Human Genetics

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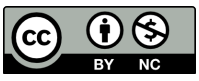


CHAPTER 1

Human Genetics

Lesson Objectives

- List the two types of chromosomes in the human genome.
- Predict patterns of inheritance for traits located on the sex chromosomes.
- Describe how some common human genetic disorders are inherited.
- Explain how changes in chromosomes can cause disorders in humans.

Check Your Understanding

- How many alleles does an individual have for each gene/trait?
- How do we predict the probability of traits being passed on to the next generation?
- What do we call complexes of DNA wound around proteins that pass on genetic information to the next generation of cells?

Vocabulary

- autosomes
- pedigree
- sex-linked inheritance
- sex-linked trait

Special Inheritance Patterns

What gene determines if a baby is male or female? How are humans born with genetic disorders, like cystic fibrosis or Down Syndrome? We can apply Mendel's rules to describe how many human traits and genetic disorders are inherited.

We can now also explain special inheritance patterns that don't fit Mendel's rules.

Sex-linked Inheritance

What determines if a baby is a male or female? Recall that you have 23 pairs of chromosomes—and one of those pairs is the sex chromosomes. Everyone has two sex chromosomes, X or Y. Females have two X chromosomes (XX), while males have one X chromosome and one Y chromosome (XY).

If a baby inherits an X from the father and an X from the mother, what will be the child's sex? Female. If the father's sperm carries the Y chromosome, the child will be male. Notice that a mother can only pass on an X chromosome,

so the sex of the baby is determined by the father. The father has a 50 percent chance of passing on the Y or X chromosome, so there is a 50 percent chance that a child will be male, and a 50 percent chance a child will be female.

One special pattern of inheritance that doesn't fit Mendel's rules is **sex-linked inheritance,** referring to the inheritance of traits that are located on genes on the sex chromosomes. Since males and females do not have the same sex chromosomes, there will be differences between the sexes in how these **sex-linked traits** —traits linked to genes located on the sex chromosomes —are expressed.

One example of a sex-linked trait is red-green colorblindness. People with this type of colorblindness cannot tell the difference between red and green. They often see these colors as shades of brown (see **Figure** 1.1 and **Table** 1.1). Boys are much more likely to be colorblind than girls. This is because colorblindness is a sex-linked recessive trait.

Boys only have one X chromosome, so if that chromosome carries the gene for colorblindness, they will be colorblind. As girls have two X chromosomes, a girl can have one X chromosome with the colorblind gene and one X chromosome with a normal gene for color vision. Since colorblindness is recessive, the dominant normal gene will mask the recessive colorblind gene. Females with one colorblindness allele and one normal allele are referred to as carriers. They carry the allele but do not express it.

How would a female become color-blind? She would have to inherit two genes for colorblindness, which is very unlikely. Many sex-linked traits are inherited in a recessive manner.

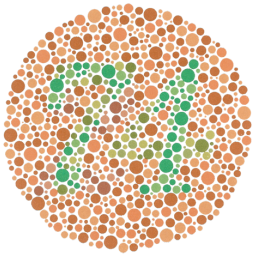


FIGURE 1.1

A person with red-green colorblindness would not be able to see the number.

TABLE 1.1: According to this Punnett square, the son of a woman who carries the colorblindness trait and a normal male has a 50% chance of being colorblind

	X^c	X
X	**X^cX** (carrier female)	**XX** (normal female)
Y	**X^cY** (colorblind male)	**XY** (normal male)

Human Genetic Disorders

Some human genetic disorders are also X-linked or Y-linked, which means the faulty gene is carried on these sex chromosomes. Other genetic disorders are carried on one of the other 22 pairs of chromosomes; these chromosomes are known as **autosomes** or autosomal (non-sex) chromosomes.

Autosomal Recessive Disorders

Some genetic disorders are caused by recessive or dominant alleles of a single gene on an autosome. An example of an autosomal recessive genetic disorder is cystic fibrosis. Children with cystic fibrosis have excessively thick mucus in their lungs, which makes it difficult for them to breathe. The inheritance of this recessive allele is the same as any other recessive allele, so a Punnett square can be used to predict the probability that two carriers of the disease will have a child with cystic fibrosis.

What are the possible genotypes of the offspring in **Table** 1.2? What are the possible phenotypes?

TABLE 1.2: According to this Punnett square, two parents that are carriers of cystic fibrosis of the cystic fibrosis gene have a 25% chance of having a child with cystic fibrosis

	F	**f**
F	**FF** (normal)	**Ff** (carrier)
f	**Ff** (carrier)	**ff** (affected)

Automsomal Dominant Disorders

Huntington's disease is an example of an autosomal dominant disorder. This means that if the dominant allele is present, then the person will express the disease.

The disease causes the brain's cells to break down, leading to muscle spasms and personality changes. Unlike most other genetic disorders, the symptoms usually do not become apparent until middle age. You can use a simple Punnett square to predict the inheritance of a dominant autosomal disorder, like Huntington's disease. If one parent has Huntington's disease, what is the chance of passing it on to their children? If you draw the Punnett square, you will find that there is a 50 percent chance of the disorder being passed on to the children.

Pedigree Analysis

A **pedigree** is a chart which shows the inheritance of a trait over several generations. A pedigree is commonly created for families, and it outlines the inheritance patterns of genetic disorders.

Figure 1.2 shows a pedigree displaying recessive inheritance of a disorder through three generations. From studying a pedigree, scientists can determine the following:

- If the trait is sex-linked (on the X or Y chromosome) or autosomal (on a chromosome that does not determine sex)
- If he trait is inherited in a dominant or recessive fashion, and possibly whether individuals with the trait are heterozygous or homozygous.

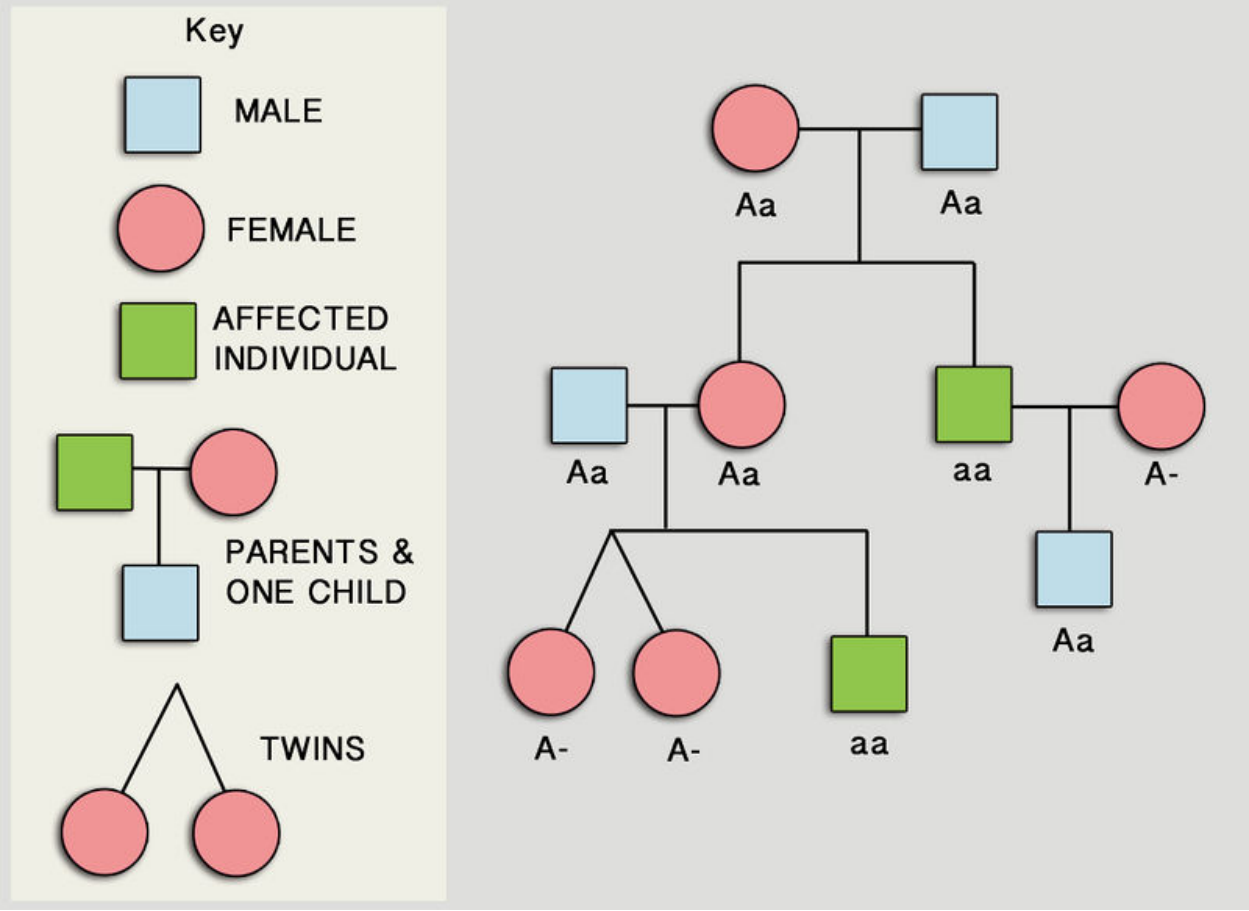
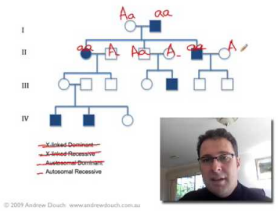


FIGURE 1.2

In a pedigree, squares symbolize males, and circles represent females. A horizontal line joining a male and female indicates that the couple had offspring. Vertical lines indicate offspring which are listed left to right, in order of birth. Shading of the circle or square indicates an individual who has the trait being traced. The inheritance of the recessive trait is being traced. "A" is the dominant allele and "a" is the recessive allele.

Pedigree analysis is discussed in http://www.youtube.com/watch?v=HbIHjsn5cHo (9:13).



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Chromosomal Disorders

Some children are born with genetic defects that are not carried by a single gene. Instead, an error in a larger part of the chromosome or even in an entire chromosome causes the disorder. Usually the error happens when the egg or sperm is forming. Having extra chromosomes or damaged chromosomes can also cause disorders.

Extra Chromosomes

One common example of an extra-chromosome disorder is Down syndrome (**Figure** 1.3). Children with Down syndrome are mentally disabled and also have physical deformities. Down syndrome occurs when a baby receives an extra chromosome from one of his or her parents. Usually, a child will receive one chromosome 21 from the mother and one chromosome 21 from the father. In an individual with Down syndrome, however, there are three copies of chromosome 21. Down syndrome is therefore also known as Trisomy 21.

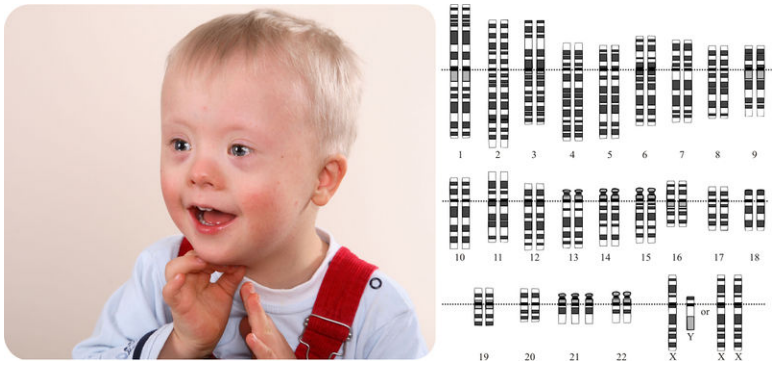

FIGURE 1.3

A child with Down syndrome.

Another example of a chromosomal disorder is Klinefelter syndrome, in which a male inherits an extra "X" chromosome. These individuals have underdeveloped sex organs and elongated limbs, and have difficulty learning new things.

Outside of chromosome 21 and the sex chromosomes, most embryos with extra chromosomes do not usually survive. Because chromosomes carry many, many genes, a disruption of a chromosome can cause severe problems with the development of a fetus.

Damaged Chromosomes

Chromosomal disorders also occur when part of a chromosome becomes damaged. For example, if a tiny portion of chromosome 5 is missing, the individual will have *cri du chat* (cat's cry) syndrome. These individuals have

misshapen facial features and the infant's cry resembles a cat's cry.

Lesson Summary

- Some human traits are controlled by genes on the sex chromosomes.
- Human genetic disorders can be inherited through recessive or dominant alleles, and they can be located on the sex chromosomes or autosomes (non-sex).
- Changes in chromosome number can lead to disorders like Down syndrome.

Review Questions

Recall

1. How many chromosomes do you have in each cell of your body?

Apply Concepts

2. How is Down's syndrome inherited?

Think Critically

3. A son cannot inherit colorblindness from his father. Why not?

4. One parent is a carrier of the cystic fibrosis gene, while the other parent does not carry the allele. Can their child have cystic fibrosis?

Further Reading / Supplemental Links

- http://www.articlesbase.com/health-articles/what-is-haemophilia-412305.html
- http://geneticdisorderinfo.wikispaces.com/
- http://www.hhmi.org/biointeractive/vlabs/cardiology/index.html

Points to Consider

- Human cloning is illegal in many countries. Do you agree with these restrictions?
- Why would it be helpful to know all the genes that make up human DNA?
- It may be possible in the future to obtain the sequence of all your genes. Would you want to take advantage of this opportunity? Why or why not?

References

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3. Left: Image copyright Tomasz Markowski, 2014; Right: Courtesy of the National Human Genome Research Institute. A child with Down syndrome has an extra chromosome 21 illustrated in a karyotype . Left: Used under license from Shutterstock.com; Right: Public Domain