

# 19 Human Genetics

## *Chapter Summary*

The information that is transferred from one generation to the next is stored in genes, which are sections of chromosomes that contain information for protein synthesis. A zygote receives twenty-three pairs of chromosomes, one of each pair from the male parent and the other of each pair from the female parent. Twenty-two of these pairs are autosomes. Autosomes are chromosomes that contain genes for the same traits. The twenty-third pair of chromosomes are sex chromosomes. Females inherit two X chromosomes and males one X and one Y as their pair of sex chromosomes. Sometimes individuals inherit an abnormal number of a particular type of chromosome. This is generally the result of failure of the chromosomes or chromatids to separate during meiosis. This type of error, which occurs during gamete formation, is called nondisjunction. Nondisjunction can cause Down syndrome, when an individual inherits three copies of chromosome number 21. It is also possible to inherit an abnormal number of sex chromosomes. This can result in Turner syndrome if only one X chromosome (and no Y) is inherited, a poly-x female if three X chromosomes are inherited, Klinefelter syndrome if two X chromosomes and a Y chromosome are inherited, or Jacob's syndrome if one X chromosome and two Y chromosomes are inherited. Characteristics associated with individuals exhibiting these abnormal chromosome numbers are discussed in the text. Just as individuals inherit pairs of chromosomes, they also inherit pairs of genes. Often, one form of a gene is dominant over another recessive form. Dominant genes are expressed when a single copy is present. Recessive genes are only expressed when they occur in pairs. Genes that occur on the sex chromosomes are called sex-linked genes. Given that males only have one X chromosome, they only have one copy of each gene associated with that chromosome. Because of this, males always express recessive genes occurring on their X chromosome. Genetic disorders result when an individual inherits abnormal, faulty genes. Dominant disorders occur when an individual inherits a copy of a faulty dominant gene, recessive disorders occur when an individual inherits two copies of a faulty recessive gene, and X-linked disorders involve inheriting faulty genes on the X chromosome. Researchers are investigating the possibility of curing genetic disorders by directly manipulating genes. This is called gene therapy. Researchers working in the field of genomics are performing a molecular analysis of the human genome, which contains all of the human genetic information. So far, a base sequence map has been completed. A genetic map that lists the locations of genes on chromosomes is under development.

## *Chapter Outline*

- I. Chromosomal Inheritance
  - A. Karyotyping
    1. Preparing the Karyotype
  - B. Nondisjunction
    1. Down Syndrome
  - C. Sex Chromosome Inheritance
    1. Too Many/Too Few Sex Chromosomes
- II. Genetic Inheritance
  - A. Inheritance of Genes on Autosomal Chromosomes
  - B. Sex-Linked Inheritance
  - C. Genetic Counseling
    1. Prenatal Testing for Genetic Disorders

- III. DNA Technology
  - A. Gene Therapy
  - B. Genomics
    - 1. The Base Sequence Map
    - 2. The Genetic Map

*Suggested Student Activities*

1. Discuss what is meant by a carrier and list some diseases in which carriers are involved.
2. Discuss the pros and cons of genetic engineering and gene therapy.
3. Discuss the genetic processes involved in sickle-cell disease.

*Answers to the Objective Questions*

- |                |                 |
|----------------|-----------------|
| 1. chromosomes | 6. one          |
| 2. chromosomes | 7. recessive    |
| 3. XY          | 8. recessive    |
| 4. three       | 9. gene therapy |
| 5. XXY         | 10. genomics    |

*Answers to Medical Terminology Reinforcement Exercise*

1. neo/genesis - new formation—a form of tissue regeneration
2. re/generat/ion - produce again or bring to life—the natural renewal of the structure as lost tissue
3. fetoscope - a device for listening for heart sounds of the fetus. It may also be a device for viewing the fetus inside the uterus.
4. chromo/some - body of color—the more readily stainable structure in the nucleus that transmits genetic information
5. poly/dys/plas/ia - condition of faulty development of many types of tissue, organs, or systems
6. con/genit/al - condition with birth—abnormality (without normal) existing at birth
7. auto/some - chromosome that is one of a pair that contain genes for the same traits

*Audiovisual Materials*

1. Film (16mm) - Laws of Heredity (15 min)(Encyclopedia Britannica Educational Corp.)
2. Film (16mm) - Biochemical Genetics in Man (The National Foundation - March of Dimes)
3. Film (16mm) - The Diagnosis of Hidden Congenital Abnormalities (The National Foundation - March of Dimes)

## Reference to Audiovisual and Computer Software Materials

Altschul Group Corporation (AGC)  
(also known as Perennial Education, Inc.)  
930 Pitner Ave.  
Evanston, IL 60202  
(800) 323-9084

Ambrose Video Publishing (AVP)  
1290 Avenue of the Americas, Suite 2245  
New York, NY 10104  
(212) 696-4545

Arthur Mokin Productions, Inc. (AMP)  
P.O. Box 1866  
Santa Rosa, CA 95402 or  
1738 Wright St.  
Santa Rosa, CA 95404  
(707) 542-4868

Carolina Biological Supply Company ©  
2700 York Rd.  
Burlington, NC 27215  
(800) 334-5551

Center for Humanities (CFH)  
Communications Park, Box 1000  
Mt. Kisco, NY 10549  
(800) 431-1242

Chariot Software Group (CSG)  
3659 India St., Suite 100C  
San Diego, CA 92102  
(800) 242-7468

Churchill Films (Ch-F)  
12210 Nebraska Ave.  
Los Angeles, CA 90025  
(213) 207-6600

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108 Wilmont Rd.  
Deerfield, IL 60015  
(800) 621-2131

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Elmira, NY 14905  
(607) 732-1090

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P.O. Box 2805  
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(800) 848-2050

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(EBE)  
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Chicago, IL 60611  
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Burr Ridge, IL 60521  
(800) 955-1177

HarperCollins Publishers (HC)  
Keystone Industrial Park  
Scranton, PA 18512  
(800) 242-7737

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P.O. Box 1029 (mailing address)  
Madison, WI 53701 or  
125 E. Gilman St.  
Madison, WI 53703  
(800) 422-4295

Human Relations Media (HRM)  
175 Tompkins Ave.  
Pleasantville, NY 10570  
(800) 431-2050

Insight Media (IM)  
121 West 85th St.  
New York, NY 10024  
(212) 721-6316  
Fax: 212-799-5309

Intellimation (INT)  
Dept. 2HF, 130 Cremona Drive  
P.O. Box 1530  
Santa Barbara, CA 93116  
(800) 346-8355  
Fax: 805-968-8899

International Film Bureau, Inc. (IFB)  
322 South Michigan Ave., Suite 522  
Chicago, IL 60604  
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Marty Stouffer Productions (MS)  
300 S. Spring St.  
Aspen, CO 81611  
(303) 925-5536

Milner Fenwick, Inc. (MF)  
2125 Greenspring Dr.  
Timonium, MD 21093  
(800) 638-8652

National Audiovisual Center (NAC)  
8700 Edgeworth Dr.  
Capital Heights, MD 20743  
(800) 638-1300

National Geographic Society (NG)  
Educational Services  
Department #90  
Washington, D.C. 20036  
(800) 368-2728

Phoenix Films, Inc. (PHO)  
468 Park Avenue South  
New York, NY 10016  
(800) 221-1274

Projected Learning Programs, Inc. (PLP)  
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Paradise, CA 95967-3008  
(800) 248-0757

Pyramid (PYR)  
P.O. Box 496  
Media, PA 19063  
(800) 523-0118

Simon & Schuster Prentice-Hall (SSPH)  
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Old Tappan, NJ 07675  
(201) 767-5000

Taped Technologies (TT)  
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(800) 995-1110

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Ridgefield, NJ 07657

Trinity Software (TS)  
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University of California Extension (UC)  
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Berkeley, CA 94704  
(510) 642-0460

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San Diego, CA 92130  
(619) 453-5000