

Book Review

Vogel and Motulsky's Human Genetics — Problems and Approaches

Michael Speicher, Stylianos E. Antonarakis, Arno G. Motulsky (eds)

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Human genetics is a vast and fascinating field that is evolving rapidly with the advent of new techniques in molecular biology and genomics. In its fourth incarnation, *Vogel and Motulsky's Human Genetics — Problems and Approaches* presents an up-to-date and extensively revised edition of the concepts and technology that are shaping our modern understanding of human genetics.

Human genetics is both a fundamental and an applied science. This makes it a complex and perhaps difficult proposition for writing text books, in that a cogent text would require the expertise of cytogeneticists, epidemiologists, population geneticists, physicians, human and medical geneticists, molecular biologists, computational biologists, etc. Vogel and Motulsky themselves wrote and edited the three earlier editions of this book, which is widely regarded as a key reference and textbook in the area of human and medical genetics. During the production of the fourth edition of this book, a new editorial team was assembled. Michael Speicher (Medical University of Graz, Austria), Stylianos Antonarakis (University of Geneva Medical School, Switzerland) and Arno Motulsky (University of Washington School of Medicine, USA) have done an excellent job as editors. They have extended, revised and added many of the new insights into the genetic basis of development and function in human health and disease that have emerged in the ten years since the publication of the third edition.

Unlike the earlier editions, all chapters in this edition were written by renowned geneticists (including the editors) who are specialists/experts in the topic discussed in each chapter, thus lending a unique insight to each chapter. The book starts out with an update on the history of human genetics, followed by a discussion

of the human genome sequence and variation. The chapter on chromosomes elegantly links the classical ideas and techniques of cytogenetic analysis to the modern techniques of detecting karyotypic abnormalities (structural or numerical) associated with human chromosomes (like fibre-fluorescence in situ hybridisation [FISH]- and array-based methods). Another chapter describes the currently popular high-throughput 'omics' approaches.

The next five chapters are dedicated to introducing the reader to Mendelian genetics as applied to the inheritance of genetic traits in humans. This includes everything from modes of inheritance to concepts of linkage and their implications for inheritance, monogenic versus oligogenic traits, multifactorial diseases in humans and findings from genome-wide association studies.

Epigenetics has garnered a lot of attention recently, and this edition includes chapters dedicated to the effects of chromatin remodelling, imprinting, etc. on the normal inheritance of genetic traits and on diseases, including cancer. This edition also contains dedicated chapters on haemoglobinopathies, infectious diseases and cancer genetics, reflecting the many advances made in the diagnosis and treatment of these diseases over the ten years since the previous edition.

The sections on population genetics and human evolution have now been expanded to include chapters dedicated to the discussion of comparative genomics, the genomics of population structure and epidemiology.

This edition also contains several new chapters on pharmacogenetics, gene therapy, gene cloning, genetic medicine, genetic counselling and prenatal diagnosis, which are fast-emerging topics that are not part of many introductory level textbooks. These areas have widespread medical, social, ethical and legal ramifications and thus are of interest to students of diverse disciplines such as medicine, human genetics, other biomedical sciences, medico-legal studies, pharmacology and sociology.

This edition also includes several chapters on databases for basic and clinical genetics, which should benefit many students and faculty members alike. Using the guidelines and explanation of database organisation and information, students will learn a lot about their purpose and applications and will be able to extract more information from their exploration of these websites.

Similarly, the behavioural genetics section has been expanded considerably to include genetic studies of

cognitive dysfunction, neurodegenerative disorders and the genetic basis of alcoholism and addictions. These are complex concepts that cross-link genetics with neural development and function and with psychology/social science. These chapters discuss modern approaches to studying the genetic basis of personality and several complex disorders in a concise manner. They bring out the pressing questions that limit our understanding/knowledge at this time.

Interestingly, although the understanding of genetics and molecular biology has advanced tremendously from the study of model organisms, the effect of these advances on human genetics are often under-appreciated in text books. Research done on human subjects has informed our knowledge, but ethical and other concerns have led to the development of several animal models as experimental systems of choice to understand the pathogenesis of diseases or normal gene function. In this book five different animal models, ranging from flies, mice, the nematode *Caenorhabditis elegans*, fish and

dogs, have been described for their individual advantages, repertoire of genetic tricks and tools that have been developed in the past several decades, along with applications of animal models to the study of genetics and to human genetics.

Overall this is an excellent reference and textbook that should interest human and medical geneticists, scientists in biomedical research, physicians, epidemiologists and students at undergraduate, graduate and post-graduate levels. The illustrations and pictures are excellent and highly informative. The authors and editors have done an excellent job of writing short and concise chapters, using good illustrations and examples that are easy to follow and written with care, so that new and old readers alike will find it an interesting and thought-provoking read.

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