Book Reviews

Journal of Medical Genetics (1975). 12, 431.

Malformation Syndromes. Edited by Daniel Bergsma. (Pp. 196; figures + tables. \$14.50.) *Birth Defects Original Article Series*, vol. 10, no. 7. National Foundation, March of Dimes. New York: Intercontinental Medical Book Corp. 1974.

A misleading title—the book records papers published at the 1973 Boston Conference on Diagnosis, Genetics and Management of Birth Defects, but is not a systematic review or bibliography of malformation syndromes. The chapter headings show it to include several general papers on nomenclature or classification of syndromes; these, by Warkany, McKusick, Smith, Herrmann, and Opitz, are of interest, and the last named particularly thorough in discussion of basic concepts: inborn error, dysplasia, malformation, etc., but, together with a check list for facial malformation syndromes, they account for only 34 pages (17%) of the book. The remainder is given up to papers of limited scope: case and family reports, and reviews of rare syndromes; and 7% of the book is virginal paper. It does not live up to expectations engendered by the publishers' puff '... the latest and most complete examination of malformation syndromes available...', and is not a good investment at the English equivalent of \$14.50.

R. H. LINDENBAUM

Skeletal Dysplasias. Edited by D. Bergsma. (Pp. 266; figures + tables. \$14.50.) *Birth Defects Original Article Series*. National Foundation, March of Dimes. New York: Intercontinental Medical Book Corp. 1974.

The once neglected field of bone dysplasias is now receiving widespread interest, and several excellent books have recently appeared which provide general reviews of the subject, despite our fragmentary knowledge of most of these disorders. This volume, the report of a conference held in Boston in 1973, does not attempt to cover the field systematically; however, the contents in general complement rather than duplicate the earlier volume on the subject produced by The National Foundation, and the standard of most contributions is high. Abundant radiographs and photomicrographs, reproduced with fair quality, add greatly to the value of the book, and the price is not excessive by present standards.

Several contributions deserve special mention. The opening chapter by Rimoin describes histological studies on bone and cartilage biopsies which in the past have been remarkably lacking and often confusing due to erroneous clinical diagnosis. Findings of particular interest to the medical geneticist are the identical appearance of changes in the mild 'variant' of diastrophic dwarfism to those of the classical disease, suggesting the same basic defect and possibly allelism; by contrast those familial cases of

thanatophoric dwarfism studied histologically showed an entirely different appearance to those seen in isolated cases, suggesting that heterogeneity may be present, and that most isolated cases represent new dominant mutations.

The article by Spranger and Langer on spondylo-epiphyseal dysplasias is valuable for its clarity in what seems at times a hopelessly confused area. They differentiate a number of disorders which can be considered well delineated, and separate them from the large remaining pool of conditions where classification is at best provisional. Pozanski and Garn tackle another neglected area, the quantitative measurement of radiological bone changes. They outline a series of key measurements and ratios which are likely to prove valuable, and which hopefully may replace the somewhat 'impressionistic' and subjective interpretation so often used at present.

Remaining chapters of the book give reviews and case reports on specific dysplasias such as Kniest syndrome, thoracic dysplasia and related disorders, and the trichophino-phalangeal syndrome. The increasing integration of histological, biochemical, and genetic data with clinical and radiological information shows that progress is steadily being made towards a fuller understanding of this complex group of inherited disorders.

PETER S. HARPER

Clinical Cytogenetics and Genetics. Edited by Daniel Bergsma. (Pp. 201; figures + tables. \$14.50.) *Birth Defects Original Article Series*, vol. 10, no. 8. National Foundation, March of Dimes, New York: Intercontinental Book Corp. 1974.

This small March of Dimes volume is the text of the conference in Boston, presumably 1974, on syndromes mostly associated with chromosomal anomalies. O. J. Miller gives a clear account of the new banding techniques and W. R. Breg the application of these new methods to identify the individual chromosomes involved in conditions such as Down's, Edward's, and Patau's syndromes, and to identify chromosomes involved in translocations. He notes for example that the usual D/G translocation in Down's syndrome is between 14 and 21; that the deleted chromosome seen in chronic myeloid leukaemia is 22; that the D chromosome with the long arm deletion associated with mental retardation in retinoblastoma is 13. E. Early and W. Stanley report the first instance of an *in vitro* cell culture system (in the rabbit), in which non-disjunction of acrocentric chromosomes occurs spontaneously. P. Neurath describes the current status of computer analysis of chromosomes. He estimates that one needs to be doing 10,000 karyotypes a year to justify running a computer system with a scanner.

The second half of the book is concerned with a variety