

# PostScript

## BOOK REVIEWS



### Dysmorphic Syndromes and Constitutional Diseases of the Skeleton

G Canepa, P Maroteaux, V Pietrogrande. Padova: Piccin Nuova Libreria, 2001.

This is a two volume book describing the clinical and radiological features of 400 bone dysplasias, dysostoses, dysmorphic syndromes, and selected non-mendelian disorders. Unlike previous similar works, it is written by two orthopaedic surgeons and a clinical geneticist, and comes with a CD-ROM that has a specially written program, which can be used as a gamut index of clinical and radiological signs and for making diagnoses based on these features.

Volume 1 contains a classification of bone dysplasias and dysmorphic syndromes involving the skeleton. The classification of osteodysplasias is mainly clinical and, although different from the 1997 International Nomenclature of Constitutional Disorders of Bone, is still quite useful diagnostically. The conditions are discussed in alphabetical order and volume 1 contains diseases from A to M. The format used to describe each syndrome and dysplasia includes major diagnostic criteria, aetiology-pathogenesis, incidence, risk of recurrence, sex ratio, age at detectability, main clinical features, radiological findings, prognosis, diagnosis (including differential diagnosis, prenatal diagnosis, and diagnosis of carriers), and therapy. Most of the conditions have been well presented. There are excellent photographs, radiographs at different ages, and line diagrams of the salient facial, clinical, and/or radiological features for almost all the conditions discussed in the book.

Volume 2 contains diseases from N to Z. This volume also includes an atlas of the histopathological features of common skeletal dysplasias by Victor and Ritta Stanesco. The authors of this section have put together an excellent collection of pathological sections (including electron micrographs) of the growth cartilage in 49 different skeletal dysplasias and syndromes. This is a very useful feature of the book but it would benefit from the inclusion of sections of normal growth cartilage for comparison. This volume of the book also includes a gamut index of clinical and radiological features, a glossary of terms used in the book for those not familiar with dysmorphology and genetic terminology, and an index.

The most useful feature of the book is the CD-ROM that accompanies it. This contains a multimedia computer program called "Philos" that can be used to search for conditions

with any combination of clinical or radiological features from the 400 conditions discussed in the book. It can also be used to find the clinical and radiological features of any of the syndromes and dysplasias discussed in the book. The software is easy to install and use and it can be used to print the list of differential diagnoses obtained by different searches. It is also an excellent teaching and learning tool.

Although the book is well written and well illustrated, there are numerous typographical and grammatical errors in the text. From a clinical geneticist's viewpoint, the book is disappointing, as the genetics of the conditions is not discussed in any detail. The inheritance pattern(s) for each condition is described and the gene loci for most conditions mentioned, where known. However, the book fails to discuss the dramatic advances that have been made in recent years in understanding the molecular genetics of several dysmorphic syndromes and skeletal dysplasias. The book contains no information about the genes that have been identified for several of the syndromes and skeletal dysplasias. In addition, several well recognised conditions, such as Langer mesomelic dysplasia, Nievergelt syndrome, osteopathia striata with cranial sclerosis, sponastrime dysplasia, and Stüve-Wiedemann syndrome have not been discussed. Many distinct lethal skeletal dysplasias such as Astley-Kendall syndrome, Blomstrand dysplasia, Greenberg dysplasia, Pacman syndrome, and Raine syndrome have also not been described. Some of the terminology used in the text is obscure and some conditions are discussed under unfamiliar names (for example, nail-patella syndrome is discussed under the title of arthro-osteonychodysplasia). There are very few recent references, and there are several references from Italian publications. It would also have been helpful to have an index in both volumes of the book for ease of reference.

Despite these shortcomings, this book is an extremely useful addition to the field of clinical dysmorphology and skeletal dysplasia. It will find a place as a reference book in clinical genetic, orthopaedic, perinatal pathology, and radiology departments and in the personal libraries of clinical geneticists, orthopaedic surgeons, and radiologists involved in the diagnosis and management of dysmorphic syndromes and skeletal dysplasias.

Mohnish Suri

### Colour Atlas of Genetics

2nd edition. Eberhard Passarge. (Pp 457.) Stuttgart: Thieme. 2001. ISBN 3-13-100362-6. New York: Thieme. 2001. ISBN 0-86577-958-9.

The second edition of this pocket sized book is successful in its aim to cover a wide range of genetics topics in a concise yet clear and well illustrated way. The book is written primarily for students of medical sciences, but also fulfils a handy role for physicians with an interest in genetics, or indeed those involved with clinical genetics, as a rapid and comprehensible reference for concepts, methods, and terminology with which the reader may be

unfamiliar. In addition, it provides some very useful ideas for approaching and illustrating undergraduate teaching in genetics.

First published in 1994, this edition has been updated and expanded, as is necessary in such an expanding field, with new sections on genomics, and many new colour plates covering areas such as cancer predisposing genes, ion channel diseases, and regulation and expression of genes. The fundamental format used in the first edition is retained, with the book divided into three sections, fundamentals, genomics, and genetics and medicine. The book includes useful references relating to each topic, in addition to relevant helpful website addresses. Disease mechanisms are well explained, although the clinical aspects are kept to a minimum.

The initial impression may be one of an over ambitious attempt to be all inclusive for significant value, with very busy pages, but further reading shows the true value of the atlas, with succinct text and informative and clear illustrations, explaining current understanding in this field.

M Lees

### Adrenal Disease in Childhood. Clinical and Molecular Aspects

Editors Ieuan A Hughes, Adrian J L Clark. Basle: Karger. 2001. ISBN 1421/7082.

A 10 year old boy has had type 1 diabetes for two years, with reasonable control. Over a few weeks he develops hypoglycaemic episodes, worse in the mornings, with vomiting. His insulin requirements are reduced, with no improvement. It turns out that his mother has autoimmune thyroid disease with premature ovarian failure and Addison disease. On short synacthen testing the boy has minimal cortisol responses.

This is a scenario straight out of a BMJ "Lesson of the Week", and reflects the pitfalls of ignoring adrenal disease in children. Indeed, on looking back at Douglas Hubble's Paediatric Endocrinology reference text of 1969, disorders of the adrenals were basically limited to congenital adrenal hyperplasia, Cushing syndrome, and Addison disease. The physiology of salt and water balance had been worked out, and steroid metabolism had established itself as a recognised branch of biochemistry. However, there was little understanding of the underlying mechanisms of disease.

Tremendous progress has occurred over the last 10 years in molecular medicine, and Hughes' and Clark's book reflects this admirably. Under their editorship, they have invited contributors who are actively engaged in research to present the most up to date information on the genetics, protein chemistry, and biochemistry of adrenal disorders in children. This ranges from the latest advances in steroidogenic factor-1 (SF-1) and DAX-1 expression in adrenal development to the clinical features of children with cyclical Cushing syndrome and primary pigmented adrenocortical disease (PPNAD).

Each chapter is laid out in a logical order, with clear figures and explanations of biochemical pathways. The review of Cushing syndrome and Addison disease places these

conditions in a logical classification, and concentrates particularly on the most up to date molecular genetics. There is an informative discussion of familial autoimmune disease which classifies the family described above as multiple endocrine abnormalities type 2 (MEA 2). An excellent chapter on X linked adrenoleucodystrophy has a detailed section on clinical and therapeutic approaches to the management of these children, together with information on prenatal diagnosis. There is a useful contribution on defects of aldosterone

biosynthesis, although it would have been helpful to refer in passing to the differential diagnosis of pseudohyperaldosteronism and the similarities in presentation. The chapter on 21-hydroxylase deficiency defects and their phenotype places the biochemical, molecular genetic, and differential diagnosis information together in an exemplary fashion. The discussion of prenatal mutation analysis and treatment brings this topic right up to date. This also introduces one of the current controversies in paediatric endo-

crinology, the safety of early glucocorticoid administration to pregnant mothers to reduce the degree of virilisation in affected female fetuses.

This book will be an essential reference for scientists and clinicians who want the latest molecular genetic information on adrenal disorders, with enough clinical and biochemical data to put it into context. This is a useful contribution in a rapidly advancing field.

**T G Barrett**



Want to extend your search?

### Cross journal searching

If you can't find what you are looking for in the *Journal of Medical Genetics* you can extend your search across many of the more than 200 journals available for selection. You can restrict your search to specific subject areas (eg, clinical medicine, basic research), or select specific journals, or search all available titles.

**[www.jmedgenet.com](http://www.jmedgenet.com)**