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Book Reviews

1. GENETICS

Abnormalities of the Sex Chromosome Complement in Man. By W. M. Court Brown, D. G. Harnden, Patricia A. Jacobs, N. Maclean and D. J. Mantle. London: H.M. Stationery Office. 1964. Pp. 239. Price £1 7*s*. 6*d*.

This M.R.C. Report was compiled and written by a group of people whose contribution to human cytogenetics is and has been considerable. It would not be an exaggeration to say that Court Brown and his co-workers have enriched our knowledge of human cytogenetics more than any other group active in this field.

The present publication is a newly encouraging step and should serve as a stimulus to colleagues who have facilities for similar projects. There can be no doubt that progress in human cytogenetics will be possible only if chromosome abnormalities are investigated not in isolation but as part of a familial study. Such thorough research requires the collaboration of a number of workers, as well as the enlightenment and willingness to co-operate of the families concerned.

The preface traces briefly recent developments of human cytogenetics, beginning in 1956 with the work of J. H. Tjio and A. Levan, who first demonstrated that man has 46 chromosomes. The introduction proper tells us how in 1959 a Registry of Abnormal Human Chromosomes was initiated by the staff of the M.R.C.'s Clinical Effects of Radiation Research Unit in Edinburgh. Then, Part I deals with "cytogenetic techniques, the normal human karyotype and the general principles of diagnosis of sex-chromosome abnormalities". Part II describes "the features of subjects with a sex chromosome abnormality". These include phenotypic males with seminiferous tubule dysgenesis; phenotypic males with other abnormalities (XYY, XO/XY, XX); phenotypic females with ovarian dysgenesis; phenotypic females with an XXX or XXXX complement; and lastly phenotypic females with a male sex chromosome complement. The rest of the book (Part III) consists of 266 case reports. They are arranged according to sexual phenotype (male, female and true hermaphrodite). "Within each phenotypic group the cases are arranged according to sex chromosome complement and within each of these groups in order of increasing age." The case reports consist of reasons for study, parental conceptional histories and deaths of siblings, clinical and histological data, X-linked traits, nuclear sex, and chromosome counts. The list of references is comprehensive and invaluable.

In many ways the Report has been published prematurely. The case reports are still incomplete; on mental illness in particular the data are not sufficient to allow an analysis. In cases with colour blindness no pedigrees are provided. The introductory text of the Report is scanty, we get no real picture of our present grasp of human cytogenetics, nor is any attempt made at interpreting the findings presented or at fitting them into a general scheme.

The authors deal at length with the diagnosis of mosaicism, but the occurrence of chimerism as a possible though much less common alternative is not mentioned. The paper by Gartler *et al.* is tucked away among the references. Case No. 36/60 (p. 228) might well have been such a chimera in spite of the chromosome analysis not having revealed an XY karyotype. The Academic Press publication on inter-sexuality edited by Overzier is not given a mention even as a reference. This is regrettable, not only because of its relevance to the subject of the Report, but also because of its usefulness as a handbook to those who are active in human cytogenetics.

The shortcomings of this Report are perhaps due to the fact that it was not conceived as a thoroughly scientific dissertation. In the author's own words: "The purpose of the first part of this section is to provide a short account of the morphology of human chromosomes, without attempting a detailed and authoritative description". Who is qualified to give an authoritative account of human chromosomes if not the authors of the Report? Elsewhere we read: "It is already apparent that the study of chromosomes has an important role to play in attempts to elucidate the nature and magnitude of radiation hazards, the causation and evolution of cancers, and the processes of ageing; and a start has been made with the X chromosome on the mapping of genetic loci". This is somewhat misleading. The study of human chromosomes so far has not solved any problems where radiation hazards, cancer and ageing are concerned; it has merely highlighted the features of these syndromes. Neither has the study of human chromosomes directly contributed to the mapping of

genetic loci. One wonders whether this will ever be accomplished by visual means except for polytene- or lampbrush-chromosomes. On the other hand, sex-linked genes in man were known long before the karyotype was established.

Other statements in the report are as yet unacceptable; in their discussion of mosaicism the authors put too much emphasis on the increase of the number of aneuploid cells with increasing age. It must be remembered that in the two papers quoted on this subject the author's conclusions were as follows: Paper (*a*), "The present data are not numerous, and the fact that the differences between the regression coefficients are not statistically significant does not necessarily mean that they are not real"; Paper (*b*), "The results reported here are open to criticism . . . the subjects were comprised of healthy volunteers, hospital in-patients, parents of children with chromosome abnormalities, etc.". In the chapter on testicular feminization (p. 32) it is stated: "The condition is hereditary in nature and is transmitted by normal mothers". However, cases No. 23/60 (p. 211) and No. 24/62 (p. 221) do not support this hypothesis; on the contrary they point to transmission by the father.

On the subject of technique the newcomer to cytology should be warned against adopting the method recommended in the report for the fixing and staining of buccal smears. This method uses 95 per cent. alcohol (one presumes ethanol), as a fixative for sex-chromatin. It is known, however, that even 100 per cent. ethanol does not fix nucleoproteins but leaves them water-soluble; the use of 95 per cent. ethanol in water followed by a watery stain (in this case cresyl-echt-violet) might easily produce undesirable artefacts in some cases. Case No. 180/61 (p. 118) will serve to illustrate this point. Anatomically the subject was a normal male. No drumsticks or sessile appendages were found in 1,000 polymorphs. All the cells analysed for chromosomes were consistent with a normal male karyotype. The only evidence to give support to the presumptive XY/XXY sex chromosome constitution was the finding that the buccal mucosa was chromatin positive on three different occasions with 16 per cent., 29 per cent., 30 per cent. single bodies respectively. We are also told that the first preparation was of poor quality. The conclusion that here we have an example of a Barr-body-like artefact, due to unsuitable fixation, cannot be excluded. There are other similar cases in the report, e.g. Case No. 106/61 (p. 112), where if the evidence from buccal smear is dismissed as artefact, we are left with a normal male child. This child was diagnosed during a nuclear sexing survey of a newborn population. It is impossible to overemphasize the unjust hardship that could be caused through counselling based on unsound methods.

The absence of a cell type *in vitro* is not necessarily unequivocal proof of its absence in the organism. In cases of mosaicism, tissues can vary with regard to their cell populations. In intersexes, gonads should be examined for karyotypes as well as histologically. This report records numerous biopsies taken from gonads for histological investigation, but only a few have been cultured. Histologically Case No. 5/61 (p. 108) was not significantly different from Case No. 1/61 (p. 110). The findings of two cells with 47 chromosomes determined the final analysis as XX/XXY (Case No. 1/61), whereas the lack of two such cells tipped the scales in favour of an XX interpretation (Cases No. 5/61 and No. 68/61). Perhaps a cytogeneticist may urge that in interpreting a patient's congenital symptoms clinical, anatomical and histological data must weigh at least as heavily as chromosomal analysis.

JACOB KAHN.

2. SEX

The Pathology and Treatment of Sexual Deviation. A Methodological Approach. Edited by ISMOND ROSEN. London: Oxford University Press. 1964. Pp. 510. Price 50*s*.

This book originated from a symposium organized by the editor under the auspices of the Portman Clinic. He also succeeded in enlisting several distinguished contributors in addition to those who had taken part in the symposium. The aim was to present a survey of the methods employed in the study and treatment of sexual deviation today. The first two chapters deal with sexual behaviour in animals. N. Tinbergen demonstrates the role of aggression and fear in normal sexual behaviour. In his opinion it is not so much the conclusions of the ethologists which are of interest to the psychopathologist, but their observational methods, especially the so-called motivational analysis. Richard P. Michael follows with a discussion of various biological aspects of sexual behaviour. He observes that Freud's theory of bisexuality has received some confirmation from general biology. Aberrations of sexual behaviour similar to those regarded as deviant in man are represented widely in vertebrates.

The next part of the book is given to the clinical approach. In this context, D. Stafford-Clark discusses the observations of the ethologists from the point of view of a practising psychiatrist. His personal experiences as a therapist in this field are of considerable interest. Peter Scott contributes an excellent treatise