Tracing Autosomal Genes

Consider the widow's peak, the pointed shape of a person's hairline, mentioned earlier in this section. The widow's peak is an autosomal trait. A pointed hairline is dominant to a straight hairline.

- People with a widow's peak have either homozygous dominant (*WW*) or heterozygous (*Ww*) genotypes.
- Two parents without a widow's peak are both homozygous recessive (*ww*), and cannot have children who have a widow's peak.
- Two parents who both have a widow's peak can have a child who does not (*ww*) if both parents are heterozygous (*Ww*).

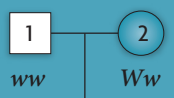
The inheritance of this trait is shown in the figure below.





TRACING AUTOSOMAL GENES: WIDOW'S PEAK

Parental generation



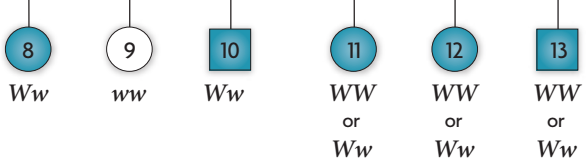
W = Dominant
w = Recessive

- Male 1 must be *ww* and female 2 must be heterozygous (*Ww*), because they have a daughter (5) with the recessive trait.

F_1 generation

- Children 4 and 6 have the widow's peak trait. They must be heterozygous, because they can inherit only one dominant allele.

F_2 generation



- Children 8 and 10 have the widow's peak trait. They must be heterozygous, because they can inherit only one dominant allele.

* ACADEMIC VOCABULARY

logic reason, critical thinking

Tracing Sex-Linked Genes

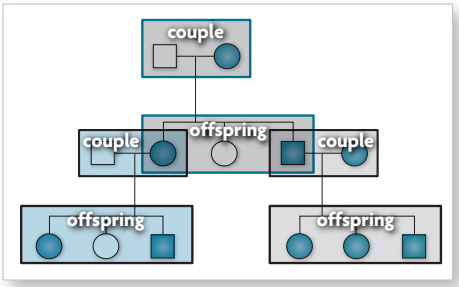
PHENOTYPE	GENOTYPES	
	MALE	FEMALE
Red-green colorblind	X^mY	X^mX^m
Normal vision	X^MY	X^MX^m or X^MX^M

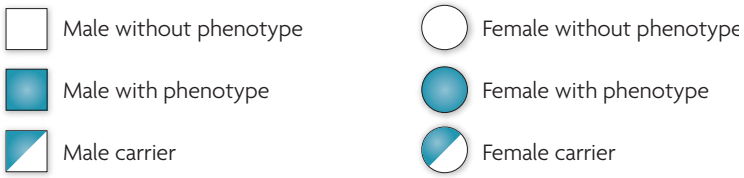
For sex-linked genes, you have to think about dominant and recessive alleles, but you also have to think about inheritance of the sex chromosomes. One example of a sex-linked trait is red-green colorblindness, a condition that causes a person to not be able to see the difference between some colors.

By using a process of elimination*, you can often figure out the possible genotypes for a given phenotype.

- Colorblind females must be homozygous recessive (X^mX^m).
- Males who are colorblind must have the recessive allele (X^mY).
- Females who are heterozygous for the alleles (X^MX^m) are not colorblind, but are carriers of the trait.

The inheritance of colorblindness is shown in the figure below.





TRACING SEX-LINKED GENES: COLORBLINDNESS

Parental generation

1 X^MY — 2 X^MX^m

X^M = Dominant
X^m = Recessive

- Male 1 must be X^MY and female 2 must be a carrier (X^MX^m) because they have two colorblind sons.

F_1 generation

3 X^MY — 4 X^MX^m — 5 ? X^MX^M or X^MX^m — 6 X^mY — 7 X^mY — 8 ? X^MX^M or X^MX^m

- Female 4 must be a carrier (X^MX^m) because she has a colorblind son. Males 6 and 7 must be X^mY. Females 5 and 8 are not colorblind, but it is not possible to determine whether they are carriers.

F_2 generation

9 ? X^MX^M or X^MX^m — 10 X^MY — 11 ? X^MX^M or X^MX^m — 12 X^mY — 13 X^MX^m — 14 X^MX^m — 15 X^MY

- Children 13 and 14 must be carriers because their father is colorblind. Females 9 and 11 are not colorblind, but it is not possible to determine whether they are carriers.

INSTANT REPLAY

What is one difference between tracing the inheritance of autosomal traits and tracing the inheritance of sex-linked traits?

* ACADEMIC VOCABULARY

process of elimination getting to the best answer by ruling out possible answers that are not correct

Several methods help map human chromosomes.

The human genome, or all of the DNA in a human cell, is so large that it is difficult to map human genes. In addition to pedigrees, other methods more directly study human chromosomes. A **karyotype** (KAR-ee-uh-TYP), for example, is a picture of all of the chromosomes in a cell. Chemicals are used to stain the chromosomes. The stains produce a pattern of bands on the chromosomes. These patterns can be used to tell different chromosomes apart. Karyotypes can also show if there are extra chromosomes or missing parts of chromosomes. Down syndrome, for example, results from an extra copy of at least part of chromosome 21, and can be identified on a karyotype. The large-scale mapping of all human genes truly began with the Human Genome Project, which you will read more about in Chapter 9.

INSTANT REPLAY

What is one example of a genetic disorder that can be seen on a karyotype? _____

7.4 Vocabulary Check

pedigree karyotype

Mark It Up

Go back and highlight each sentence that has a vocabulary word in **bold.**

Label the drawings below with the proper term.

1. 

2. 

_____ _____

7.4 The Big Picture

3. Why can the genetics of fruit flies be applied to humans?

_____

4. What is the genotype of a female carrier of a sex-linked genetic disorder? _____

5. What are pedigree charts used for? _____

_____

6. What types of information can a karyotype provide?

_____