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Why Is a Knowledge of Medical Genetics Important for Today's Health-Care Practitioner?

There are several reasons health-care practitioners must understand medical genetics. Genetic diseases make up a large percentage of the total disease burden in pediatric and adult populations (Table 1.1). This percentage will continue to grow as our understanding of the genetic basis of disease grows. In addition, modern medicine is placing increasing emphasis on prevention. Because genetics provides a basis for understanding the fundamental biological makeup of the organism, it naturally leads to a better understanding of the disease process. In some cases, this knowledge can lead to prevention of the disorder. It also leads to more effective disease treatment. Prevention and effective treatment are among the highest goals of medicine. The chapters that follow provide many examples of the ways genetics contributes to these goals. But first, this chapter reviews the foundations upon which current practice is built.

Genetics as it is known today is largely the result of research performed during the 20th century. Mendel's principles were independently rediscovered in 1900 by three different scientists working in three different countries. This was also the year in which Landsteiner discovered the ABO blood group system, in 1902, Archibald Garrod described alkaptonuria as the first "inborn error of metabolism," in 1909, Johannsen coined the term gene to denote the basic unit of heredity.

The next several decades were a period of considerable experimental and theoretical work. Several organisms, including *Drosophila melanogaster* (fruit fly) and *Neurospora crassa* (bread mold) served as useful experimental systems in which to study the actions and interactions of genes. For example, H. J. Muller demonstrated the genetic consequences of ionizing radiation in the fruit fly. During this period, much of the theoretical basis of population genetics was developed by three central figures: Ronald Fisher, J. B. S. Haldane, and Sewall Wright. In addition, the modes of inheritance of several important genetic diseases, including phenylketonuria, sickle-cell disease, Huntington disease, and cystic fibrosis, were established. In 1944, Oswald Avery showed that genes are composed of deoxyribonucleic acid (DNA).

Probably the most significant achievement of the 1950s was the specification of the physical structure of DNA by James Watson and Francis Crick in 1953. Their seminal paper, which was only one page long, formed the basis for