BOOK REVIEW

Catalogue of Unbalanced Chromosome Aberrations in Man

2nd edition. Albert Schinzel. (Pp 966; SFr 397.) Berlin: Walter de Gruyter, 2001. ISBN 3-11-011607-3.

It is difficult to do justice to the scale of achievement that this book represents. Here, in a single volume, are the phenotypes associated with the great majority of cytogenetically visible unbalanced chromosome abnormalities. At the turn of the century, this volume contains much of the information gathered since the modern era of chromosomal pathology began with the identification of the extra chromosome 21 in Down syndrome in 1959.

The first edition in 1984 was an essential publication for both clinical genetic and cytogenetic departments. The new edition contains a great deal of additional information with many references as recent as 1998 and some from 1999. The new edition also follows the same overall pattern as the first, with the cytogenetic findings dictating the order of the clinical summaries; thus, the abnormalities are ordered first by chromosome number, secondly by the type of unbalanced rearrangement (monosomies, deletions, duplications, trisomies), and thirdly according to the chromosomal segment ordered from centromere to telomere. The different types of chromosomal imbalance are reviewed in a beautifully illustrated introductory chapter, but some familiarity with the International System for Human Cytogenetic Nomenclature is useful to navigate the catalogue. Non-mosaic imbalances are followed by mosaic imbalances of the same chromosomal segment and "pure" imbalances of a particular chromosomal segment are followed by the "mixed" imbalances, as in unbalanced translocations, where there are two or more concomitant imbalances. Rings and marker chromosomes appear in sections depending on whether their presence leads to a net gain or loss of genetic material. Uniparental disomies which require molecular genetic analysis are included, as are some submicroscopic rearrangements detectable only by FISH.

The majority of the book and its key components are the phenotypic summaries which follow each and every specific imbalance. These summaries are at the same time both comprehensive and concise. They are effectively mini-reviews of the majority of features which have been associated with a particular imbalance. The references on which the summaries are based are listed underneath each summary. Many of the classical or commoner chromosomal syndromes are illustrated with one or more sets of photographs from which the facial "gestalt" can be immediately appreciated. Listing every feature can give the impression that a phenotype is relatively severe. Thus, cases of XYY with features such as renal agenesis, cystic dysplasia, iris coloboma, and multiple anomalies are included. At the same time, these observations are tempered by reference to the much milder picture of XYY patients that has emerged by following up cases of XYY ascertained at prenatal diagnosis. Similarly, it is useful to have a catalogue of the growing list of the less common manifestations of del(22)(q11.2q11.2) at the same time as an acknowledgement that mental retardation is not an invariable feature and that transmitting carriers are frequently less severely affected than the offspring through which the parents were ascertained. Naturally, in a work of this size and scope, there are individual entries and interpretations with which some people might quibble, but this does not detract from the whole as a work of reference.

Readers may ask why a catalogue of this kind is still needed when resources such as PubMed are so widely available and, indeed, the author's own data are the basis of the Oxford Cytogenetic Database. The strength of the current volume is, however, that a wealth of specific edited information is gathered in a single place. It is also a useful source of references from the 1960s and 1970s, which are not always so easy to find using electronic means. The time saved in chasing relevant published reports is likely rapidly to repay the cost of this volume in most professional genetic centres. It is also a pleasure to hold, handle, and browse through. No doubt an electronic version will appear but, even so, this reviewer believes that this is a volume which no genetic centre should be without, even in the 21st century. Let us hope that the publishers will not attempt to create artificial differences between electronic and hard copy versions in an attempt to make it necessary to pay for both; this publication is an international resource that should be as widely available as possible.

The Americans may have McKusick but the Europeans have Schinzel. This catalogue is a unique achievement, which will never be repeated unless the author has the time and energy for a third edition. As the foreword proclaims, this "really is a monumental work" and the genetic community should be grateful to the author for his diligence, erudition, and Herculean persistence over so many years.

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