Book reviews


Genetic Biochemical Disorders

The stated purpose of this book is to summarise the salient features of inborn errors for a wide readership. After a short introduction there are chapters on lysosomal storage disorders, inborn errors of amino acids, organic acids, purines and pyrimidines, carbohydrates, erythrocyte enzymes, porphyrins, steroids, lipoproteins, thyroid hormones, and copper metabolism. This list serves to emphasise the enormity of the task facing anyone setting out to write a book, even a short one, about inborn errors. A very large number of inborn errors is included and the text is clear, but the discussion about each inborn error does reflect the authors’ particular interests. Details of the molecular defects and prenatal diagnosis tend to outweigh the discussion about the clinical presentation, diagnosis, and treatment. All of these are often brief, particularly those concerning treatment. There is no discussion about treatment at all in the section on the porphyrias, and elsewhere important details about therapy are not always covered clearly.

An important feature of the whole of the book is the emphasis on genetic counselling, but the rapidity of change makes it almost impossible to publish a book that is fully up to date, both because of the introduction of new methods of prenatal diagnosis and because of the identification of new inborn errors or the discovery of the biochemical basis of other ones. Thus, in the introduction, the authors discuss chorionic villus biopsy and DNA probes but these are difficult to find in the main body of the text. Some disorders are missing, such as Zellweger’s syndrome and all the associated peroxisomal disorders including Conradi’s syndrome. The references are also rather outdated, although the total number is huge, approximately 3700. More than 50% of the 300 I counted predated 1975 and fewer than 5% were published in 1981 or after. All the references are listed together at the end of the book making it tedious to find individual ones quickly.

This book provides a useful and concise introduction for those with limited experience of the field of inborn errors. However, it does not give much precise detail about treatment and it will always be necessary to check that there have not been recent advances.

Peter Goodfellow

The Y Chromosome. Part A: Basic Characteristics of the Y Chromosome

For around £100 you can buy 100 000 units of the restriction enzyme EcoRI, fly to America from Britain (or vice versa), or buy a copy of ‘The Y Chromosome: part A’. If you chose the latter alternative you will have purchased 23 reviews and almost 600 pages describing Y chromosome cyto- genetics and molecular genetics.

The Y chromosome can be roughly divided into euchromatin and heterochromatin. The euchromatin constitutes about 40% of the human Y and contains the sex determining gene (or genes). The heterochromatin is composed of two repetitive sequences. It was a puzzle to discover that it was possible to write 600 pages about the human Y chromosome (and another 600 pages in part B?). Perhaps this is a direct reflection of the structure of the Y chromosome; the chapters emphasising the cyto- genetics of the heterochromatin are highly repetitive. Even the chapters which discuss the molecular genetics of the euchromatin and genetics of the H-Y antigen show evidence of low copy number repeats. If I were editor, I would have reduced the number of chapters by half while being careful to retain several excellent reviews, such as those by Daiger and Chakraborty (Mapping the Human Y Chromosome) and Willard (Molecular Organisation of Repeated DNA Sequences on the Human Y Chromosome). However, the book is very detailed in its coverage and the repeats are probably the price to be paid for the exhaustive treatment.

Scientific views about H-Y antigen are often propounded with religious fervour. The monotheists believe that there is but one H-Y antigen which controls male sexual development. Polytheists recognise separate serological and T cell defined H-Y antigens and usually reserve judgement on function. Apart from the T cell defined antigen, my own views are atheistic. In accordance with most publications, only monotheist views are presented in this volume.

With a few tubes of restriction enzyme you can begin to study the molecular genetics of the Y chromosome. With a transatlantic flight you can learn about the experiments your colleagues are performing. With ‘The Y Chromosome: part A’ you can read about experiments done in the past. My inclination would be to buy the restriction enzyme.

J V Leonard

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