SECOND EDITION Colleen Belk • Virginia Borden

Chapter 7

DNA Detective: Extensions of Mendelism, Sex Linkage, Pedigree Analysis, and DNA Fingerprinting

7.1 Extension of Mendelian Genetics

- Patterns of inheritance that are not straightforward are called extensions of Mendelian genetics

- When the offspring of two different parents has a phenotype that is unlike either parent
  - the trait is said to display **incomplete dominance**

Extension	Example
Incomplete dominance Heterozygote is intermediate to either homozygote.	Flower color in snapdragons  Red = *RR* ×  White = *rr* =  Pink = *Rr*

Table 7-1 part 1 Biology: Science for Life, 2/e

Extension of Mendelian Genetics

- **Polygenic**
  - traits that are controlled by many genes
  - Most human traits
- Predicting inheritance is more difficult for **polygenic** traits
  - especially when the environmental also influences

Extension	Example
Polygenic inheritance More than one gene determines the phenotype.	Skin color, eye color, height, IQ

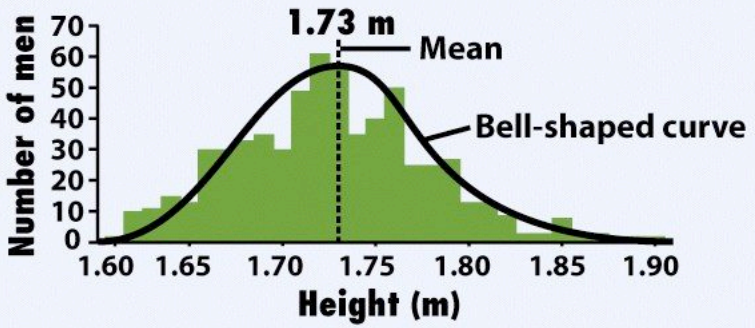


Table 7-1 part 4 Biology: Science for Life, 2/e

Extension of Mendelian Genetics

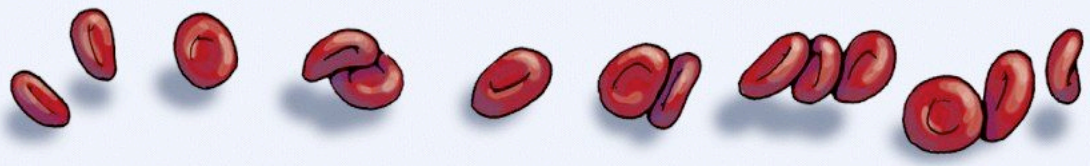
Two other extensions are:

- **Codominance**
  - both alleles are expressed
- **Multiple allelism**
  - there are more than two allele options for the gene

- These extensions are used in the **ABO blood system** of blood typing

Extension of Mendelian Genetics
Essay 7.1

- Codominance:
  - If you are blood type AB, you have one allele for A and one allele for B
  - Both are seen in the codominant phenotype
    - A does not mask B and B does not mask A

Extension	Example
Codominance Both alleles are expressed in a heterozygote.	ABO blood system 

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Extension of Mendelian Genetics/Essay 7.1

- **Multiple allelism:**
  - There are 3 options for alleles in ABO blood types
    - I^A, I^B, and i
  - There are 3 options but each person only has 2 alleles
    - Each person only has a pair or 2 chromosomes that carry the gene for blood type

- I^AI^A - blood type A
- I^Ai - blood type A
- I^BI^B - blood type B
- I^Bi - blood type B
- I^AI^B - blood type AB
- ii - blood type O

Extension of Mendelian Genetics/Essay 7.1

- Inheritance of the **Rh factor** of the red blood cells is determined by two alleles that show complete dominance
- If your blood type is A negative
  - You can be $I^A I^A$ - - or $I^A i$ - -
- If your blood type is A positive
  - You can be $I^A I^A$ + - or $I^A I^A$ + + or $I^A i$ + - or $I^A i$ + +
- Blood typing can only show that two people may be related or that they are not related
  - Can sometimes be used to exclude men in a paternity suit

Extension of Mendelian Genetics another exampole of multiple allelism

Extension	Example
Multiple allelism One gene has many alleles.	Coat color in rabbits is controlled by four different alleles. **Genotype** — **Phenotype** $c^{}c^{}$ — Albino  $C^{h}C^{h}$ — Himalayan  $C^{ch}C^{ch}$ — Chinchilla  $C^{+}C^{+}$ — Darkly pigmented rabbit 

Table 7-1 part 3 Biology: Science for Life, 2/e

Extension of Mendelian Genetics

- The Singed gene in drosophila an example of **pleiotropy**
  - the ability of one gene to affect at least 2 different functions

Extension	Example
Pleiotropy One gene has many effects.	The singed gene in fruit flies controls both bristle formation and fertility. **Genotype** *BB* and *Bb* — **Phenotype** Normal bristles and fertile  *bb* — Singed bristles, infertile 

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7.2 Sex Determination and Sex Linkage

- Some genes are on the X chromosome and are inherited in a specific manner
- In humans, **sex determination** involves the X and Y chromosomes
- There are two types of sex chromosomes in humans: X and Y
  - If you have XX you are female
  - If you have XY you are male

Sex Determination

- All egg cells contain
  - an X chromosome
- Sperm cells can either contain
  - an X or a Y chromosome
- Fathers determine the sex of the offspring

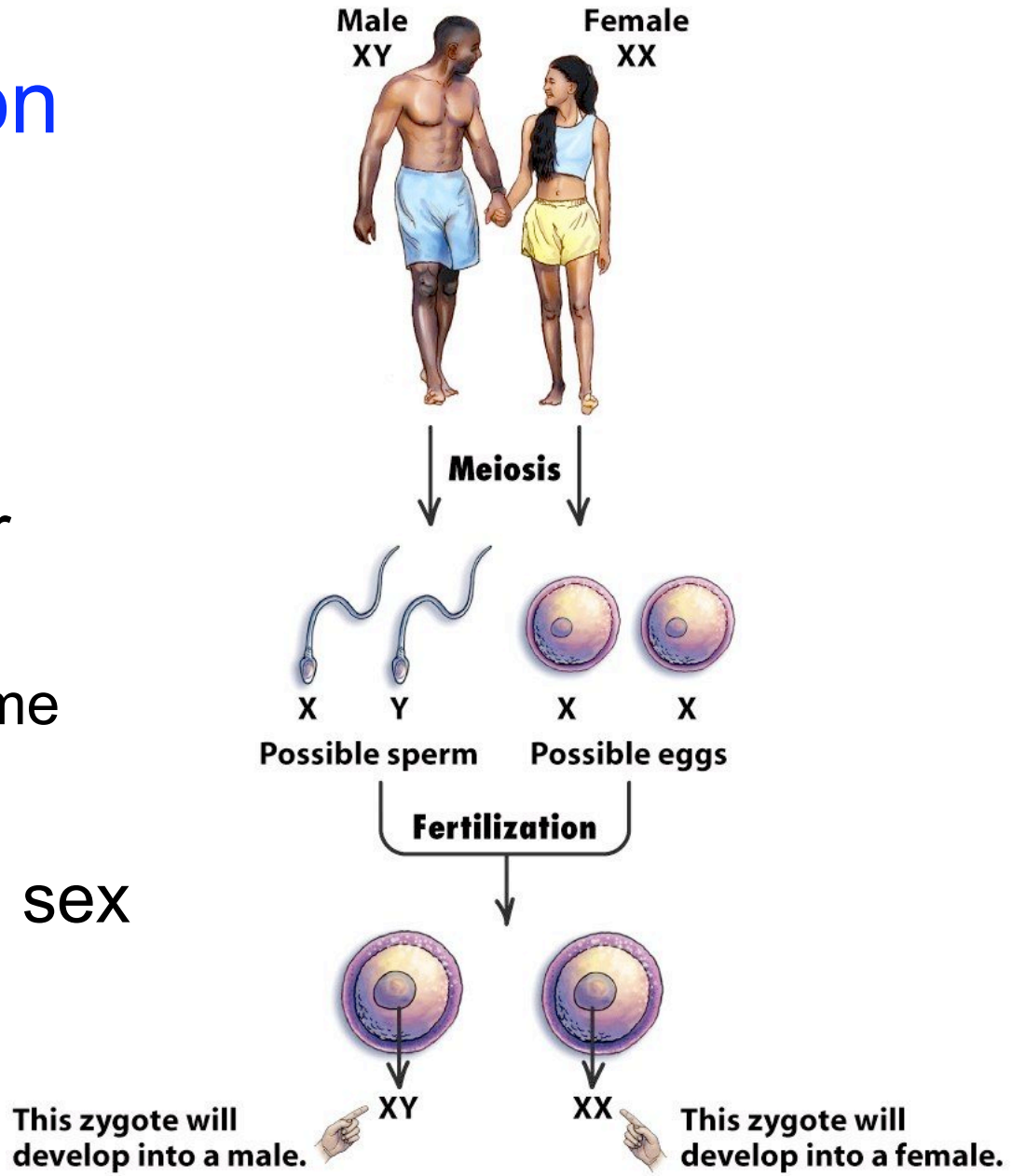

Figure 7-1 Biology: Science for Life, 2/e
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Meiosis and Sex Chromosomes

- Sometimes the sex chromosomes do not separate during meiosis

- This is called **nondisjunction** and can occur in either meiosis I or meiosis II
  - Just like it can happen for autosomes
  - In females, both X chromosomes go to one cell
  - In males, X and Y chromosomes go to one cell

Sex Chromosomes and Nondisjunction

Conditions Caused by Nondisjunction of Sex Chromosomes	Approximate Frequency Among Live Births	Comments
XO— Turner syndrome 	1 in 5000 females	People with only one X chromosome are females with retarded sexual development. They are usually sterile since their ovaries often fail to develop. They can have webbing of the neck, shorter stature, and some hearing impairment. Since they are missing an X chromosome, affected females have 45 chromosomes.

Table E7-3 part 4 Biology: Science for Life, 2/e

Conditions Caused by Nondisjunction of Sex Chromosomes	Approximate Frequency Among Live Births	Comments
Trisomy X— Meta Female 	1 in 1000 females	Meta females have three X chromosomes. Two of the X chromosomes are condensed to Barr bodies and most XXX females develop normally. Since these women have an extra X chromosome, the cells of their bodies have 47 chromosomes.

Table E7-3 part 5 Biology: Science for Life, 2/e

Sex Chromosomes and Nondisjunction

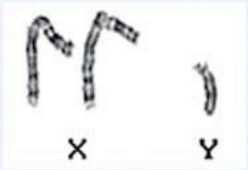
Conditions Caused by Nondisjunction of Sex Chromosomes	Approximate Frequency Among Live Births	Comments
XXY— Kleinfelter syndrome 	1 in 1000 males	Males with the XXY genotype are less fertile than XY males, have small testes, sparse body hair, some breast enlargement, and may have mental retardation. Testosterone injections can reverse some of the anatomical abnormalities but not the mental retardation and lowered fertility.

Table E7-3 part 6 Biology: Science for Life, 2/e

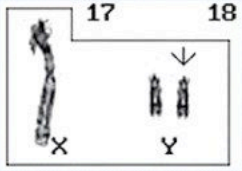
Conditions Caused by Nondisjunction of Sex Chromosomes	Approximate Frequency Among Live Births	Comments
XYY syndrome 	1 in 1000 males	Males with two Y chromosomes tend to be taller than average but have a normal male phenotype.

Table E7-3 part 7 Biology: Science for Life, 2/e

Sex Linkage

- The genes on the X or Y chromosomes are called **sex-linked genes**
- Genes on X are called “X-linked,” while those on Y are called “Y-linked”
- The X chromosome is much larger and carries far more genetic information

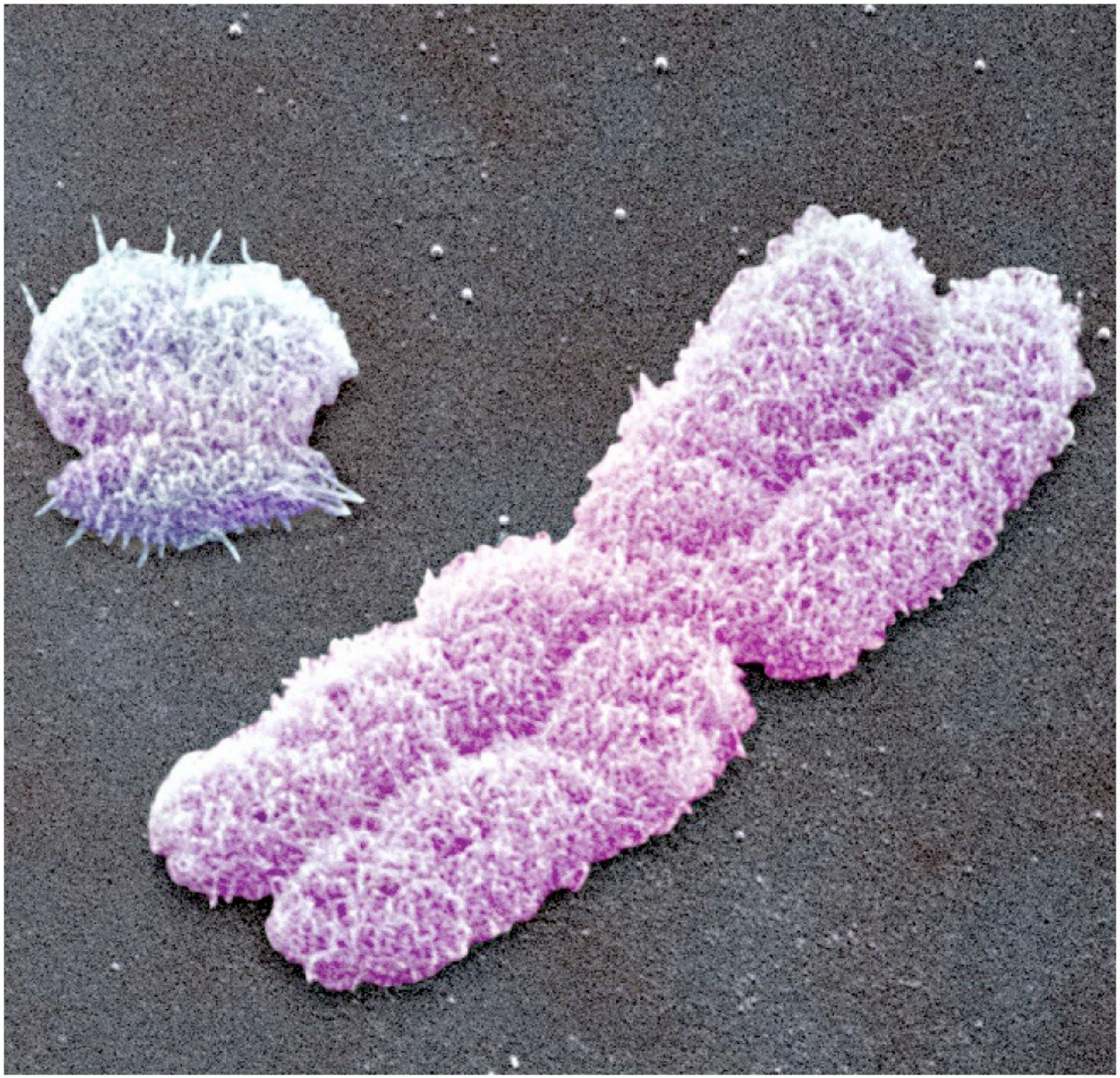


Figure 7-2 Biology: Science for Life, 2/e

X-Linked Genes

- Males inherit their X chromosomes from their mothers
- Males get the Y chromosome from their fathers
- Males only have one X chromosome
  - they are more likely to suffer from recessive X-linked diseases or traits
  - Only have one x; if it has the disease allele, he has the disease
    - Like hemophilia
    - Like pattern baldness

X-Linked Genes

- Females get one X chromosome from each parent
  - have two copies
  - are less likely to suffer from X-linked recessive diseases
    - Both X's have to have disease allele for female to have disease
- Females can be **carriers** of disease
  - They are not affected because they have one allele of a recessive trait
  - Need two alleles of a recessive trait to have disease
- and pass the disease on to offspring, especially sons
  - Because they only have one X chromosome

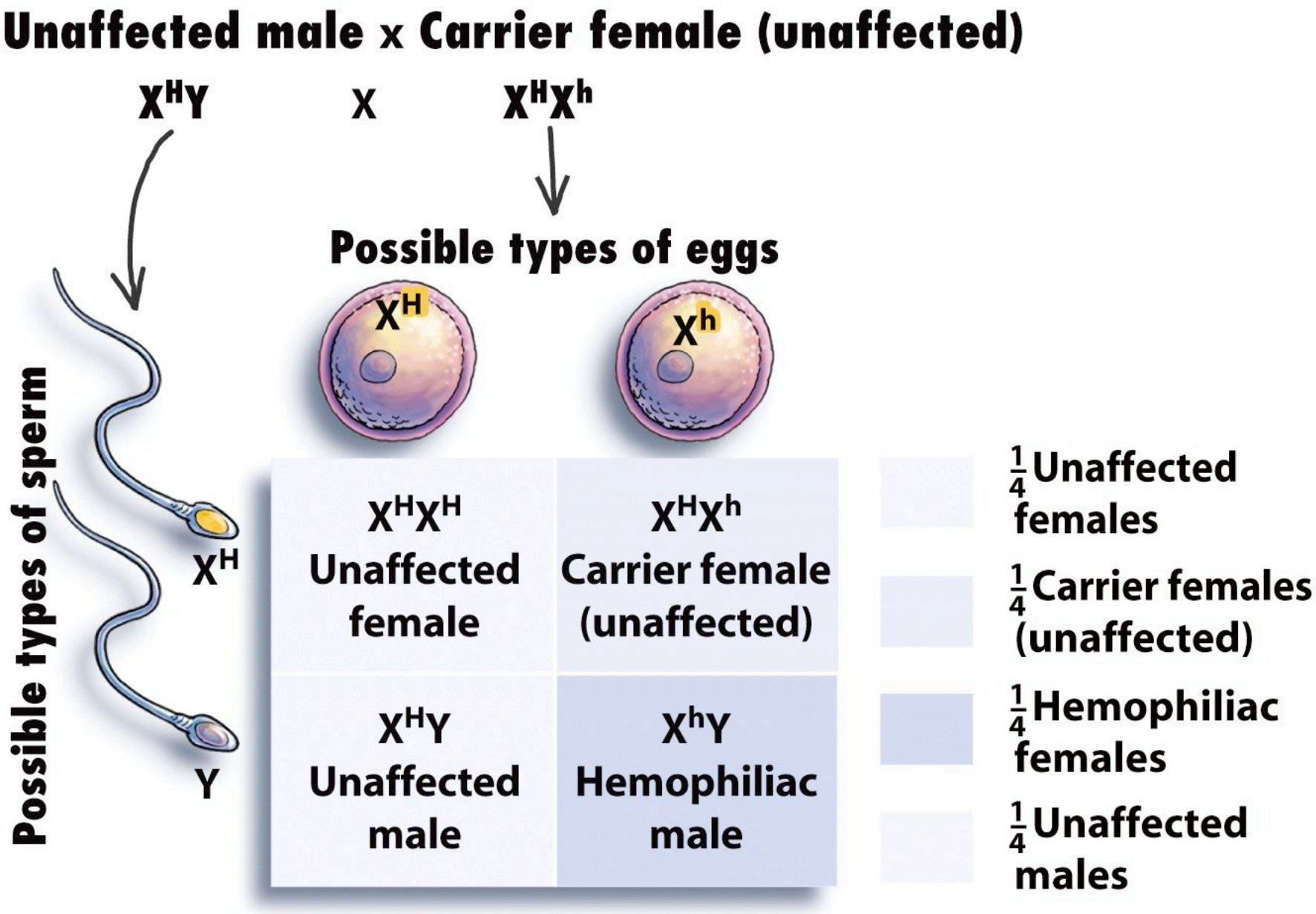


Figure 7-3a Biology: Science for Life, 2/e
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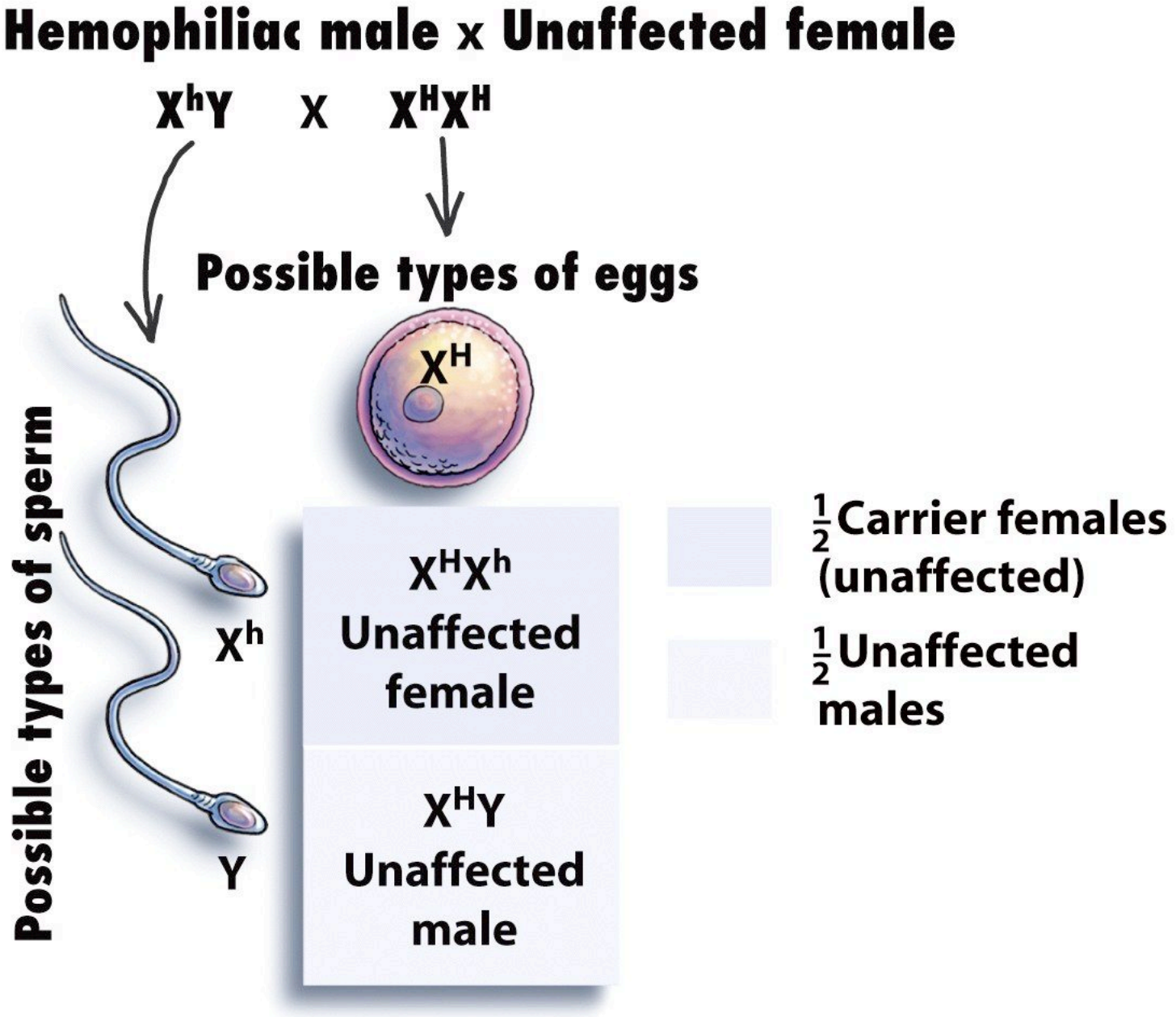


Figure 7-3b Biology: Science for Life, 2/e

X-Linked Genes

- There are several other X-linked traits in humans

X-Linked Genes	Example
Red-green color blindness 	Red-green color *blindness* affects approximately 4% of all human males. Red blindness is an inability to see red as a distinct color. Green blindness is an inability to see green as a distinct color. When normal (in this case, the dominant alleles are normal), these genes code for the production of proteins called opsins that help absorb different wavelengths of light. A lack of opsins causes insensitivity to light of red and green wavelengths.

Table 7-3 part 2 Biology: Science for Life, 2/e

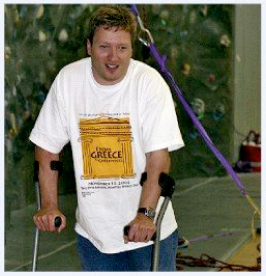
X-Linked Genes	Example
Muscular dystrophy 	*Muscular dystrophy* is a progressive, fatal disease of muscle wasting that affects approximately 1 in 3,500 males. The onset of muscle wasting occurs between 1 and 6 years of age, and by 12 years of age, affected boys are often confined to a wheelchair. The gene is one that normally codes for the dystrophin protein and is located on the X chromosome. When at least one allele is normal, dystrophin stabilizes cell membranes during muscle contraction. It is thought that the absence of normal dystrophin proteins causes muscle cells to break down and muscle tissue to die.

Table 7-3 part 3 Biology: Science for Life, 2/e