Essentials of Human Genetics
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Second Edition

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Published by
Jitendar P Vij
Jaypee Brothers Medical Publishers (P) Ltd

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Essentials of Human Genetics
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First Edition: 2002
Second Edition: 2009
ISBN/978-81-8448-535-6
Typeset at JPBMP typesetting unit
Printed at Ajanta Offset & Packagings Ltd., New Delhi
To
My Granddaughter
Sree
Preface to the Second Edition

Medical genetics is one of the most rapidly advancing branch of medicine and is recognized as a clinical specialty which provides an insight into the functioning of the human body, in health and disease. Understanding the principles of human genetics is an important component in the diagnosis, management and prevention of several medical disorders.

To be a good medical practitioner, every medical student and professional needs to know the fundamentals of human genetics. The book is written with this view in mind and covers various basic aspects of the field. This is written after fifteen years of teaching experience in a medical college in the field of anatomy, embryology and genetics and twenty eight years of working experience in the clinical and laboratory aspects of medical genetics, and includes cases I have seen over the years.

Today, the treatment of genetic disorders is within reach due to advances in biotechnology. However, this is not available for most genetic disorders. As of now, the emphasis in medical genetics is on prenatal diagnosis, including presymptomatic testing and screening in pregnancy, the neonatal period and population screening.

Identification of genes responsible for human disease is a key factor in the progress in medical genetics. In February 2001, a major landmark was the mapping of 95% of the human genome, and the progress and benefits of this achievement are reviewed. Lastly but importantly, the ethical, legal and social issues concerning this field need to be understood and studied in context to the law of the land.

Self test is the best test to learn how much you know. Multiple-choice questions can quench this thirst. Most importantly, it is the work of other scientists and genetic
professionals whose published works help us to substantiate our knowledge and help as reference.

I am confident that the basics given in the book will open minds of the readers to this exciting branch of new medicine. However, medicine is an everchanging science with continuous research and clinical experience altering the management. The efforts made in the making of this book, though are after the proper review to best of my ability, I disclaim all the responsibility of any errors or omissions and readers are therefore encouraged to confirm the same before putting into actual practice.

Hema Purandarey
Medical genetics is one of the most rapidly advancing branch of medicine and is recognised as a clinical speciality which provides an insight into the functioning of the human body, in health and disease. Understanding the principle of human genetics is an important component in the diagnosis, management and prevention of several disorders.

To be a good medical practitioner, every medical student and professional needs to know the fundamentals of human genetics. This book is written with this view in mind, and covers various basic aspects of the field. It was written after fifteen years of my teaching experience in a medical college and twenty years of working experience in the clinical and laboratory aspects of medical genetics, and includes cases I have seen over the years.

The initial chapters cover the brief history and burden of genetic diseases and factors predisposing to Mendelian and multifactorial diseases. The application of this knowledge in the diagnosis and prevention of genetic disease is also discussed. Chromosomes are the basic units of heredity and methods of their studies and types of abnormalities are discussed next. The disorders occurring due to these abnormalities are discussed later. The structure of DNA and factors altering DNA structure leading to various diseases, and methods of analysis are discussed next. The chapter on cancer genetics deals with constitutional and acquired genetic changes leading to malignant disorders.

Inborn errors of metabolism form an important group of disorders in pediatric practice and the chapter on biochemical genetics deals with the causes and classification of these disorders.

Preface to the First Edition

Medical genetics is one of the most rapidly advancing branch of medicine and is recognised as a clinical speciality which provides an insight into the functioning of the human body, in health and disease. Understanding the principle of human genetics is an important component in the diagnosis, management and prevention of several disorders.

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Inborn errors of metabolism form an important group of disorders in pediatric practice and the chapter on biochemical genetics deals with the causes and classification of these disorders.
There are many congenital malformations which arise due to environmental insults or infections or teratogens, in addition to those occurring sporadically. These are described in the chapter on dysmorphology and congenital malformations.

Today the treatment of genetic disorders is within reach due to advances in biotechnology. However, this is not available for most genetic disorders. As of now, the emphasis in medical genetics is on prenatal diagnosis, including presymptomatic testing and screening in pregnancy, the neonatal period and population screening. These aspects are dealt with in the respective chapters.

An important chapter is that of genetic counseling as it is the first step in the patient’s medical history for arriving at a preliminary diagnosis and planning and discussing management options.

The chapter on population genetics deals with methods to study populations and statistical methods to estimate the incidence and recurrence of genetic diseases.

Identification of genes responsible for human diseases is a key factor in the progress in medical genetics. In February 2001, a major landmark was the mapping of 95% of the human genome, and the progress and benefits of this achievement are reviewed in the chapter on the human genome project. Lastly but importantly, the ethical, legal and social issues concerning this field are briefly discussed. Multiple-choice questions cover some important aspects of the field, and answers to the questions with the explanations have been provided.

This book covers the basics of medical genetics and references are provided at the end of the book for further reading. I hope this text provides something of interest to every reader; that it will be a basic introduction to those new to the field of medical genetics, and will provide a useful reference to those more experienced in the field of medical genetics.

Hema Purandarey
Acknowledgements

Dr Smita Purandare for so zealously going through and editing the entire manuscript and Dr Usha Desai, Dr Shilpa Purandare, Dr Anil Jalan, Dr Shrikant Purandare, Dr Madan Naik for their inputs.

My staff, technicians and research students who have willingly helped me at all stages.
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