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BOOK REVIEWS

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Clinical and Genetic Aspects of the X Linked Hydrocephalus/MASA Spectrum. Editor C Schrander-Stumpel. (Pp 137.) Maastricht: Datawyse/Universitaire Pers. 1995.

This small volume is effectively a monograph recounting the progressive recognition that three disorders, X linked hydrocephalus, MASA syndrome, and X linked complicated spastic paraparesis linked to Xq28, in fact constitute a clinical spectrum. The variability in clinical features within families and the overlap between families led to the suggestion that the disorders may be allelic. The disorders all mapped to Xq28, and a search for mutations in the LICAM gene was fruitful.

Although the volume consists largely of a series of papers, there is a logic to the presentation that allows this book to rank as more than that.

I would recommend this as a fine example of a dissertation/monograph, sure to be of interest to all clinicians and scientists in genetics who recognise the value of a broad understanding of the disorders that we attempt to diagnose, manage, and study. It would also be of interest to our colleagues in paediatric neurology.

ANGUS CLARKE

From Genotype to Phenotype. Editors S E Humphries, S Malcolm. (Pp 310; £55.00.) Oxford: Bios Scientific Publishers Ltd. 1994. ISBN 1-872748-62-7.

This book is the second in the Human Molecular Genetics series, following on from the excellent "Human Mutation" volume, and is aimed at both clinical and scientific research geneticists. The first volume had a relatively easy task in explaining and illustrating the history of genetic mutations, while this second volume takes on the more difficult job of describing the present state of knowledge regarding how a genotype is related to a specific phenotype. This could just have been an exercise in presenting lists of disorders, their mutations and a description of the correlations with phenotype known to date. This would not be a bad thing but without suitable commentary to place the conclusions in context such a book would have been of little use. Thankfully, the editors have ensured that there are several chapters that describe the types of mutation involved in genotypes, notably the first chapter by Susan Malcolm that clearly and concisely describes most of the mutations involved in human genetic disease. Chapters on cystic fibrosis, Gaucher's disease, collagen disorders, familial hypercholesterolaemia, Charcot-Marie-Tooth disease, and Wilms' tumour make up the first half of the book and cover these areas admirably. Two chapters on myotonic dystrophy and fragile X disease clearly describe the new field of short tandem repeat length variation as a cause of genetic disease. The remaining mechanisms of mutation are then covered in the next chapter, including somatic mosaicism, chimerism, and X inactivation. A chapter on mitochondrial DNA associated disease is then followed by three chapters covering the emerging challenge in the field of human genetics, namely multigenic disorders. Three subjects are covered: diabetes, coronary artery disease, and dyslipidaemia. These chapters are necessarily weaker than the preceding ones and the chapter on diabetes does not discuss non-insulin dependent diabetes which is unfortunate given the recent work by Todd *et al* in describing techniques by which loci involved in a heterogeneous genetic disorder can be investigated. Besides these minor criticisms the book is clear and well laid out, with high quality black and white illustrations and each chapter has been concisely referenced. As Professor Kåre Berg says in his introduction, "It is a pleasure to read a book that describes these recent advances so well and which is so full of novelty". This is a well written introduction to the state of the art in one of the most rapidly advancing fields in medical research. It is to be recommended to both the clinician and scientist who are taking part in the fascinating search for the elusive links between genotype and phenotype.

ANDREW J WALLEY

Haldane's *Daedalus* Revisited. Editor K R Dronamraju. (Pp 147; £19.99.) Oxford: Oxford University Press. 1995.

Haldane's reason for choosing the title *Daedalus* for his slim volume of 93 pages on science in the future is not very clear. But Bertrand Russell's choice of *Icarus* for his counter attack, also published in 1924, is more obvious. Daedalus was a legendary Athenian craftsman of great skill and ingenuity. He made wings for himself and his son, Icarus, to enable them to escape the Labyrinth of Minos, but Icarus flew too near the sun and was destroyed. This mythological tale reflects Haldane's and Russell's different attitudes to science.

Haldane prophesied that science would play an increasing role in human affairs. He championed particularly the Soviet Communist attitude to science (though of course Lysenko had not yet appeared on the scene) and considered religion an irrelevance. Some of his predictions have proved correct but others were silly or wrong (windpower to replace completely more conventional energy sources and food from coal products, but he dismissed atomic power as ever having any commercial possibility). His idea of "ectogenesis", with the culture of human embryos in the laboratory, foreshadowed Aldous Huxley's *Brave New World* published in 1932.

Haldane, recognised nowadays mainly as a mathematical geneticist, belonged to a class of rebellious liberal intellectuals before the Second World War. They were often privileged and sometimes arrogant and intolerant. His book, which set out to shock the more conventional mind, was a great success at the time. I first read it as an undergraduate in the 1940s and found it very exciting but was less impressed by Russell's book. But now, on rereading these two essays, I more clearly appreciate the latter's concern. Technological developments have far outstripped even Haldane's fertile imagination and have generated many moral and ethical problems which are proving ever more difficult to resolve. It is impossible to stop scientific and technological advances. And as Russell says "Technical scientific knowledge does not make men sensible in their aims and administrators in the future will be presumably no less stupid and no less prejudiced than they are at present."

Dronamraju, a one time student of Haldane, has gathered together several eminent people to comment on *Daedalus* based on their own opinion of the work and in the light of subsequent developments. Unfortunately these contributions prove to be somewhat repetitious and even irrelevant: Perutz merely reflects briefly on Haldane in Cambridge. Ezrahi considers the work from a philosophical point of view. The other contributions are mainly concerned with scientific matters. I especially enjoyed Weatherall's contribution, perhaps because he writes as a physician-scientist. I would find a modern day *Daedalus* written by him especially rewarding, instructive, and thought provoking.

ALAN EMERY

Notice to contributors (general guidance)

The readership of *Journal of Medical Genetics* is world wide and covers a broad range of workers, including clinical geneticists, scientists in the different fields of medical genetics, clinicians in other specialities, and basic research workers in a variety of disciplines. It publishes original research on all areas of medical genetics, along with reviews, annotations, and editorials on important and topical subjects. It also acts as a forum for discussion, debate, and information exchange through its Letters to the Editor columns, conference reports, and notices. The editor is always grateful for suggestions or criticisms from readers and authors.

ORIGINAL PAPERS

These may be on any aspect of medical and human genetics and may involve clinical or laboratory based and theoretical genetic studies. Guidance on length can be obtained from studying the Journal. Case and family reports may be submitted as *Brief papers*. *Short reports* should in general not exceed 500 words, with one or two illustrations, and the text should be continuous with no headings. An abstract should be provided for all papers. Contributions may also be submitted as *Hypotheses* or *Technical notes*. Accelerated publication of papers of particular importance will be considered.

REVIEWS

Short or longer reviews on all aspects of medical genetics are welcome, but should be discussed first with the Reviews Editor. Contributions on historical topics, or which could form part of specific series, are particularly acceptable.

ANNOTATIONS AND EDITORIALS

These are written or commissioned by the editors, but suggestions are welcome regarding possible topics and authors.

LETTERS

These are welcome on any relevant topic and will be published rapidly. Those relating to or responding to previously published items in the Journal will be shown to those authors, where appropriate. Although a paper submitted as an original report may sometimes be published in shortened form as a letter, it is preferable for initial submissions to be as a short report, unless directly related to a previous journal article.

CONFERENCE REPORTS

Reports from small to medium sized meetings, especially international workshops on specific topics, will be appreciated. Authors intending to submit conference reports should liaise with the Reviews Editor to avoid duplication.

SPECIAL ISSUES AND SUPPLEMENTS

These are published at intervals on topics of particular relevance. Enquiries are welcome from those organising workshops or symposia who may have material suitable for such an issue.

BOOK REVIEWS

The Journal aims to review as wide a range of relevant books as possible. Authors or others wishing to check if a book has been received may check with the Journal office. Computer programs and databases, official reports, and other material relevant to the field may all be appropriate for review. Enquiries about such items are welcome.

OBITUARIES

The Journal would like to be informed rapidly of the death of any senior or important person in the field of medical or human genetics, regardless of geographical location. In general, a brief notice would be published rapidly, with a longer obituary as appropriate. Since such deaths often occur many years after retirement, it will be appreciated if readers will contact the Reviews Editor so that appropriate arrangements can be made.

NOTICES

Notice of forthcoming meetings in different countries should be sent as far ahead as possible. Extensive descriptions should be placed as advertisements.

'CALLS FOR PATIENTS'

The Journal receives an increasing number of requests to publish notices of proposed studies involving patients or families with rare genetic disorders. In general such notices are appropriate only for major international collaborations; the proposer should ensure that such a notice does not conflict with existing studies or proposals.

ILLUSTRATIONS

High quality black and white photographs are preferred for most illustrations, particularly of patients. Colour illustrations can be accepted; however, authors are asked to pay part of the cost, so their desirability should be discussed in advance of submission. All identifiable photographs of patients must be accompanied by written permission for use.

NOTES ON NOMENCLATURE

Authors should refer to the following publications.

(1) Chromosomes: *ISCN 1985. An international system for human cytogenetic nomenclature.* Basel: Karger, 1985.

(2) Genes: Shows TB, *et al.* In: Human Gene Mapping 5 and 7. *Cytogenet Cell Genet* 1979;**25**:96–116, 1984;**37**:340–3.

(3) Loci: Conventional nomenclature should be used with lower case lettering as appropriate (for example, Race RR, Sanger R. *Blood groups in man.* 6th ed. Oxford, London: Blackwell, 1975; and Giblett ER. *Genetic markers in human blood.* Oxford, London: Blackwell, 1969).

(4) Blood coagulation: International Committee of Haemostasis and Thrombosis (Graham JB, *et al*). A genetic nomenclature for human blood coagulation. *Thromb Haemostas* 1973;**30**:2–11.

(5) Enzymes: *Enzyme nomenclature: recommendations of the nomenclature committee of the International Union of Biochemistry.* New York: Academic Press, 1984.

Specific instructions to authors

Papers, which should be in triplicate and in the Vancouver style (*BMJ* 1988;**296**:401–5), should be sent to the Editor, *Journal of Medical Genetics*, Box 238, Level 3, Laboratories Block, Addenbrooke's Hospital, Hills Road, Cambridge CB2 2QQ, UK and not to individual editors, with the exception of papers from the USA, which can be submitted to the North American Editor, Dr P M Conneally, Department of Medical Genetics, James Whitcomb Riley Hospital for Children RR129, Indiana University Medical Center, Indianapolis, Indiana 46223, USA. Submission of a paper will be held to imply that it contains original work which has not been previously published. It is the responsibility of the submitting author to ensure that all the co-authors are agreeable for their names to appear on the manuscript. A fax number should be provided. Permission to republish must be obtained from the Editor.

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