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BOOK REVIEWS

Authors muscle in on history

'The History of a Genetic Disease Duchenne Muscular Dystrophy or Meryon's disease',
Edited by AEH Emery and MLH Emery
Published by Oxford University Press
ISBN: 978-0-19-959147-3 price: £80.00, $155.00

Usha Kini and Deirdre Cilliers

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Duchenne muscular dystrophy (DMD) has been regarded as one of the archetypal genetic conditions as far as the defined phenotype and identification of the gene is concerned. This book, a second edition, tells the story of how the knowledge of DMD unfolded. It then describes the realisation that there were other types of muscular dystrophy with their own particular clinical picture and genetic basis.

The book begins with a short overview. The authors then examine each aspect of DMD in detail, starting with the early descriptions of cases and progressing to the refining of the clinical phenotype, together with the pathological and biochemical basis of the condition. The history of the recognition of the inheritance pattern with the implications for family members is analysed, leading to the discovery of the gene and the function of its protein.

The most absorbing aspect of the book is the comprehensive and insightful description of the people that contributed to the investigation of DMD. In addition to the effort and research that the authors have put into finding out about the lives of the discoverers, the authors have known several of the people discussed in these pages, and provide a personal appreciation of these clinicians and scientists.

An intriguing feature of the book is the description of the prevailing scientific and cultural climate of the time in which the discoveries were made. This places the people, their new theories and the implications of their discoveries into a context, which provides the reader with a far greater insight than if the history of the discoveries alone was told.

The authors particularly wish to highlight one contributor to DMD, namely Edward Meryon. They suggest that he has not received the recognition that his counterpart, Duchenne de Boulogne, has done. They describe how Meryon was the first to link the clinical description, the muscular basis for the condition and the inheritance pattern.

In the last few chapters of the book there is a change of focus. These chapters concentrate on the scientific aspects of the DMD gene and protein and the history of genetic counselling of this condition. Future researchers are invited to address several unanswered questions about Duchenne and the other muscular dystrophies in the final chapter.

The book tells the extraordinary story of DMD well. It is a book that can be read with pleasure from start to finish, although some people may be slightly distracted by the many italicised words. This book would be enjoyed by anyone with a basic genetic knowledge and particularly those who are involved with the care of patients with muscular dystrophy. It could also be read with benefit by those interested in medical history and the investigation of disease.

Perhaps a future edition may have an additional chapter in which the story of the discovery of an effective treatment for the condition is told■

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Deeper than the skin

A review of Genetic Skin Disorders by Virginia P Sybert, Second Edition

'Genetic Skin Disorders', Second Edition, 2012, Virginia P Sybert
ISBN: 978-0-19-539766-6
Published by: Oxford University Press
Price £110
USD 164

Dr Vadakke Kanakath Ajith Kumar

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The second edition of 'Genetic Skin Disorders' was long awaited. The book is well-illustrated with colour photographs and provides easily accessible information on inherited skin disorders. It has a very useful section on differential diagnosis by skin signs. It does not aim to be a comprehensive text book of Dermatology, but an aid to the clinician in establishing the diagnosis, inheritance pattern and syndromic associations of inherited skin disorders. It succeeds admirably in the task it sets out to accomplish.

Skin manifestations occur in many genetic disorders, which may involve other organ systems. In some ways, the title of the book is as understated as Gorlin's 'Syndromes of the Head and Neck'. The annotated bibliography is very helpful and Dr Sybert's comments make interesting reading.

The book is available only in hardback. It would be good to have the book in electronic format, which would make it more portable and easier to search.

'Genetic Skin disorders' is reasonably priced for a text book of this kind. It is recommended for Clinical Geneticists, Dermatologists, Paediatricians and Physicians in Internal Medicine■

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