

## BOOK REVIEWS

If you wish to order or require further information regarding the titles reviewed here, please write to or telephone the BMJ Bookshop, PO Box 295, London WC1H 9JR. Tel 020 7383 6244. Fax 020 7383 6662. Payment can be made by cheque in sterling drawn on a UK bank or by credit card (Mastercard, Visa, or American Express) stating card number, expiry date, and full name. (The price and availability are occasionally subject to revision by the Publishers.)

### The Principles of Clinical Cytogenetics.

Editors S L Gersen, M B Keagle. (\$79.50). Totowa, New Jersey: Humana Press. 1999. ISBN 0-89603-553-0.

The recent considerable expansion in our understanding of human chromosome pathology in medicine has been reflected in the production of some excellent textbooks from the "how to" of the encyclopaedic AGT Cytogenetics Manual to the more discursive but invaluable second edition of "Gardner and Sutherland". This multi-authored book, entitled, *The Principles of Clinical Cytogenetics*, aims to provide a comprehensive description of the basic concepts of clinical cytogenetics in a single volume.

This book is divided into four sections: "Basic Concepts and Background", "Examining and Analysing Chromosomes", "Clinical Cytogenetics", and "Beyond Chromosomes". The first chapter, "Basic Concepts and Background", gets off to a good start with an entertaining history of clinical cytogenetics. This includes an account of Painter's understandable indecision in the 1920s as to whether to plump for 46 or 48 as the diploid chromosome number for man and a reminder that terms like "super-female" were common currency in papers of the late 1950s. "Examining and Analysing Chromosomes" contains chapters on "Basic Laboratory Procedures", "Quality Control and Assurance", and "Automation" (reflecting one of the editor's own fields of interest). Disappointingly, the issue of laboratory safety and its management is dealt with in a single paragraph. Mention is made of approaches to Quality Assurance/Quality Control (QA/QC) outside the United States but only a brief overview is provided. Examination of different international approaches to QA/QC in more detail would have been useful.

"Clinical Cytogenetics" is the strongest section in the book with comprehensive, well referenced chapters on constitutional, prenatal, and cancer clinical cytogenetics. The reviewer was disappointed that there was little or no discussion of the role of cytogenetic analysis in the diagnosis of "Breakage" syndromes. Partly because of the rarity of these disorders, this topic would benefit from a review of the different procedures (often

involving extremely time consuming methods) used by laboratories. "Beyond Chromosomes" includes chapters on "Fragile X", "Imprinting", "Genetic Counselling", and on "FISH". The latter chapter is let down by some cramped, poor quality black and white figures illustrating techniques and the near universal reliance on a commercial company for illustrations for the text.

Whether this volume realises the editors' ambition to transcend the role of a reference work or a "how to" manual is open to question. There is, for instance, little reflection or discussion as to how laboratories should regulate or define the scope of their workload (for example, with respect to fragile X testing) in consultation with clinicians. This book is, however, reasonably priced and does provide an overview of many of the topics which form part of the working knowledge base of clinical cytogeneticists.

JONATHAN J WATERS

### Eye and Face in Syndromes - The Clinical Examination of Eyes and their Surroundings. Video. Mette Warburg, Ringsbjergvej 29, DK-4682 Tureby, Denmark.

This video has been produced as a training aid for ophthalmologists, ophthalmic nurses, orthoptists, genetic counsellors, and nurses. The commentary and content of the video are the work of Mette Warburg, a respected Danish geneticist who has published extensively in the field of ophthalmic genetics. The aim is to outline a system for examination of the face and eyes of patients where a syndromic diagnosis is a possibility. She explains that the video does not describe the embryology or aetiology of the conditions nor the intraocular signs unless they form an important part of the syndrome.

The introduction details the frequency of congenital eye anomalies in dysmorphic syndromes before going through an approach to examination, starting with observation of the child's eye contact and use of vision. She then goes on to discuss examination of the face followed by the ears, neck, and hands. Possible findings are illustrated in turn, using both well recognised and rare conditions. The length of the video (40 minutes) means that only brief details can be given about each case and there are a few technical inaccuracies in the commentary which may have resulted during translation. The video has been shot as a series of still pictures and although this results in the loss of some information, especially when discussing observation of children's behaviour, the unusual nature of some of the conditions indicates that they have been collected over a period of time, making it difficult to produce the video in any other way.

From a training point of view, the video illustrates an interesting set of dysmorphology cases linked to a system for examination. Most of the cases will be familiar to doctors training in clinical genetics and so the video will probably be most useful to ophthalmolo-

gists interested in dysmorphology, who wish to familiarise themselves with some syndromic diagnoses and signs to look for in a general examination.

NORA SHANNON

## NOTICES

### British Human Genetics Conference

This conference will be held at the University of York on 11-13 September 2000. It will include a one day joint symposium "Technologies in Genome Analysis" with the Genetical Society on 13 September 2000. For further information contact the Conference Office, British Society for Human Genetics, Clinical Genetics Unit, Birmingham Women's Hospital, Edgbaston, Birmingham B15 2TG, UK. Tel/fax: 0121 627 2634. Email: bshg@bham.ac.uk Website: <http://www.bham.ac.uk/bshg>

### 7th European Meeting on Psychosocial Aspects of Genetics (EMPAG 2000)

This conference will be held in Manchester on 21-23 September 2000. Further information is available on our web site at [www.cmht.nwest.nhs.uk/directorate/deptstmary/empag2000.htm](http://www.cmht.nwest.nhs.uk/directorate/deptstmary/empag2000.htm) or from the Conference Secretary, Barbara Egan, Department of Clinical Genetics, St Mary's Hospital, Hathersage Road, Manchester M13 0JH, UK.

## CORRECTIONS

In the paper by Chotai and Payne (*J Med Genet* 1998;35:472-475) on "A rapid, PCR based test for differential molecular diagnosis of Prader-Willi and Angelman syndromes", the sequence of primer S2 (antisense) was wrong in two places. The correct sequence is as follows with the corrected letters in bold: 5'-CCCCTCCTCTACACAGCAATCAT-3'. The authors apologise to any readers who encountered difficulties in trying to amplify the above sequence.

In the letter by Webster *et al* (*J Med Genet* 2000;37:62-63) on "Risk of multisystem disease in isolated ocular angioma (haemangioblastoma)", there was an error in the second equation where two " | " signs that denote conditional probability were omitted. The equation should have been as follows:

$$\frac{\Pi[\Pr(C_i | VHL)] \times \Pr(VHL)}{\Pi[\Pr(C_i | VHL)] \times \Pr(VHL) + \Pi[\Pr(C_i | \overline{VHL})] \times \Pr(\overline{VHL})}$$