

# 14-1 Human Heredity

## Human Chromosomes

Cell biologists analyze chromosomes by looking at karyotypes.

A karyotype is a photograph of all paired chromosomes.

Human Karyotype



## Human Chromosomes

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

- Females have two copies of an X chromosome.
- Males have one X chromosome and one Y chromosome.

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

### How is sex determined?



**All human egg cells carry a single X chromosome.**

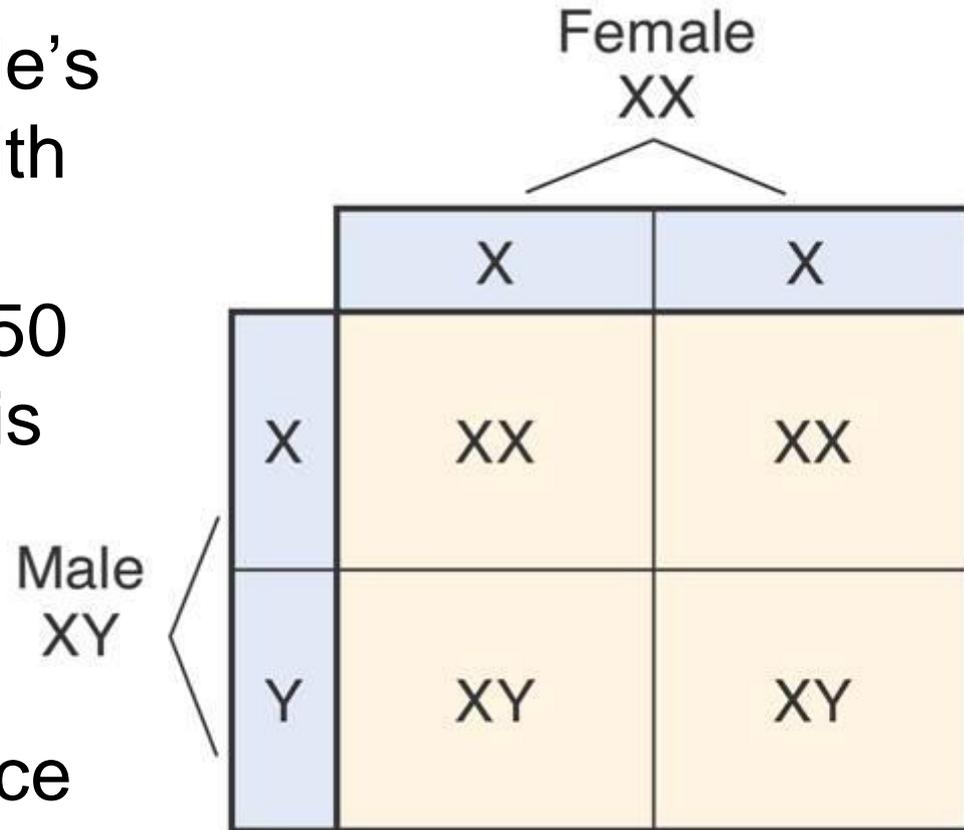
**Half of all sperm cells carry an X chromosome and half carry a Y chromosome.**

**Sex is determined by which sperm, carrying the X or Y chromosome, is fertilizing the egg.**

## Human Chromosomes

When crossing a male's sex chromosomes with a female's sex chromosomes, a 50:50 ratio of the offspring is produced.

There is a 50% chance a female will be born and a 50% chance a male will be born.



# Human Genes

The human genome includes tens of thousands of genes.

In 2003, the DNA sequence of the human genome was published.

In a few cases, biologists were able to identify genes that directly control a single human trait such as blood type.

### Blood Group Genes

Human blood comes in a variety of genetically determined blood groups.

A number of genes are responsible for human blood groups.

The best known are the ABO blood groups and the Rh blood groups.

## Human Genes

The Rh blood group is determined by a single gene with two alleles—positive and negative.

The positive ( $Rh^+$ ) allele is dominant, so individuals who are  $Rh^+/Rh^+$  or  $Rh^+/Rh$  are said to be Rh-positive.

Individuals with two  $Rh^-$  alleles are said to be Rh-negative.

### ABO blood group

- There are three alleles for this gene,  $I^A$ ,  $I^B$ , and  $i$ .
- Alleles  $I^A$  and  $I^B$  are codominant.

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
AB	$I^A I^B$	A and B	AB	A, B, AB, O

The *i* allele is recessive.

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

Blood Groups				
Phenotype (Blood Type)	Genotype	Antigen on Red Blood Cell	Safe Transfusions	
			To	From
A	$I^A I^A$ or $I^A i$	A	A, AB	A, O

Individuals with  $I^B I^B$  or  $I^B i$  alleles are type B.

Blood Groups				
Phenotype (Blood Type)	Genotype	Antigen on Red Blood Cell	Safe Transfusions	
			To	From
B	$I^B I^B$ or $I^B i$	B	B, AB	B, O

Individuals who are homozygous for the *i* allele (*ii*) produce no antigen and have blood type O.

Blood Groups				
Phenotype (Blood Type)	Genotype	Antigen on Red Blood Cell	Safe Transfusions	
			To	From
O	<i>ii</i>	none	A, B, AB, O	O

# 14–2 Human Chromosomes

### Sex-Linked Genes

- The X chromosome and the Y chromosomes determine sex.
- Genes located on these chromosomes are called **sex-linked genes**.
- More than 100 sex-linked genetic disorders have now been mapped to the X chromosome.

## Sex-Linked Genes

The Y chromosome is much smaller than the X chromosome and appears to contain only a few genes.

### X Chromosome



Duchenne muscular dystrophy

Melanoma

X-inactivation center

X-linked severe combined immunodeficiency (SCID)

Colorblindness

Hemophilia

### Y Chromosome



Testis-determining factor

## Sex-Linked Genes

Why are sex-linked disorders more common in males than in females?

## Sex-Linked Genes

For a recessive allele to be expressed in females, there must be two copies of the allele, one on each of the two X chromosomes.



**Males have just one X chromosome. Thus, all X-linked alleles are expressed in males, even if they are recessive.**

### Colorblindness

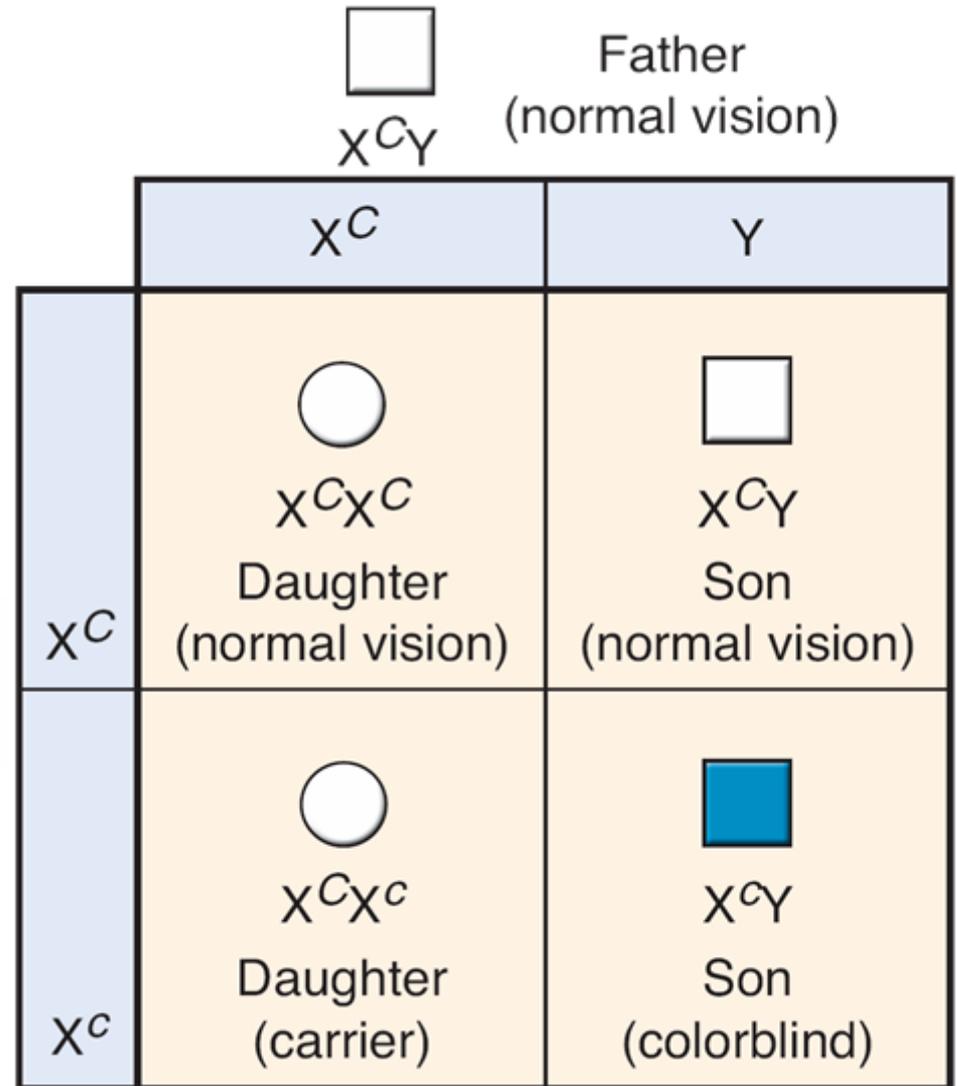
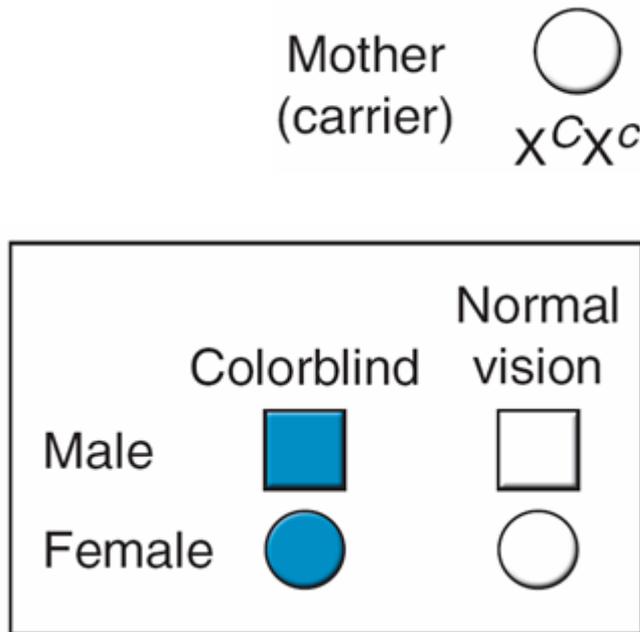
Three human genes associated with color vision are located on the X chromosome.

In males, a defective version of any one of these genes produces colorblindness.

Color blindness is the inability to perceive certain colors.

## Sex-Linked Genes

### Example of Inheritance for Colorblindness Allele



### Hemophilia

- The X chromosome also carries genes that help control blood clotting. A recessive allele in either of these two genes may produce hemophilia.
- Hemophilia is the inability for blood to clot.
- A hemophiliac is a person who has hemophilia.