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CHAPTER 1

The Cell as a Unit of Health and Disease

Richard N. Mitchell

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Pathology literally translates to the study of *suffering* (Greek *pathos* = suffering, *logos* = study); more prosaically, the term *pathology* is invoked to represent the study of *disease.* Germane to this opening chapter, Virchow coined the term *cellular pathology* to emphasize the basic tenet that all diseases originate at the cellular level. Thus, modern pathology is basically the study of *cellular* abnormalities. Therefore, diseases and the underlying mechanisms are best understood in the context of *normal* cellular structure and function.

It is unrealistic (and even undesirable) to condense the vast and fascinating field of cell biology into a single chapter. Moreover, students of biology are likely quite familiar with many of the broader concepts of cell structure and function. Consequently, rather than attempting a comprehensive review, our goal is to survey some basic principles and highlight some recent advances that are relevant to the pathologic basis of disease that is emphasized throughout the text. We hope this chapter will be useful to review key topics in normal cell biology as they apply to the areas of Pathology that are covered from Chapter 2 onwards.

The Genome

The sequencing of the human genome represented a landmark achievement of biomedical science. Published in draft form in 2001 and more completely detailed in 2003, the information has already led to remarkable advances in science and medicine. Since then there has been an exponential decrease in the cost of sequencing and an exponential increase in data accrual; this new information, now literally at our fingertips, promises to revolutionize our understanding of health and disease. However, the sheer volume of the data is formidable, and there is a dawning realization that we have only begun to scratch the surface of its complexity; uncovering the relevance to disease and then developing new therapies remain challenges that both excite and inspire scientists and the lay public alike.

Noncoding DNA

The human genome contains roughly 3.2 billion DNA base pairs. Within the genome there are about 20,000 protein-encoding genes, comprising only about 1.5% of the genome. These proteins variously function as enzymes, structural components, and signaling molecules and are used to assemble and maintain all of the cells in the body. Although 20,000 is an underestimation of the number of proteins encoded in the human genome (given that many genes produce multiple RNA transcripts encoding different protein isoforms), it is nevertheless startling to realize that worms composed of fewer than 1000 cells—with genomes of only about 0.1 billion DNA base pairs—are also assembled using about 20,000 genes to produce proteins. Even more surprising is that many of these proteins are recognizable homologs of molecules expressed in humans. What then separates humans from worms?

The answer is not completely known, but the weight of current evidence suggests that much of the difference

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