mosome breakage syndromes (2 pages). Extensive coverage seems to be the primary aim of this type of publication and it is the most complete catalogue of human chromosome variation there is to date. It should be, as the author hopes, the first source to be consulted when references on a specific chromosomal abnormality are sought.

The basic idea and cataloguing arrangement are good but Chromosomal Variation in Man will have to be regularly updated and extended to realize its full potential and become a chromosomal 'McKusick'.

MICHAEL BARAITSER

New Chromosomal and Malformation Syndromes.


This is Vol. XI. No. 5 in the Birth Defects: Original Article Series and contains selected papers from a conference held in California in 1974. In a previous publication in this series called Malformation Syndromes (1974) the editor was criticized for the choice of title which misleadingly implied a comprehensive review of the subject. The present volume has thus been entitled New Chromosomal and Malformation Syndromes. Geneticists seem not only to be running out of titles for books, they are running out of titles for syndromes. In this edition we have the 3M syndrome (a name derived from the first letter of the surnames of three of the five authors—one can only feel sorry for the two left out), the KGB syndrome, the G syndrome, and the Floating Harbor syndrome.

What is clear from looking at the superb photographs is that despite all misgivings about the publication of conference papers, the clinical geneticist cannot see enough of the new malformation and chromosomal syndromes, because of the number of malformed infants he sees and is unable to diagnose.

The book is almost equally divided into the chromosomal and non-chromosomal malformation syndromes. There is the inevitable endeavour to describe recognizable syndromes associated with chromosomal abnormalities, and trisomy 9p and trisomy 22 might qualify for recognition. Evidence is presented for the acceptance of three new short stature syndromes and among the better known entities, diagnostic criteria for the whistling face syndrome are reviewed and the variable limb malformations in the de Lange syndrome are documented.

New Chromosomal and Malformation Syndromes is, like the loose-leaf series 'Syndrome Identification', another step forward in trying to make clinical meaning of the multitude of congenital malformations.

MICHAEL BARAITSER


Clinical geneticists have been eagerly awaiting this book. The great experience of Dr Bussey in this field is unique, having been associated with Cuthbert Dukes at St Mark's Hospital from 1924. When he retired in 1974 he had been closely concerned with the investigation and recording of nearly 300 families with intestinal polyposes of all types. The book is somewhat disappointing in that it is much shorter than had been hoped—only about 100 pages including 64 figures and 14 tables. In addition Dr Bussey does not give his own personal opinion on several controversial aspects of the subject, but merely quotes the opposing theories. There is little discussion of the possible reasons for the remarkable 45% incidence of solitary cases, nor why in only 9 out of 99 of these patients presenting with a negative family history, did a child or other relative subsequently develop polyposis during a presumably lengthy follow-up period.

Notwithstanding these criticisms, the book is of great value. It contains many tables of data which reveal much about the condition. Dr Bussey's research has been connected mainly with the cancer aspect of the subject and the chapter on the relation to carcinoma is one of the high spots of the book. In these data 62.2% of the 293 patients presenting because of symptoms, already had malignant disease, and multiple carcinoma were found to be 12 times more frequently encountered in polyposis patients than in sporadic colonic cancer.

Dr Bussey sees the main value of the study of polyposis as the light it may throw on the wider problem of intestinal adenomas when solitary or few in number. He considers the suggestion that all adenomas of the large intestine have a genetic origin, though he does not rule out an interaction of environmental and genetic factors even in multiple polyposis families. The intriguing regression of rectal polyps after colectomy is discussed in this context.

For 25 years Gardner's syndrome has been regarded as a separate genetic entity, though the Swedish data of Thor Alm have thrown doubts on this by revealing many families in which only one of several polyposis members had an extracolonic lesion. The St Mark's data also contain 30 families in which only one member had the lesion of Gardner's syndrome. Nor was any difference found between the number of polyps and their distribution in polyposis families with and without Gardner's syndrome. Dr Bussey does not dismiss the idea that all adenomatous polyposis may be Gardner's syndrome with different degrees of manifestation of the subsidiary lesions.

The book is well written and the numerous photographs are excellently reproduced. The references grouped at the end of the book are not comprehensive on the genetics of the subject, but are a useful collection on its neoplastic and clinical aspects. It can be recommended not only to clinical geneticists but also to gastroenterologists and surgeons who meet patients with
colonic polyps in their clinical practice. Pathologists and others interested in the development of cancer will be particularly pleased to have this book on their shelves.

R. B. McCONNELL


Congenital malformations impose a heavy psychological burden on the family and an economic burden on society. In recent years their relative contribution to morbidity and mortality in childhood has increased dramatically as other causes have declined. However, as Professor McKeeown points out in his thought-provoking introduction to this monograph, apart from one or two notable exceptions (e.g. rubella and thalidomide embryopathies) and despite several decades of research we still have little idea of their causation. Yet only when we have identified a specific causative agent may true prevention be really possible. Until then we shall have to rely on identifying those who are at risk of having affected children and then providing them with genetic counselling and antenatal diagnosis. This is by no means an entirely satisfactory solution and certainly selective abortion should only be considered a 'holding measure'. This volume is, therefore, particularly valuable because all the contributors address themselves to these various problems.

The genetics of congenital malformations (Carter) and developments in the antenatal diagnosis of chromosomal disorders (Laurence and Gregory) and CNS malformations (Brock) are well summarized, though one might hope that non-invasive techniques, such as sonography, might one day replace amniocentesis for the antenatal diagnosis of certain malformations (MacVicar). However, if, as one suspects, more investigators are now turning their attention to the possibility of primary prevention through an understanding of causation, then the remaining contributions are particularly relevant. Chapters on environmental teratogens (Smithells), drugs (Berry and Barlow), and infective agents (Dudgeon, Mims) are well documented accounts of our present knowledge in these fields. At present most investigators seem to favour an epidemiological approach to understanding aetiology from studying incidence figures in various population groups (Leck) and the development of surveillance systems (Weatherall and Haskey) whereby it is hoped that an 'epidemic' of a particular malformation might be recognized as soon as it occurs and that this is then more likely to lead to the identification of the causative agent. A hypothesis generated from data obtained from such epidemiological studies may then be tested either in the general population (e.g. to determine if the incidence declines with avoidance of a suspected teratogen) or on animal models. Alternatively, drugs and infective agents known to be teratogenic in animals may be given particular consideration in studying the incidence of congenital malformations in man. Several contributors deal with the problems of animal models: the numerous models which are available (Beck), and ideas of pathogenesis which such models can produce (Poswillo, Wolpert). The possibility of mechanical factors operating in utero as a cause of congenital malformations is also discussed (Dunn).

It is a little disconcerting, however, that in only one instance so far has an epidemiological observation on a human congenital malformation ever led to any idea of a mechanism (Penrose's observation of two maternal age groups in Down's syndrome which ultimately proved to be two different chromosomal abnormalities). Further, that the two most clear-cut teratogenic agents so far identified in man (rubella and thalidomide) were recognized as such by astute physicians without recourse to statistics or laboratory studies. Nevertheless with the increasing attention which this field is now attracting, it seems more likely that epidemiological and animal studies may prove more helpful in the future in identifying causative agents and thus lead to prevention.

ALAN E. H. EMERY


The books successfully summarize the clinical aspects and the advances in understanding of the pathogenesis of haemophilia from recent and continuing research. As with most multi-author texts there is some overlap between chapters. This might have been avoided in some instances by condensing related chapters into a single chapter.

It is unfortunate that these books should be so expensive as this may prevent them being as widely and fully read as the authors had hoped. To reduce publishing costs the editors could perhaps have eliminated chapters which contribute little to the books, for example, 'Incidence of Haemophilia in South Africa' is condemned by its authors' opening sentence and the chapter on transplantation is superfluous when the site of procoagulant FVIII is unknown.

Although the chapters on specific blood products are of interest to the specialist, these chapters might have been better replaced by a chapter comparing the properties of different products. Among topics that might have benefited from further discussion is the detection of haemophilia carriers. In Chapter 30 it is unfortunate that the achievements of present therapy have been swamped by the discussion of the inadequacies.

Despite these criticisms, the book contains many useful chapters and readers wishing to pursue any subject further will find the comprehensive reference list invaluable.

F. G. H. HILL