gorilla. Chiarelli was writing soon after fluorescence and banding of chromosomes was discovered. On morphological grounds and from replication patterns he suggests that the human chromosome 1 corresponds to a Robertsonian translocation of the chimpanzee 15 and 13. This has presumably already been confirmed or contradicted with the new techniques.

C. O. CARTER

Book Reviews


As presenting up-to-date authoritative reviews of various aspects of medical genetics this series has clearly established itself. This new volume covers a number of widely differing disciplines. Fenner deals with genetic aspects of viral diseases of animals. At first sight this might not seem of particular relevance to medical geneticists. However the unravelling of the genetic constitution of animal viruses is an enthralling story which will encourage those who hope that one day a similar level of sophistication may be attained in higher organisms, including man.

German considers in detail the relationship between chromosomal rearrangements and cancer. Put simply the argument runs as follows. Many human cancers have one or more demonstrable marker chromosomes derived from normal chromosomes by breakage and rearrangement. There are essentially two different ways in which such marker chromosomes might arise. Firstly the particular cell which first underwent conversion to a cancerous cell and became the progenitor of all the cells of the cancer itself contained the mutated chromosome. Alternatively the chromosome rearrangement might arise in one of the descendants of the first cell to undergo neoplastic change which had a normal karyotype. German argues that evidence from patients with certain Mendelian disorders associated with an increased risk of cancer (such as Bloom's syndrome, Fanconi's anaemia, Louis-Bar syndrome, and xeroderma pigmentosum) tends to favour the former explanation, i.e., that chromosomal rearrangements occur in the cell which originally becomes neoplastic. However it is probable that such chromosomal rearrangements may have little to do directly with cancer other than to provide a predisposing background.

Morton writes eloquently, though to a medical practitioner perhaps not entirely convincingly, on the future of human population genetics. Kirkman's chapter on enzyme defects is a very clear and comprehensive review. Mechanisms of enzymic deficiencies, detection of heterozygotes, and treatment of enzyme defects are discussed. A particularly intriguing problem for the human biochemical geneticist is the molecular mechanism of dominant disorders other than the haemoglobinopathies. Apart from angioneurotic oedema in which there is a deficiency of an inhibitor of C'1 esterase, a deficiency of a specific protein has so far not been demonstrated in any autosomal dominant disorder. Kirkman discuss several possibilities for the molecular basis of such disorders, including defect in a rate-limiting enzyme, mutant gene producing an abnormal protein with deleterious effect or altered substrate specificity, or a mutant gene resulting in excessive enzymic activity.

Clarke discusses in detail the whole problem of the prevention of Rh isoimmunization which he and his colleagues in Liverpool have pioneered. It is gratifying to learn that there are virtually no risks in giving anti-D as a prophylaxis even if given again in a subsequent pregnancy. Yet another area pioneered by the author is the subject of disorders of ganglioside metabolism reviewed by Brady. Not only are chemical methods now available for establishing the diagnosis in affected individuals (and detecting heterozygotes in some instances), but also for the antenatal diagnosis of these disorders. Finally there is a very comprehensive review of the genetics of short stature by Scott. It is gratifying to realise that by clinical and radiographic studies much of the heterogeneity within this group of diseases is being clarified, an essential step for giving reliable genetic counselling.

As with previous volumes in this series, this one can also be highly recommended to all interested in developments in medical genetics.

ALAN E. H. EMERY


This book contains the three papers read at a symposium held at Sunland Training Centre at Miami, Florida. An opening statement by Arnold Cortazzo precedes the papers, which are followed by three brief closing statements.

A problem for speakers at a symposium of this kind is to judge the level of understanding of those whom he has to address and to set his standard and style accordingly. This symposium was sponsored by the South Florida Foundation for Retarded Children, the Sunland Training Centre, and the Hospital Improvement Program and no doubt there would be a mixed lay and professional audience of parents, social workers, psychologists, doctors, and others.

The opening statement by Arnold Cortazzo is clearly intended to soften up the audience for what is to follow and is aimed at those with little or no scientific knowledge. It describes the cell and cell division with a brief explanation of genes, Mendel's laws, and the molecular structure of genes. Too much is attempted in too small a space (about 10 pages of text). It would have been better without any mention of the 'DNA staircase', code letters, messenger RNA, and so on; none of which is necessary for an understanding of what follows. The account of the reduction division is a little confusing.

Dr Smith talks about the clinical aspects of genetics in mental retardation and, of the three chapters, this is the
Aminoacidopathies, Immunoglobulinopathies, Neuro-genetics and Neuro-Ophthalmology.


Volume 6 in the series of Monographs on Human Genetics is the proceedings of a conference held in 1970 on Neuro-Genetics and Neuro-Ophthalmology, where the two main chosen topics were aminoacidopathies and immunoglobulinopathies.

In the first section there has been some repetition, particularly concerning phenylketonuria. The late D. Hsia comments on the allelic variants in several of the aminoacidopathies and discusses the excess of phenylketonuric males ascertained in screening surveys. He reports briefly on the early work with somatic cell cultures, a field which has rapidly advanced since then. T. Gerritsen and F. Siegel describe animal models for hyperphenylalaninaemia and discuss the possible mechanisms of brain disturbance. If such mechanisms were understood one outcome could be helpful suggestions for treatment. J. Frézal reviews enzyme defects in the aminoacidopathies; some tables here would be helpful. There are two useful accounts of clinical manifestations: one by C. Hooft and D. Carton on the ways in which these disorders may present in a paediatric department, emphasizing, as do other authors, how there may be no disease with an aminoacidopathy. J. François gives an excellent account of ophthalmological findings. The last in this section is a good review article by A. Barbeau on dopamine in the nervous system, a subject, where, again, several advances have been made more recently.

Probably the best article is that by S. Barndun, who presents for neurologists and ophthalmologists a beautifully clear account of immunodeficiency disorders. This is followed by an interesting chapter on slow viruses and neurological damage, where J. Hotchin discusses the occurrence of several strains of lymphocytic chorion meningitic viruses with different affinities for liver and brain; and the probable role in persistent infection of an auto-immune component.

The offered contributions include an account by F. Mollica et al of symptomless hyperprolinemia in a family: an account by B. Neville of 4 patients with hypogammaglobulinaemia who developed progressive neurological disease; an account of a prospective study by C. Rose of 170 patients with optic neuritis, in which he concludes that all cases of optic neuritis are a manifestation of demyelinating disease; and a description by P. Thomas and D. Calne of two autosomal dominant forms of hypertrophic neuropathy. It is unfortunate that the editors did not insist on all authors reporting their results for this book; some summaries, presumably prepared in advance for the conference, mention methods only, as for example, the tantalizing bare three sentences on family studies of immunoglobulin levels by W. Billewicz et al.

This is a nicely presented and prepared monograph. However its field is too wide to be useful in its entirety to neurologists, paediatricians, or ophthalmologists. Certainly an account of the conference is valuable, but perhaps a hard-backed well-printed book is too lavish a way to do it.
The Role of Genetics in Mental Retardation

B. W. Richards

doi: 10.1136/jmg.9.4.486-a

Updated information and services can be found at:
http://jmg.bmj.com/content/9/4/486.2.citation

These include:

Email alerting service
Receive free email alerts when new articles cite this article. Sign up in the box at the top right corner of the online article.

Notes

To request permissions go to:
http://group.bmj.com/group/rights-licensing/permissions

To order reprints go to:
http://journals.bmj.com/cgi/reprintform

To subscribe to BMJ go to:
http://group.bmj.com/subscribe/