
This is a new and extensively rewritten edition of a respected textbook. Dr Cohen has significantly reorganised and updated the book to reflect the monumental changes which have occurred in the field since the first edition was published in 1982. The book begins with essential chapters on nomenclature, followed by chapters on possible approaches to the clinical assessment of dysmorphic patients, basic genetics, teratology, syndrome classification, delineation and diagnosis, abnormal growth and development, mental retardation, and finally on psychological and social implications of birth defects. It has no pretensions towards being a comprehensive atlas of birth defect syndromes. Instead, as Robert Gorlin states in the foreword, it is a great "how-to" book. Dr Cohen clearly describes a rational, critical approach to the problems of syndrome delineation and diagnosis. In every chapter there are concepts which will allow the reader to refine his or her own diagnostic skills, to understand the fundamental processes by which new syndromes are delineated, and to be better equipped to appraise the presentations in the field critically. Throughout, the book is highly readable and the text is supplemented by many informative and often entertaining illustrations. There is a school of thought which says that great dysmorphologists are born rather than made. This book will certainly help those of us lacking the requisite genes to reach a competent level.

Evan Reid


The lysosome is an important cellular organelle involved in the intracellular digestion of macromolecules. When something goes wrong with this system, such as the deficiency of the digestive enzyme (e.g. acid hydrolase, lysosomal enzymes are over 40) or transport protein, the organelle becomes "constipated". The seriousness of this constipation is illustrated by the severity of the various resulting lysosomal storage disorders, many of which are associated with debilitating neurodegenerative disorders. This well written and timely account of the lysosome is divided into three parts which deal with the delivery routes to the lysosome, its metabolic processes, and its role in the cellular environment. The life history of the lysosome and its dynamic relationship with the endosome and other vesicular components is discussed initially. A fully functional lysosome contains its own complement of enzymes and macromolecular substrates in aproaches to the clinical. The assessment by which proteins are taken up into the lysosome is a complex but ordered one which may or may not involve the mannose-6-phosphate receptor. In this process is crucial if we are ever to succeed in treating these disorders by enzyme replacement therapy. As well as the various uptake processes which are discussed, there are chapters on the lysosomal metabolism of proteins, glycosaminoglycans, lipids, and finally nucleic acids and phosphates. The biochemistry and molecular biology of many of the lysosomal storage disorders are well covered in the glycoconjugate and lipid chapters. It is perhaps unfortunate that the recently described cathepsin K, deficiency leading to cathepsin K deficiency (exhibited by Toulouse-Laurent) was too late to gain an entry. The final section deals with the important ATP dependent proton pump, responsible for acidification of the lysosome and endosome, the mechanisms of efflux and influx of solutes across the lysosomal membrane, and finally the role of the lysosome in pharmacology and toxicology is discussed. One of the editor’s main chapters on lysosomal membrane permeability, recalls the pioneering work of De Duve in the early 1950s on liver glucose-6-phosphatase activity (the enzyme responsible for type 1 glycogen storage disease). Interference from acid phosphatase led to a study of the latter enzyme and its location to an organelle whose membrane was impermeable to sucrose and which De Duve named the lysosome. This book achieves its aim of bringing us up to date with the lysosome and its biology. Each chapter is authoritatively written with many recent references. It will be of interest to all those involved in the study of this organelle and for those interested in the medical and genetic aspects of lysosomal disorders, and is recommended to complement the various clinical texts that are available.

J. E. Wraith


I am biased, but I do not believe any other medical specialty attracts as much media attention for its social and ethical implications as clinical genetics. The reason is real. It is the intrigue that we are when we discuss with families their plans for children and what their feelings are towards what we frequently term "normal" and "abnormal" references. It is the reality that we discuss with the physically disabled and people with learning difficulties? How much more complex does this become when people from two very different cultures meet in the genetic counselling clinic? Nancy Fisher has looked at 13 different ethnic cultures in the United States as they present to genetic counsellors.

Each chapter of this timely book is written in a similar format and describes the religion and main beliefs of that culture. Those factors that might influence their decision making are discussed: the family structure, who makes the decisions, who they may consult outside the family, and the influence that they have. Each chapter tries to give us an insight into whom we are meeting in the counselling room. There is a useful bullet point summary at the end of each chapter. The book’s layout and easy writing style makes it easy to dip into. Reading the book does not make you feel that the non-directive approach might not be the family’s approach, when they have to follow the advice of a knowing grandmother. This is clearly a North American book, devoted to an understanding of their ethnic minorities, with far less relevance to Britain and Europe.

The informativeness of chapters varies. The chapter on African-American culture is very helpful (avoid staring), looking at relationships and decision making within the family (be aware of the extended family support system). The chapter on Hispanic origins of the south west, is fascinating general reading. Chapter 1, describing people from a European culture, seeking control of time, decision making, their fertility, and ultimately future events that strike familiar chords. Contrast this with people from cultures that are less technocratic, who are more accepting of time and of events. They are orientated to present events rather than future goals, and success. The book raises the idea of our own competitive individualism, versus the non-competitive group culture of south east Asia. For those from a traditional Chinese culture, described as a group of recessive inheritance seem irrelevant as they present a calm stoic acceptance. Haven’t many of us had my experience of “I don’t understand about it being genetic, it’s just God’s will”, an example of how other cultures are formed? My experience is that other cultures are more adjusted to these situations than so-called westerners. I cannot comment on the accuracy of much of the book’s content. The two chapters on European and on Christian culture in North America convey a very narrow view of our society. They frequently refer to the stereotypical (if not diminishing) two parent families, living in stable relationships. There is scant reference to single parent families, family break ups, and divorces. What about multi-parental families and blended families with fluid parenting that we are now seeing. Chapter 2 on Latino culture is well written and is the standard I would have liked to see throughout the book. I can vouch that chapter 12 on Jewish culture is accurate and informative.

North Americans would certainly find this book relevant. There are chapters devoted to Korean, Latian, and Vietnamese cultures and for those of us from Europe European geneticists would be disappointed by the content. There are only 12 pages devoted to Indian cultures. I would encourage the Editor to produce a European version. More coverage on Muslim, Sikh, and Bengali cultures and migrant groups seen across Europe, for example, Turkish and North African workers, would broaden the book’s readership. I concluded that the world’s ethnic groups are something like genetic traits. There are groupings seen today that have evolved over time and share common ethnic threads. The book succeeds in making the reader aware of the ethnic groups that we might meet. It raises issues in cross cultural genetic counselling that we should be aware of and sensitive to. I would have looked more for discussion on decision making and the competing issues of the individual choice versus cultural background, particularly with second generation ethnic groups. What role for the individual person, the family elders, or their religious leader? If you see this book on your shelf, do pick it up and leaf through it. You will find the book fascinating.

Ian Ellis