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CHAPTER 19**Ethical and Social Issues****in Genetics and Genomics 383***Principles of Biomedical Ethics, 383**Ethical Dilemmas**Arising in Medical Genetics, 383**Privacy of Genetic Information, 386**Eugenic and Dysgenic**Effects of Medical Genetics, 388**Genetics in Medicine, 390***THE BIRTH AND DEVELOPMENT OF GENETICS AND GENOMICS**

few areas of science and medicine are moving as quickly at the pace we are experiencing in the related fields of genetics and genomics. It may appear surprising to many students today, then, to learn that an appreciation of the role of genetics in medicine dates back well over a century, to the recognition by the British physician Sir Richard Garrad and others that Mendel's laws of inheritance could explain the recurrence of certain clinical disorders in families. During the ensuing years with developments in cellular and molecular biology, the field of medical genetics grew from a small clinical subspecialty concerned with a few rare hereditary disorders to a recognized medical specialty whose concepts, approaches are important components of the diagnosis and management of many disorders, both common and rare.

At the beginning of the 21st century, the Human Genome Project provided a virtually complete sequence of human DNA—our genomic code written in letters from the Greek for “all” or “complete”—which now serves as the foundation of efforts to catalogue all human genes, understand their structure and regulation, determine the extent of variation in these genes in different populations, and understand how genetic variations contribute to disease. The human genome of any individual can now be studied in its entirety, rather than only a part at a time. These developments are making possible the field of genomic medicine, which seeks to apply large-scale analysis of the human genome and its products, including the control of gene expression, human gene variation, and interactions between genes and the environment, to medical care.

GENETICS AND GENOMICS IN MEDICINE**The Practice of Genetics**

The medical geneticist is usually a physician who works as part of a team of health-care providers, working with other physicians, nurses and genetic counselors to evaluate patients for possibly hereditary diseases. They characterize the patient's illness through careful history taking and physical examination, assess possible modes of inheritance, arrange for diagnostic testing,

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develop the patient's management plan, and participate in research to determine ways to test for the disorder.

However, while Principles and Genomics are not restricted to case studies, a significant portion of the text, chapters 10 and 11, and portions of several of the other chapters, are devoted to the examples of the genetics and genomics as applied to medicine today.

- A pediatrician explains a child with multiple congenital malformations and enters a high-resolution genotyping system to determine if chromosomal deletions or duplications are below the level of resolution of standard diagnostic methods (Case 1).
- A forensic pathologist explains the hereditary aspect of cancer, the genetic basis of the disease, and the impact of the environment on the disease (Case 2).
- A geneticist explains a child's condition, the sample taken from the child's cells, and the results of the analysis in the context of the child's condition (Case 3).
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- A geneticist explains a child's condition, the sample taken from the child's cells, and the results of the analysis in the context of the child's condition (Case 5).
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