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# LETTER TO THE EDITOR

## Marfan syndrome

In the May issue of the Journal, Gray and Davies[1] present a nice short overview of the clinical features, natural history, and molecular genetic aspects of Marfan syndrome. For the clinical diagnosis of Marfan syndrome, they refer to the so-called "Berlin Nosology",[2] a set of criteria established in 1986 by a group of experts at the ISHG Meeting in Berlin. With these criteria Marfan syndrome is diagnosed in the absence of an unequivocally affected first degree relative if involvement of the skeleton and at least two other systems is found, with at least one of those systems showing a major manifestation. In the presence of one or more unequivocally affected first degree relatives, involvement of only two organ systems and preferably, although not necessarily, a major manifestation of the disorder is required.

In a reply in the December issue of the Journal, Galasko[3] rightly points to the danger of mistakenly diagnosing Marfan syndrome in unaffected relatives of Marfan patients if the Berlin criteria are strictly applied. It was widely realised that, in particular with the advent of molecular testing, weaknesses in the Berlin Nosology existed. Therefore, recently, a revised version of the diagnostic criteria for Marfan syndrome has been put forward.[4] These new criteria (which may be called the "Ghent Nosology", in analogy to the "Berlin Nosology") are still based on a combination of major and minor clinical manifestations in different organ systems. The major differences from the Berlin Nosology are: (1) skeletal involvement as a major criterion if at least four of eight typical skeletal manifestations are present; (2) more stringent requirements for diagnosis in relatives of an unequivocally affected subject (a major criterion in the family history and one major criterion in an organ system and involvement of a second organ system, the major criteria being dilatation/dissection of the ascending aorta, ectopia lentis, dural ectasia, and the skeleton); (3) potential contribution of molecular analysis to the diagnosis (presence of a causal FBN1 mutation or disease associated FBN1 haplotype).

In addition, initial criteria for diagnosis of the conditions partially overlapping Marfan syndrome are presented. It is hoped that these new and more stringent criteria can serve as an international standard for clinicians who are confronted with the problems of diagnosing this variable and pleiotropic syndrome.

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1 Gray JR, Davies SJ. Marfan syndrome. *J Med Genet* 1996;33:403-8.
2 Galasko G. Marfan syndrome. *J Med Genet* 1996;33:1051-4.
3 Beighton P, De Paepe A, Danks D, *et al.* International Nosology of Heritable Disorders of Connective Tissue, Berlin, 1986. *Am J Med Genet* 1988;27:139-40.
4 De Paepe A, Devereux RB, Dietz HC, Hennekam RCM, Pyeritz RE. Revised diagnostic criteria for the Marfan syndrome. *Am J Med Genet* 1996;62:417-26.

# BOOK REVIEWS

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**Emery and Rimoin's Principles and Practice of Medical Genetics.** 3rd edition. Editors D L Rimoin, J M Connor, R E Pyeritz. Senior advisory editor A E H Emery. (£295.00, 2 volumes.) Edinburgh: Churchill Livingstone. 1996. ISBN 0-443-04851-7.

The title of this work is correct, this is a state of the art study of the principles and practice of medical genetics. However, the title underplays the breadth, quality, and usefulness of the book that the three distinguished editors, David Rimoin, John Connor, and Reed Pyeritz, have produced.

It is seven years since this work was last published and now it is in its third edition. While it is again published in two volumes the amount of new information included has led to an increase in both its size and usefulness. Despite this, the style and layout of the book is clear, making it easy to read, scan, or to find information about a particular question. The reference lists at the end of each chapter and the index printed in both volumes are comprehensive. The editors have written chapters and assembled contributions from 234 authors. The editors have clearly kept the focus of the book on clinical relevance and also enforced a clear style, kept repetition to a minimum, and avoided any major deficiencies.

The book is arranged by clinical approach, genetic methodology, and body system and not by specific diseases. There are 142 chapters arranged in three sections: Basic Principles, Clinical Applications, and Future Considerations. The Clinical Applications section is subdivided into "General Principles", from clinical genetic methodologies to ethical and legal issues, and "Approaches to Clinical Problems", containing seven particularly useful chapters, and 19 system specific sections each usually with multiple chapters covering specific topics. Some of the chapters are relatively short reflecting a tight focus, but all are written by experts in their field. There are few books with such an impressive list of contributors.

If you have long, lonely winter nights you can try and find faults in this work. You will have to be patient, for while there are a few, for example, Cockayne syndrome does not lead to a strong cancer predisposition as stated on page 431, they are infrequent.

In many ways this book is a testament to medical genetics as it has evolved from a mathematically based science to a broadly applicable and useful part of established medical care. This clinically useful, comprehensive, clearly written, authoritative book is an essential work for any department involved in clinical practice or training in medical genetics.

C G WOODS

**London Dysmorphology Database and London Neurogenetics Database with Photo Library.** Windows versions. R M Winter, M Baraitser. (£395.00 for each of three components, £750.00 for any two, £1100 for all three, +VAT.) Oxford: OUP, Oxford Medical Databases. 1996.

All the medical specialities have their own touchstones, to which they turn to gain reassurance and a sense of belonging. Cardiologists have angiography and balloon technology, orthopaedic surgeons have rows of gleaming metalwork, and psychiatrists have comfortable leather chairs. In clinical genetics, one turns again and again to the excellent London Dysmorphology and Neurogenetics databases by Robin Winter and Michael Baraitser.

The databases have always described the known and less well known genetic syndromes in succinct and perceptive summaries, along with a list of the features of the syndromes, and the important references. Individual case reports, which are thought to represent distinct clinical entities, are also included. They are an unparalleled resource to turn to for a quick resumé of the many conditions that may not be in the forefront of our memory. The search facility lets us check our differential diagnoses using a cluster of dysmorphic or malformation handles, to make sure we haven't overlooked any diagnosis. The authors quite correctly make the point that the databases are not meant to substitute for the diagnostic process, simply to aid it. However, it is often useful to have the databases on a laptop at an outreach clinic. One can miraculously produce knowledge about a rare condition, with a discreet consultation with your trusty laptop!

The previous edition introduced on CD-ROM photographs of many of the conditions, to try and answer the often difficult task of translating written summaries of facial dysmorphology into a characteristic picture. The number of photographs was limited, access to photographs could be slow, and installation could be somewhat tricky (speaking from personal difficulty, solved eventually by several phone calls to Oxford University Press). Until now the databases have been supplied as a DOS based system, with no concessions to Windows point and clickers, or to the die-hard band of Macintosh users.

The latest Windows version of the databases is a great improvement, and is more than a simple DOS to Windows conversion. For the technical minded, the system ran extremely well under Windows 95 on a Pentium 133 MHz processor, with 16 MB RAM, a 1.6 GB hard disk, an 8x CD-ROM drive, and a super VGA screen. This is a considerably higher specification than the requirements suggested by the system manual. In