
Ophthalmological genetics has an extensive literature going back in a substantial way to as early as 1858 when the hereditary character of retinitis pigmentosa was first fully appreciated. The monographs by Julia Bell in the Treasury of Human Inheritance summarized much of that literature and began a tradition which has been well maintained in recent years by the sumptuous volumes currently brought out by Waardenburg and his associates, and by the present comprehensive study on the chorio-retinal heredo-degenerations; this covers a very wide field in great detail. Some 200 pages are devoted to the anatomy of the retina and the clinical methods employed in assessing function, considerable attention being paid to the testing of the colour sense and the use of electro-diagnostic procedures. This section—Part I of the book—is a monograph on its own; it is up to date and includes electronmicroscopy studies. Part II, which runs to about a thousand pages, carries five chapters. The first is devoted to autosomal chorio-retinal affections and covers retinitis pigmentosa and its variants; the second chapter deals largely with choroidal lesions; sex-linked chorio-retinal affections are the subject of the third chapter, while retinal cyst formation, vitreous anomalies, and allied affections are dealt with in the succeeding chapter. The last chapter in this part is devoted to chorio-retinal affections with associated general and ocular disorders. This contains a most useful and well-documented account of syndromes not readily found in textbooks. Part II is relatively short. Its 300 pages cover nightblindness, the different varieties of colour anomalies, as also the retinopathies of viral, toxic, or infective origin. The concluding 130 pages are given to three further parts dealing respectively with morbid anatomy of the heredo-degenerative lesions, treatment, and experimental induction of retinal degeneration.

In a book of this scope, no reviewer would be at a loss to find views and statements from which to differ. What is relevant is that this is a careful, well-documented account of current teaching. If for nothing else, this volume is welcome for the considerable elimination of mere labels in its presentation—an admirable feature that might perhaps have been carried still further.

A.S.


In 1959 the Medical Research Council’s Clinical Effects of Radiation Research Unit in Edinburgh set up a Registry of Abnormal Karyotypes with the idea of facilitating the study of causes of death of people with chromosome abnormalities, and particularly of finding out whether death from malignant disease was commoner among them than among people with an originally normal chromosome complement. The majority of persons notified to the Registry have anomalies of their sex chromosome make-up, and by the end of 1962 good cytological data, together with a variety of other data, were available on 266 of them. The bulk of the report (Part III) sets out the cytological and other information collected on these persons. Among them there are 134 with male phenotype and 128 with female phenotype. In addition the findings are described in four true hermaphrodites. Among the males there are 99 subjects with an XXY complement and 24 with sex-chromosome mosaicism, mostly with two cell lines, one of which has an XY and the other an XXY complement. Among the females the four largest groups are: an XO group (38 subjects), 22 examples of ovarian dysgenesis with sex-chromosome mosaicism, 33 triplo-X women, and 26 with an XY complement, among whom 20 have
the syndrome of feminizing testis. Chromosomal and nuclear sexing data are listed for each case, and clinical data, and the results of body measurements and of special tests (e.g. Ishihara tests) are indicated. Reasons for referral for cytological studies are given and the conceptional history of the parents is tabulated. This has been checked, wherever possible, against the information in the registers of births, marriages, and deaths kept by the Registrar-General for Scotland. A proportion of the material has already been published and appropriate reference to this is made.

Part I of the Report is devoted to technical consideration and to the problems of sex chromosome diagnosis, and deals with the difficult problem of the definition and detection of sex-chromosome mosaicism, both developmental and proliferative—to use C. E. Ford's terminology. In Part II the literature relevant to the cases studied is briefly reviewed.

This Report is a great service to all interested in human genetics, and cytogenetics in particular, for it brings forward a great deal of painstaking work. The findings recorded, together with those that are being published from other sources, offer a great deal of data for study and analysis and will no doubt act, as indicated in the preface, as a stimulus to speculation and further inquiry. It is hoped that the Medical Research Council, as is their present intention, will publish supplements of new material at suitable time intervals.

P. E. POLANI


This is part of a five-volume handbook of which the first will be devoted to general principles; the remaining four are planned to cover the whole of clinical genetics. Of the six contributors to Vol. II, which is the first to appear and the subject of this review, two, Degenhardt of Frankfurt and Lenz of Hamburg, are in charge of medical genetic units, while the others are clinicians. Degenhardt contributes the sections on the skeleton other than the extremities, while Lenz deals with anomalies of growth and bodily form. The extremities are discussed fully by Grebe of Frankenberg/Eder, while Cocchi of Zürich deals with bony anomalies. A particularly full section on teeth, mouth, and jaws comes from Schulze of Göttingen, while embryopathies in general are dealt with by Goerttler of Kiel in an admirably concise chapter. The book is profusely and most usefully illustrated and the bibliographies carried by the different chapters are full, though not exhaustive. If the rest of the work maintains the high standard of the present volume, this handbook should prove a worthy successor to the earlier encyclopaedia by Just, Bauer, Hanhart, and Lange.

A.S.