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Book Review
Human Genetics 2nd Edition

By: Anne Gardner and Teresa Davies

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The field of human genetics is a rapidly evolving area, often spurred on by changes and advances in technology. This revised edition of Human Genetics covers an impressive variety of topics within the field, and would certainly be suitable for an audience ranging from basic science undergraduates to people working within genetics in the medical profession.

The book starts off with the expected and necessary sections on DNA structure, replication, and mechanisms of gene expression. From personal preference, the third chapter (Mechanisms of Disease) may have been better placed after the subsequent chapter on Patterns of Inheritance, but the clear descriptions associated with each section make it easy for readers to pick and choose the order in which they want to study the material. Subsequent chapters cover the techniques used in the traditional areas of cytogenetics and molecular genetics, with a useful intervening chapter on the more molecular aspects of cytogenetics, including the relatively recent uptake of array-based methods in clinical cytogenetics. This leads nicely into the final sections on prenatal diagnosis, genetic counselling and ethical issues, which provide a necessary appreciation of the implications of this technology.

The material in this book is clearly laid out, and provides a comprehensive overview of the subject. The explicit list of learning objectives at the start of each chapter will be valued by students and teachers alike, and the explanatory sentence or two after each one provides a quick reference to the key take-home messages. Within the text, important facts and definitions are highlighted in boxes, and reinforced by straightforward and logical figures. The authors acknowledge that this is an introductory text, but also provide key references to further reading, enabling readers to easily expand their knowledge in specific areas of interest, yet achieving a fundamental grasp of the areas being presented. This further reading, along with the incredibly valuable self-assessment questions, will allow students to identify and address the limits of their knowledge.

The emphasis on genetic disorders throughout the book will make it relevant to those studying the science of human genetics, but also to medical students throughout their studies. The glossary of disorders at the end of the book provides an excellent quick reference point, and the comprehensive list of internet sites directs the reader to many other well recognised resources for further information. In the age of information overload via the internet, such a list is incredibly valuable for helping students assess the quality of the information they are retrieving from online sources.

With the emphasis on those areas most relevant to a diagnostic laboratory, an area lacking in coverage is that of complex multifactorial diseases such as coronary artery disease or diabetes, which recent genome-wide association studies have shown to have a genetic component. However, this shouldn't detract from the overall usefulness of the text. This book will appeal to anyone with an interest in human genetics, particularly those approaching the subject from the point of view of a diagnostic laboratory.