

## PREFACE

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In the last few years a new word has entered the medical and scientific vocabulary. This word, *channelopathy*, describes those human and animal diseases that result from defects in ion channel function. Ion channels are membrane proteins that act as gated pathways for the movement of ions across cell membranes. They play essential roles in the physiology and pathophysiology of all cells and it is therefore not very surprising that an ever increasing number of human and animal diseases have been found to be caused by defective ion channel function. This book aims to provide an informative and up to date account of our present understanding of ion channels and the molecular basis of ion channel diseases. It is intended for graduates and final year undergraduates in the medical and biological sciences. I hope it will also be of value to clinicians who wish to know more about the molecular basis of the channelopathies, and to research workers interested in the physiological role of ion channels and the diseases that result from their defective function.

To understand how ion channel mutations give rise to human and animal disease requires at least a basic understanding of the genetics, molecular structure, biophysical properties and physiological role of ion channels. It is difficult to give a comprehensive review of all these areas in a single book. However, I have provided a basic introduction to the relevant aspects of molecular biology and biophysics in the next two chapters, for the benefit of those unfamiliar with these topics. I have also included a brief description of the principal methods used to study channelopathies (Chapter 4). For more detailed descriptions of these topics readers are advised to consult one of the many excellent textbooks in the field, such as those by Hille (1992) or Aidley & Stanfield (1996). Chapters 5-23 each consider a single class of ion channel, while Chapter 24 includes a medley of channels which are distinct from those discussed in earlier chapters but about which too little is known to warrant an individual chapter. In each chapter, the initial section summarizes the physiological roles, subunit composition, molecular structure and chromosomal location of the channel in question. The relationship between channel structure and function is then considered. The third section describes those diseases associated with defective channel structure and/or regulation. For those channelopathies that result from mutations in ion channel genes, I consider how the mutation(s) affects channel function and to what extent this change in channel function can account for the clinical phenotype.

Identification of new ion channel disorders is progressing at an ever increasing rate. Many were identified during the course of writing this book, so that at times I felt like the Red Queen in Alice in Wonderland, who had

# Preface To Donne

**Lauren Gardner**



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