European Journal of Human Genetics (2005) 13, 1312

www.nature.com/ejhg

BOOK REVIEW

Myotonic dystrophy - time to improve patient care and prepare for pathogenesis based treatments

Myotonic dystrophy: present management, future therapy

Edited by PS Harper *et al*, Oxford University Press.

Susan M Huson

European Journal of Human Genetics (2005) **13,** 1312.
doi:10.1038/sj.ejhg.5201500

Myotonic dystrophy is one of the many genetic diseases viewed as a medical curiosity. It is a diagnosis medical students and physicians in training 'swot up' for exams, but then rapidly forget about! The problem for patients with Myotonic dystrophy is that few specialists recognise the multisystem nature of their disease, even though it is the commonest from of muscular dystrophy. Fortunately, this is now changing and this book is witness to developments in understanding of pathogenesis and disease management in recent years.

The idea for the book arose at an international workshop held by the European Neuromuscular centre in 2001. The 18 chapters are all written by international experts in the field and include two chapters written from the patient's perspective, which I particularly appreciated. The other chapters cover clinical and counselling issues and currant understanding of disease pathogenesis. The book ends with a review of recent therapeutic trials and speculates on likely future developments.

Reading the book through, I was slightly frustrated by the repetition of core facts at the start of each chapter. The advantage of this is that the book can be 'dipped' into by different specialists wanting an update about a particular aspect of the disease. All the authors cover their topic in a clear and concise manner. Some very useful summary tables and flow diagrams of management support the text. The relevant cellular pathways are also well illustrated.

Who should read this book? As a reference work there is something for everyone and as such every genetic and neurology department library should have a copy. It is a must for all those working in the field either clinically or in research. Personally, I would still recommend Harper's monograph on Myotonic dystrophy to those entering the field, but as the third edition was published in 2001 this book provides a useful update.

Those involved in teaching and training in neurology and genetics will find this book an invaluable source of teaching material. For example, the chapter looking at missed diagnosis in Myotonic dystrophy provides useful histories and data for those teaching about multisystem disease or variability in dominant disorders. Too few genetic texts contain chapters written by people personally affected. The chapter written by Sharon Miller Lord describes her personal journey through diagnosis and the benefit of contact with a support group. This would be a useful 'core text' for genetic counselling students and clinical genetic trainees alike. (and perhaps also as continuing professional development for established clinical geneticists!) ■

Susan M Huson is a Hon Clinical Geneticist at Regional genetics Service, St Mary's Hospital, Manchester, UK.
E-mail: smhuson@aol.com