
Human Pathobiochemistry

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Editors

Human Pathobiochemistry

From Clinical Studies to Molecular
Mechanisms

 Springer

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Preface

In just the past few decades, advances in molecular biology, cell biology, and biochemistry have allowed biomedical researchers to identify the mechanisms of action that belie many processes in our physiology. So much of what has been learned about these biochemical processes or mechanisms has been learned from studying human disease. More than ever, we need researchers and clinicians to be trained to think past the symptoms and seek to understand the underlying molecular or biochemical pathology that presents in a patient. Much of the research done on these diseases in the clinic and laboratory have led to the discovery of novel, targeted therapies. The shift in clinical practice is already happening. The question being asked is no longer only “how do I treat these symptoms” but also “how can we target the cause of the disease?” This culture shift in medicine will surely beget a generation of clinicians who practice medicine in a more informed way and contribute to elucidating new therapies. The purpose of this text is to teach clinicians-in-training to understand the pathology not only on a topical level but a deep understanding of the molecular underpinnings of human disease.

This textbook uses a case study approach to present the core principles of biochemistry and molecular biology in the context of human disease to students who will be involved in patient care. The 29 clinical cases are carefully selected to cover key scientific concepts and some common, and other not so common, diseases. This textbook covers many topics on metabolic disease as a major part but contains other topics on connective tissue disorders, neurological disorders, auto-inflammatory disorders, infective diseases, cancer, etc. Each chapter provides a specific patient report that includes the natural history, pertinent clinical laboratory data, physical findings, subsequent diagnosis, and therapy. This is followed by a comprehensive discussion of the normal biochemical processes and reactions pertaining to the case, along with the pathophysiological mechanisms of the disease.

The book is organized into three parts, the first of which considers diseases of metabolism. The diseases range from those that might already be familiar to a student, such as familial hypercholesterolemia and phenylketonuria (PKU), to others that are more rare, such as Fabry or Wilson disease. The second part of the book focuses on genetic disorders. The manifestations of the genetic disorders included are myriad. In some cases, the genetic insult results in physical deformities, such as achondroplasia, Marfan syndrome, and cherubism. In others, the result can be malignant disease, such as acute myeloid leukemia. The last section encompasses a host of other diseases,

infections, and conditions which can now also be studied to the molecular level. Some of these have known etiologies; others are still under investigation. Hopefully, clinicians-in-training will appreciate the opportunity to think about all conditions as having a molecular basis and thereby have a target to which a therapy can be developed. Again, this book would hope to emphasize the importance of the *process* of diagnosis, as it relates to the biochemistry of the patient, rather than memorizing a list of symptoms for each disease.

This textbook has several unique features which we hope will enable instructors to get the most out of using it in a classroom setting. The chapter authors have provided discussion questions meant to help students in identifying important information or consider why therapies will or will not work based on the understanding of the pertinent biochemistry. The text is also meant to be usable by many different clinical students or even those in basic research. It provides various clinical cases for preclinical students in medical school and schools for other health professionals, including nurses, nutritionists, physiotherapists, or radiographers, for example. In all of these settings, we hope the instructors will use the discussion questions in various modalities, including techniques such as active deep learning, small group discussion (SGD), and/or problem-based learning (PBL). These teaching methods have been experimentally shown to improve student learning. In its design, this text has an advantage over a standard textbook where content delivery is emphasized, in that this text includes studying the *approach* clinicians and researchers used to discover the mechanism of disease. Instructors and students can also study similarities and differences in techniques and methodology between cases, in addition to content targeted to each molecular pathology being investigated. Inherently, it emphasizes the role and importance in “bench-to-bedside” training of health-care professionals. We hope that it encourages independent inquiry outside of the textbook, in that ongoing research of disease mechanisms and of novel therapeutic approaches are constantly being updated, changed, and improved. We hope that a new text with this approach will have wide appeal in many disciplines related to pathobiochemistry.

Okayama University Medical School has a long history of experience in delivering a pathobiochemistry tutorial course. Since its initiation in the 1990s by Prof. Ninomiya, it has conducted the course in English for approximately 30% of the students each year. The students and instructors use English in all discussions, written assignments, and presentations. The faculty who have been involved in the course have unanimously agreed on the need of a new pathobiochemistry text dedicated to a *project*-based learning for the clinician in training. Recently our school has begun accepting undergraduate or graduate (Master’s level) students from other Asian countries (e.g., China, Korea, Indonesia, and Myanmar) as part of exchange programs. Their inclusion in the English-speaking student groups provides a positive stimulus for Japanese students to communicate in English. Furthermore, the faculty from those countries have also joined the course as tutors. It is therefore likely that their experience may translate in using the same English textbook for their own teaching. This international collaboration has clearly benefitted all involved.

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