

# PostScript

## BOOK REVIEWS

### Corneal dystrophies and degenerations: a molecular genetics approach

Edited by M X Wang. Oxford: Oxford University Press, 2003, £67.50. ISBN 0-19-516881-X (paperback).

The corneal dystrophies represent a large and varied group of inherited conditions, and the underlying molecular basis of many has been elucidated over the past decade or so. This exciting progress has been rapid and now allows a re-evaluation of our clinical and morphological classifications. This monograph has been produced in association with the American Academy of Ophthalmology, and is published by Oxford University Press. The major body of the text comprises six chapters and a total of 123 pages. In addition, there is a short self-study examination directed towards US CME accreditation. As would be expected from such a collaboration, the production qualities are high. Dr Wang, the editor, is a clinical academic in Nashville with major interests in the cornea, external eye disease, and refractive surgery. He lists the book's educational objectives: to bridge the gap between the new molecular information and the knowledge base for today's ophthalmologists; to discuss current understanding of the molecular pathogenesis of these conditions; to outline the use of excimer laser for the treatment of corneal diseases; and to review the most recent literature on corneal dystrophies and degenerations. This monograph, and its two first objectives in particular, are therefore timely in their conception.

The editor is co-author of five of the six chapters. The first chapter, written in collaboration with Dr Francis Munier, discusses the inheritance patterns of the corneal dystrophies, and in particular covers the range of epithelial and stromal dystrophies caused by defects in *TGFBI/BIGH3*, including a detailed and well constructed examination of their molecular pathology. The following three chapters describe, respectively, the epithelial, stromal, and endothelial dystrophies. In general the clinical, histopathological, and ultrastructural features of the disorders are clearly described, illustrated, and referenced. Here, the molecular focus is generally on gene identification and, whereas for certain disorders—for example, Meesmann epithelial dystrophy—there is a clear description of the underlying molecular mechanisms, this is disappointingly covered for many conditions. The final two chapters, concerning corneal and conjunctival degenerations and excimer laser therapies for corneal dystrophies, are likely to be of limited interest to the geneticist and carry little molecular information.

The key difficulty, when producing a monograph such as this, is ensuring that what is produced is as recent as possible and is not simply a replication of information that is available in other ophthalmological texts. In the first regard, the book unfortunately

appears to have taken a disappointingly long time from completion to publication. However, the degree of illustration—all figures are included on the excellent CD ROM that accompanies the book—when allied to the molecular details ensures that the phenotypic descriptions of the dystrophies are covered in a manner that will be both familiar and useful for ophthalmic clinicians and trainees alike.

Conflicts of interest: none declared

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### Nucleotide and protein expansions and human disease

Edited by J Gecz, G R Sutherland. Basel: Karger publishers, Reprint of Cytogenetic and Genome Research Vol 100 Nos 1–4, 2003, £67.50, pp 298. ISBN 3-8055-7621-8 (hardback)

Since 1991, when the CAG repeat expansion causing spinobulbar muscular dystrophy and the CCG repeat expansion in fragile X syndrome were discovered, there has been great progress in understanding the biology of triplet repeat instability and the diseases associated with these types of mutation. The number of diseases and classes of mutations has grown such that there are currently nine CAG repeat diseases where the repeats are translated into polyglutamine tracts, a recessive triplet mutation (Friedreich's ataxia), more than a handful of different diseases caused by expanded polyalanine tracts (for example, oculopharyngeal muscular dystrophy), diseases associated with untranslated triplet repeats (for example, myotonic dystrophy), and diseases caused by expansion of other micro or minisatellites (for example, progressive myoclonus epilepsy).

Soon after the first group of triplet repeat mutations was discovered, the biological mechanism was revealed behind the previously baffling (and controversial) phenomenon of anticipation, where the disease tends to increase in severity or present at an earlier age in successive generations in families. In many cases there has been rapid progress in developing cell and animal models of disease, and in some cases we have a much better understanding of pathogenesis. Yet many mysteries and controversies remain, even for diseases where the mutation was identified a decade or so ago (for example, myotonic dystrophy and Huntington's disease) and cures and treatments for the human diseases are still elusive.

This book addresses certain aspects of the field with a collection of freestanding articles covering aspects ranging from trinucleotide repeat instability, through epidemiology of spinocerebellar ataxias, to pathogenetic mechanisms. The nature of the book means that there are sometimes issues that are duplicated between chapters. This is not a problem, as the book is probably not designed to be read from cover to cover, but rather as a reference source. The topic coverage is not complete and gaps include discussions of the pathogenesis of Friedreich's ataxia and spinocerebellar ataxia type 1 (both where considerable progress has been made). However,

in contrast to many other books where there are collections of chapters, for instance focused on neurological diseases, this book deals with some of the more esoteric areas very well. There are fine chapters on repeat instability (by Lenzmeier and Freudenreich; and by Cleary and Pearson), oculopharyngeal muscular dystrophy (Brajs) and on transgenic models of myotonic dystrophy (Wansink and Wieringa), spinobulbar muscular dystrophy (Sobue and colleagues), Huntington's disease (Hickey and Chesselet) and Fragile X syndrome (Bakker and Oostra). It was good to see a chapter devoted to the interesting and possibly under recognised cerebellar ataxia syndrome associated with FRAXA premutation carriers (Hagerman et al) and the excellent chapter on SCA3 by Kobayashi and Kakizuka (a group that have made a number of key contributions to the polyglutamine disease field). In general, the chapters are written by authorities in their fields (including Brice, Ashizawa, Margolis, La Spada, Nelson, Usdin, and Ranum).

In conclusion, this book includes many chapters that add to and complement existing texts dealing with these diseases. The articles are generally of a high standard and are concisely written. This book would be of particular value to human geneticists, genetic counsellors, and researchers working on this class of diseases.

D C Rubinsztein

## CORRECTIONS

doi: 10.1136/jmg.2002.005546corr1

The acknowledgements for the original article by Bentley et al (*J Med Genet* 2003;40:249–56) were omitted and should read:

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We thank Dr Katarina Hannula and Prof Juha Kere for preliminary allelic expression analysis of CPA genes in lymphoblastoid cell lines. We also thank the Dunhill Medical Trust, the Wellcome Trust (LB, ZB, FK), the MRC (DM, JP, CB), and the Canadian Institutes of Health Research (CIHR) for supporting this research. K Nakabayashi is a Research Fellow of the CIHR and S W Scherer is a Scientist of the CIHR and the International Scholar of the Howard Hughes Medical Institute. All animal studies were carried out under the guidance issued by the Medical Research Council (MRC) in The Use of Animals for Medical Research (July 1993) and Home Office Project Licence No 30/1517.

We apologise for this error.

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Ressiniotis T, Griffiths P G, Birch M, et al. Primary open angle glaucoma is strongly associated with a specific *p53* gene haplotype (*J Med Genet* 2004;41:296–8). An erratum has been detected in the second paragraph of the Key points box. In the penultimate sentence "arginine residue" and "proline residue" should have been transposed. The authors apologise for this error.