Book reviews

Journal of Medical Genetics, 1985. 22, 237–240


This special issue should become a vade mecum for any cytogeneticist involved in prenatal diagnosis because it contains such immediately practical and useful information, especially about those relatively rare situations where no individual laboratory can collect enough data to arrive at a firm conclusion about the significance of a finding. I hope that this sort of data will continue to be collected and reported on regularly in the future.

The first section gives maternal age specific rates for chromosome aberrations and exhaustively analyses paternal age data. It would have been interesting to have seen the analysis of parental age effects done for male and female trisomy 21 cases separately in view of the findings of Hassold et al (1984), but perhaps this can be done next time.

Boué and Gallano give differential risks of an abnormal outcome in pregnancies to parents carrying varied balanced translocations. Translocations ascertained through amniocentesis for other reasons are excluded from these figures and I did wonder whether the elimination of these previously trouble-free rearrangements might lead to an overestimation of risk. By sampling error alone some families would be expected to show no adverse outcome despite the risk, but they still form part of the population of outcomes on which risk figures should be based.

There is a particularly informative section on de novo structural rearrangements. These cases are among the most difficult on which to give advice and this information will make that job much easier. The section highlights the need for more extended follow up of such cases. Most assessments are made immediately, at birth, and few cases are followed to the time when developmental delay or retardation might be expected to be detectable.

It is an indication of the difficulty of assessing mosaicism that three studies of this problem are presented. The Canadian survey produces particularly helpful guidelines for establishing the most effective way of distinguishing between ‘true’ and pseudomosaicism and in this, and the European study, there are helpful notes about presumptive maternal cell contamination. Again the necessity for full and extended follow up is stressed, without which the long term significance of such aberrations cannot be assessed.

For me, this presentation reinforced with hard data impressions gained from a relatively small number of cases. My job is going to be easier because of the broad based information included in this special issue.

P Cooke

Catalogue of Unbalanced Chromosome Aberrations in Man

This book, as indicated in the title, is primarily a catalogue of chromosome imbalances reported in man. It is intended for cytogenetists and clinicians who deal with chromosome disorders and is an endeavour to provide a comprehensive review consisting of concise, easily accessible information together with recent references for further studies.

The first chapter introduces the reader to a variety of cytogenetic topics including nomenclature, techniques, variants, fragile X, population studies, and rearrangements. The latter refreshingly describes some of the rarer structural aberrations and, despite the occasional factual error and omission, is both well presented and informative. The notes that follow on clinical findings in autosomal and sex chromosome aberrations succeed in identifying patterns of abnormality, while an index of selected malformations and minor anomalies, which correlates specific abnormalities of the phenotype and karyotype, is provided towards the end of the book.

A gene map (1982) is another useful inclusion.

The catalogue of aberrations, which comprises the major part of the book, lists imbalances of chromosomes 1 to 22 with sections also on polyploidy and the sex chromosomes. Combinations of imbalances are also included and a diagram (ISCN, 1978) of the relevant chromosome on each page allows the relative position of any segment to be determined quickly. An attempt is made to correlate significant clinical findings with imbalances of specific chromosomal segments. Clinical entities of questionable validity based, for example, on overlapping deletions or duplications are therefore avoided and an emphasis is placed on recording malformations,
mental development, and survival; dysmorphic features are only briefly mentioned. A large number of minor syndromes are thus delineated. However, within major syndromes there is little or no subdivision of the text so that specific information is not readily retrieved. Furthermore, the author departs with little justification from ISCN (1978), and the clinical illustrations might be more conveniently located interspersed within the text than in a group at the end of each chromosome section. On the other hand, published reports up to 1982 appear to be thoroughly and critically reviewed, with the more recent important references listed for each aberration, while the latest reports are included as addenda. The references are completed by a first author index.

The field of clinical cytogenetics, after the technical advances of the last 15 years, has seen an almost bewildering increase in the number and variety of published cases with imbalances resulting from loss or gain or both of parts of chromosomes. It has reached the stage where the need to review and catalogue comprehensively has become acute. This volume, which will require regular updating, makes a very significant contribution to that process and is likely to become an indispensable acquisition for cytogenetic laboratories and clinicians with an interest in cytogenetics. However, there are some deficiencies in its organisation and presentation that might usefully be corrected in a future edition.

S H Roberts

Clinical Atlas of Human Chromosomes

The first edition of this useful book was published in 1977 and contained 319 pages. The second edition, in similar format, is 487 pages long. Not only is there more information on all conditions, but more than 30 chromosomal syndromes—collections of cases with duplication or deletions of chromosome segments with recognisable clinical similarity—are described.

The authors take one through the human karyotype, chromosome by chromosome, and arm by arm. They document the morphology of each chromosome, normal genes located on it, and described abnormalities with their clinical correlates. The clinical illustrations, perforce largely reproduced from previous original publications, are sometimes less than excellent, but are adequate for their purpose.

In place of a conventional index, the book contains a ‘syndrome finder’, a list of major clinical features, leading one to the chromosomal disorders in which the clinical sign occurs. I failed to find the Prader-Willi syndrome (or chromosome 15) mentioned under hypotonia, obesity, or genital anomalies, but the system in general seems to work well. I would personally have welcomed some way in which named syndromes could be located by their eponyms, rather than by their individual clinical components.

Although dated 1984, I suspect that this is a translation of the French edition, which is a little older. Perhaps for this reason, the Langer-Giedion syndrome, associated with abnormalities of 8q, appears in the foreword, but not in the text.

These are, however, minor blemishes in what is generally a thoroughly useful book. As the range of detectable chromosomal abnormalities increases, and particularly with the advent of prometaphase chromosome analysis, it becomes less and less satisfactory simply to give the laboratory a blood sample labelled ‘abnormal’ and assume that they will inevitably find whatever is wrong. With long chromosomes, directed attention will efficiently lead the person at the microscope to confirm or exclude particular abnormalities, and I would guess that this will become increasingly true over the coming years.

The authors hope that this current edition will have a long and happy life; I think that we may all be crying out for a third edition sooner than they expect, as we continue to explore the world of single band deletions.

Cytogenetics laboratories have a need to compare the clinical information on a patient with previous descriptions of the same abnormality as a check on their cytogenetic diagnoses. Clinical geneticists need ready references to dysmorphic syndromes, to provide maximum prior information to the laboratories. Both groups will find this book excellent for their purposes.

Martin Bobrow

Principles and Practice of Medical Genetics

This book marks, as the editors say in their introduction, the coming of age of Medical Genetics as a speciality in its own right. The fact that it has been written at all shows how extensively the subject has developed over the past 20 years, and both editors and publisher deserve much credit for putting this major synthesis on the record.

In many ways this book complements another