



Rare Diseases and Orphan Drugs: Keys to Understanding and Treating the Common Diseases

By Jules J. Berman

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Rare Diseases and Orphan Drugs shows that much of what we now know about common diseases has been achieved by studying rare diseases. It proposes that future advances in the prevention, diagnosis, and treatment of common diseases will come as a consequence of our accelerating progress in the field of rare diseases.

Understanding the complex steps in the development of common diseases, such as cancer, cardiovascular disease, and metabolic diseases, has proven a difficult problem. Rare diseases, however, are often caused by aberrations of a single gene. In rare diseases, we may study how specific genetic defects can trigger a series of events that lead to the expression of a particular disease. Often, the disease process manifested in a certain rare disease is strikingly similar to the disease process observed in a common disease.

This work ties the lessons learned about rare diseases to our understanding of common ones. Chapters covering the number of common diseases are minimized, while rare diseases are introduced as single diseases or as members of diseases classes. After reading this book, readers will appreciate how further research into the rare diseases may lead to new methods for preventing, diagnosing, and treating all diseases, rare or common.

- Makes rare diseases relevant to clinicians and researchers by tying lessons learned about the rare diseases to our understanding of the common diseases
- Stresses basic pathologic mechanisms that account for human disease (e.g., disorders of cell development, replication, maintenance, function and structure), that can be understood without prior training in pathology
- Discusses advanced concepts in molecular biology and genetics in a simple, functional context appropriate for medical trainees and new researchers
- Offers insights into how further research into rare diseases may lead to new methods for preventing, diagnosing, and treating all diseases.

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Editorial Review

Review

"This book may appeal to clinicians and researchers by drawing on lessons learnt about rare diseases and their relevance for common diseases...covers disease pathological mechanisms, understandable to non-experts, concepts in molecular biology and genetics, and how future research into rare diseases may provide answers for the treatment of all diseases."--***Pediatrics Institute online, June 18, 2014***

"The book flows well and makes good use of illustration, making it an engaging read...The first of its kind, this book provides justification for funding research in the rare diseases. This book is strongly recommended for the intended audience. 3 Stars - Score: 86"--***Doody's.com, Nov 14 2014***

"This book about rare diseases is a rare book. Few authors have the breadth of knowledge to cover the field of rare diseases. Jules J. Berman, well known in medical informatics, tutors the reader with a mix of history, genetics, medicine and social commentary on common versus rare disorders. The book is readable, interesting and unexpectedly entertaining. It is not a textbook or a compendium, but for anyone interested in rare diseases generally it is a unique and informative read."

-M. Ian Phillips, Editor, Expert Opinion on Orphan Drugs, Director, Center for Rare Disease Therapies, Keck Graduate Institute, Claremont, CA, USA

"At last, a book that makes these complex matters comprehensible. Dr. Berman has produced a very significant work here. He manages to bring together the complexities of genetics and inheritance and the complex relationship of rare diseases to common diseases, and weave them together to explain many things about human biology and disease. His use of over 100 rules, with a rationale for each, is a novel and very useful way of summarising key points of his work. These rules will be a valuable aid for students as well as lay readers wanting to better understand this complex area."

-John Forman, Executive Director, NZ Organisation for Rare Disorders, Wellington, New Zealand

"Rare Diseases and Orphan Drugs: Keys to Understanding and Treating the Common Diseases bridges our understanding of the common diseases and the rare diseases. This unique and much-needed book provides an insightful glimpse of how biomedical research will play out as the rare diseases take an increasing role in the way we understand and treat the common diseases. Healthcare professionals, students, biomedical researchers, and advocates for rare disease research will find that this book applies common sense to a rare subject."

-Stephen G. Groft, Pharm.D., Director of the Office of Rare Diseases Research, U.S. National Institutes of Health, 1993-2014 (retired)

From the Back Cover

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About the Author

Jules Berman holds two bachelor of science degrees from MIT (Mathematics, and Earth and Planetary Sciences), a PhD from Temple University, and an MD, from the University of Miami. He was a graduate researcher in the Fels Cancer Research Institute, at Temple University, and at the American Health Foundation in Valhalla, New York. His post-doctoral studies were completed at the U.S. National Institutes of Health, and his residency was completed at the George Washington University Medical Center in Washington, D.C. Dr. Berman served as Chief of Anatomic Pathology, Surgical Pathology and Cytopathology at the Veterans Administration Medical Center in Baltimore, Maryland, where he held joint appointments at the University of Maryland Medical Center and at the Johns Hopkins Medical Institutions. In 1998, he transferred to the U.S. National Institutes of Health, as a Medical Officer, and as the Program Director for Pathology Informatics in the Cancer Diagnosis Program at the National Cancer Institute. Dr. Berman is a past President of the Association for Pathology Informatics, and the 2011 recipient of the association's Lifetime Achievement Award. He is a listed author on over 200 scientific publications and has written more than a dozen books in his three areas of expertise: informatics, computer programming, and cancer biology. Dr. Berman is currently a free-lance writer.

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