presentation of the material is excellent throughout, and the picture diagrams are of great benefit to the reader. The book is beautifully illustrated with the emphasis on clinical teaching.

Diseases of waning significance, i.e. vitamin D deficiency rickets, acroacia, and other vitamin deficiencies have been reduced in length, but the size of the book has not altered; though it is perhaps a formidable size for undergraduates, it is nevertheless an extremely valuable teaching and reference book for paediatricians and practitioners alike. It should hold a high place in international paediatrics.

Information is readily obtained by virtue of a detailed index covering more than 80 pages.

A revised English edition is eagerly awaited.


The main and original part of this investigation deals with a cytogenetic and clinical study of 119 subjects with Down's syndrome or suspected Down's syndrome, referred to the Institute for Medical Genetics at Uppsala since 1959. Of these persons, 57 were referred because the diagnosis was in doubt, 18 for counselling because of young maternal age (30 years and less), 9 because of a positive family history, and 35, all born to women over 30, for other reasons.

In 18 of the 57 patients whose original diagnosis was in doubt, a diagnosis of Down's syndrome could not be confirmed clinically when they were reviewed and, of these, 16 had a normal chromosome make-up, one had a chromosome anomaly difficult to relate to Down's syndrome (Case 6), and one was a chromosome mosaic of normal and 21-trisomic cells. In the other 39, in whom a confident clinical diagnosis of Down's syndrome was made, the cytological findings confirmed the clinical assessment.

There were 5 patients with chromosome translocations, in one between a presumptive 21 and a member of group 13-15 (D/G), and in 4 between a presumptive 21 and another member of the 21-22 group (G/G translocation). Excluding familial cases there were 33 infants of younger mothers (30 years and younger at the birth of the affected children) four of whom carried translocations, and 60 children of older mothers only one of whom carried a translocation. None of the translocations was apparently inherited, though one G/G translocation the father could not be studied chromosomally. A study of the five families with two Down's syndromes in the sibship, and of one family with two in the kinship, showed that the index cases were trisomic for chromosome 21 (one was a mosaic), that the three secondary cases that were examined were also trisomic 21, and that the parents were chromosomally normal.

Of the 119 patients in this survey the author personally examined 111, and a part of the report is devoted to the analysis and discussion of the clinical aspects of Down's syndrome.

The clinical and cytological features of the cases studied are summarized and tabulated in the Appendix.


The main object of this report is to make available to those interested in human genetics and cytogenetics, particularly, data collected since 1959 in connexion with the Registry of Abnormal Human Karyotypes set up in the Medical Research Council's Effects of Radiation Research Unit in Edinburgh.

The first two parts are introductory and deal with techniques and with the methods of detection of human sex-chromosome anomalies, and summarize briefly the abnormalities of sex chromosomes that have been described in the literature and that are relevant to the third part of the report. This forms its bulk and gives in detail the cytological, clinical history, and other findings in 266 individuals with sex chromosome abnormalities and discrepancies, reported to the Registry and studied from 1959 to the end of 1962.

Most of the subjects are adults, but there are some children, either specially referred, for instance for investigation of failure to grow satisfactorily, or discovered because of neonatal signs (e.g. congenital lymphoedema and webbing of neck) and in the course of routine nuclear sexing of newborn babies. Of the 266 persons, 134 have a male phenotype (99 are XXY and 24 have sex chromosome mosaicism), 128 are of feminine appearance (38 are XO, 33 are XXX, 22 are sex chromosome mosaics, and 26, among whom 20 are thought to be examples of testicular feminization, have an XY complement), and there are also 5 true hermaphrodites. The information in each case is clearly set out and the authors are to be congratulated on making their carefully collected data available to those interested in this rapidly expanding field.