Book Reviews

Chromosome Studies on Adults. (Eugenics Laboratory Memoirs No. 42.) By W. M. Court Brown, Karin E. Buckton, Patricia A. Jacobs, Isbel M. Tough, E. V. Keunssberg, and J. D. E. Knox. (Pp. v + 91; 22 figures and 65 tables. 32s. 6d.) London and New York: Cambridge University Press. 1966.

This is an important and painstaking study of fundamental significance to all those interested in human cytogenetics and its relation to developmental anomaly, malignancy, and ageing.

Attention has been concentrated on the pathological, but this quantitative study attempts to remedy some of the deficiencies of this approach and to determine the frequency of chromosomal variations, the significance of which is as yet obscure, and of rearrangements; and to investigate numerical (and structural) alterations, both dependent and independent of age.

The object is the investigation of these chromosome changes as revealed by the peripheral blood culture technique, in a sample of people in the general population. Some of the changes are a variability in size and appearance of certain easily recognizable (and sometimes identifiable) chromosomes like those of the 13-15 or 21-22 groups, chromosome 16, and the Y. These changes generally are not associated with a harmful effect and thus can be considered 'normal variations rather than aberrations', but it would be inappropriate to conclude that to all such changes there corresponds an exactly identical genetic and chromosomal make-up. Clearly the variation that is inheritable, without detriment, is most likely to be appropriately called normal variation, as it stands the test of transmission with a normal phenotype in different genotypes. The other important chromosomal variable studied is the presence of proliferative chromosome mosaicism, involving presumptive loss of one X chromosome of females and the Y of males, in a proportion of cells and in relation to age. Standardized procedures were used of blood culture, duration of culture, analysis of cells, and interpretation of findings, and, in the case of the structural variation, confirmation by fibroblast cultures was obtained.

A total of 1020 people was studied. There were 438 male and female subjects taken from general medical practice lists, while the rest (582) were miscellaneous collected, with certain safeguards to exclude groups of subjects in whom there was an *a priori* likelihood of chromosome anomaly. In about three-quarters of the cultures, all cells counted were also analysed, and it is

with the findings in these 756 subjects that the first part of the report, on morphological chromosome changes, is concerned. The total prevalence of detectable autosomal variation in both sexes is 2.25% (affecting chromosome No. 16 and those of the 13-15, 17-18, and 21/22 groups), while rearrangements are detected in 0.53%. In addition, among males, variations in length of the Y are found in 2.6%, and over-all changes, including rearrangements, in 2.86%. The variations recorded are small, involve chromosomes with secondary constrictions, are present in all cells, and are generally inheritable without apparent harm, though, as the authors stress, this remains to be proven. The reason for and means by which such variations attain the frequencies that they seem to still remains to be discovered.

The second part of the report deals with chromosome changes detected in cells from blood cultures of the randomly-chosen male and female subjects. The results are classified by age and sex, according to whether the chromosomes are intact, whether they show chromatid changes, or chromosome aberrations. The latter show an increase with age, especially in the male. But the main analysis concerns the relation between aneuploidy and ageing. Taking first the age-dependent changes, and on the grounds of the relatively easily identified Y chromosome findings in male cells, it can be shown that males tend to lose a Y chromosome from their cells which become XO, reaching a maximum of 1.5%. Female cells appear to lose one X but here the maximum could be 6%. In addition, the age when the two sexes begin to show 'true' loss of the relevant chromosome varies, being about 55 in women and 65 in men. The presence of presumptive XO cells in women may lead to difficulty in diagnosing XO/XX developmental mosaicism from blood cultures, and a helpful guide table is given in the Appendix. As for age-independent chromosome losses, which may or may not be the consistent outcome of technical artefacts, the losses involve unevenly the chromosomes, with preferential retention of the larger autosomes (1, 2, 3, 4, and 5), preferential loss of chromosomes 17-18 and, in women, of 21-22.

The report is clearly written and the Appendix contains numerous tables of crude data. The use (and spelling) of 'ideogram' in the captions to most illustrations (but not in the charts and text) is in contrast to the recommendations of the 1960 Denver Conference, which the authors otherwise follow.

PAUL E. POLANI