



Prenatal Diagnosis. Special Issue. Collaborative Studies in Prenatal Diagnosis of Chromosome Aberrations

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Book reviews

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Prenatal Diagnosis. Special Issue. Collaborative Studies in Prenatal Diagnosis of Chromosome Aberrations.

Volume 4. Edited by J L Hamerton and M A Ferguson-Smith. (Pp 162; figures + tables. £25.00.) Chichester: John Wiley, 1984.

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

By Albert Schinzel. (Pp xx + 913; figures + tables. DM 298.) Berlin, New York: Walter de Gruyter, 1984.

This book, as indicated in the title, is primarily a catalogue of chromosome imbalances reported in man. It is intended for cytogeneticists 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,