Book Reviews

Birth Defects: Original Article Series, V, 3. First Conference on The Clinical Delineation of Birth Defects, Part III, 'Limb Malformations'. (Pp. 234; figures+tables. £7.) New York: The National Foundation—March of Dimes. Edinburgh: E. and S. Livingstone for Williams and Wilkins.

This is a report of the proceedings at the First conference on the 'Clinical Delineation of Birth Defects' held at the Johns Hopkins Hospital, Baltimore in May 1968. Volume III is devoted to limb malformations and attempts to distinguish fundamentally distinct anomalies or groups of anomalies as a first step in the elucidation of pathogenesis, and as a guide to genetic counselling and planning of possible future treatment.

A review of the whole problem with particular reference to the classification of anomalies was given by Dr Lenz, Dr Freire-Maia, and by Dr O'Rahilly. Dr Pfeiffer reviewed the numerous 'head and hand' anomalies and dermatoglyphs were dealt with by Dr Alter and Dr Warburton. Much of the volume deals with a variety of rare syndromes, some never previously reported, and there are a large number of excellent illustrations throughout.

The largest single contribution is from Dr Temtamy and Dr McKusick who are to be congratulated on their classification of limb anomalies based on anatomical and genetic characteristics. This was arrived at by a careful study of many families seen personally and appraisal of other reported cases. They take each type of deformity and consider it first as an isolated phenomenon and then where it occurs as part of a generalized disorder or syndrome. This is a more helpful way of considering the numerous malformations of the limbs than those classifications based solely on details of anatomical variation.

The anomalies dealt with are still only at the stage of clinical description and genetic analysis and the book will be mainly of value to clinicians and clinical geneticists. It is to be welcomed for its presentation of carefully observed cases and it is to be hoped that there will be future conferences where material collected from many centres can be presented and compared in this way.

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The first conference of the 'Clinical Delineation of Birth Defects' was held over 5 days in May 1968 at the Johns Hopkins Hospital and proceedings have now been published in 5 volumes, of which this is the second. It is aptly placed between Part I, a thin volume of three essays on biological development and nosology of genetic disease and Part III which is made up of a series of articles on limb malformations. Part IV was devoted to skeletal dysplasias and Part V to the phenotypic aspects of chromosomal aberrations.

The prologue to this volume by Dr Opitz clearly underlines the difficulties that face those who study and try to classify the malformation syndromes. This introduces a series of articles on what may be specific malformation syndromes. Some of these are reviews; Rubinstein's paper on the broad thumb and big toe syndrome is one excellent example. Others are restricted to the description of very rare and familial conditions. In all, 19 conditions are covered. Many of these articles are written by the authorities on the subject though the Wisconsin group, headed by Dr Opitz, appears to overwhelm the others. This volume is not aimed at giving a comprehensive list and description of all the malformation syndromes, so it should not be bought in the hope that it will provide this information. It is perhaps unfortunate that the Wisconsin group have seen fit to use the initial letter of the index family as titles for some of the conditions they describe, and although this should be considered a brave attempt to avoid eponyms single or even multiple letters are not impressive as *aide memoirs* and are certainly not descriptive. The articles themselves are clear and are beautifully illustrated with numerous photographs. There is in addition a small number of articles which consider malformations on a rather broader basis; Gorlin describes 8 specific syndromes that affect the face and George Fraser considers malformations with eye and ear involvement. Pruzansky shows that careful x-ray measurements of the developing mandible gives precision to diagnosis and provides a useful record which will define the degree of defect associated with a specific condition. A further one sixth of the book is filled with straight case reports that are without comment.

The volume is almost closed by an epilogue by Polani who reminds us that disorders that bear a resemblance to each other can only be effectively separated from each other when linked to some recognizable metabolic or cytological label. Right at the end David W. Smith sneaks in with an appendix of simple tables that attempt to classify abnormalities according to the most obvious affected tissue or organ or to the underlying defect when it is known.

Birth Defects: Original Article Series, V, 2. First Conference on The Clinical Delineation of Birth Defects, Part II 'Malformation Syndromes'. (Pp. 284; figures+tables. £7.) New York: The National Foundation—March of Dimes. Edinburgh: E. and S. Livingstone for Williams and Wilkins.