Some chapters, however, seem only loosely connected with antenatal diagnosis (eg, screening for inherited traits, detection of heterozygotes). There is also considerable overlap between chapters and the lack of an index makes it difficult to find information. The last 2 chapters on the legal (Sadler) and eugenic (Neel) implications of antenatal diagnosis are very interesting, but no one but a lawyer would be able to make sense of the references at the end of the legal chapter.

This book is a readable and inexpensive account of work being carried out by some of the best known investigators in antenatal diagnosis and related fields. It is unfortunate, however, that there is a certain lack of cohesion and uniformity in the various presentations.

ALAN EMERY


This is the proceedings of the First Inter-American Symposium on Hemoglobins held in Caracas in December 1969. The meeting brought together workers in many different disciplines who had in common an interest in the haemoglobin molecule.

The book falls into 3 sections on the geographic distribution and genetic problems of haemoglobins, the structure and function of haemoglobins, and physical studies of the haemoglobin molecule. The first section deals primarily with the occurrence and incidence of abnormal haemoglobins and of thalassaemia, and presents information from Mexico, Jamaica, Brazil, El Salvador, Colombia, and Venezuela. These form a valuable statement on the extent of haemoglobinopathies in this part of the world, much of which was not previously available in the English language. Two observations of especial interest were the rarity of abnormal haemoglobins among the indigenous Indian populations of Mexico, Colombia, and Venezuela, and the surprisingly high frequency (13.6% in Colombia) in certain selected populations.

The middle section describes studies on the function of normal haemoglobin including the properties of haemoglobin subunits, the effect of pyridoxaline and of 2,3-DPG on haemoglobin function, the oxygen binding sites of haemoglobin, and certain functional aspects of ligand binding. The ways in which the abnormal functional properties of certain haemoglobin variants add to our knowledge of haemoglobin function are illustrated in studies on the M haemoglobins and on haemoglobin Gun Hill.

The last section deals with the physical properties of the haemoglobin molecule and of some variants investigated by procedures which included electron paramagnetic resonance (EPR), electron spin resonance (ESR), nuclear magnetic resonance (NMR), circular dichroism (CD), and far infra-red spectroscopy. The proceedings are well printed and illustrated and form a useful review of the knowledge and investigative approaches to the structure and function of the haemoglobin molecule. It can be recommended to all workers in this field.

GRAND SERJEANT


The phenotypical effects of trisomy G/normal mosaicism are of great interest in the phenomenon of cytogenetics. As well, they are of practical diagnostic importance to the clinician. It is possible that they may also be of significance to the clinician from a predictive point of view as regards development, although there has been considerable variation of opinion with respect to possible correlation between intelligence and the percentage of abnormal cells. Moreover, there is some evidence that a shift of proportions of cell lines may take place after the age of between 1 and 2 years, when, it has been said, as a rule equilibrium is reached. This poses the interesting question as to whether or not such fluctuations influence the course of development.

The study of trisomy G/normal mosaicism from both a cytogenetical and a clinical point of view has been hampered not so much by the relative paucity of material as by the means by which the subjects are ascertained. Cases that have been investigated have for the most part come to light by virtue of abnormality in themselves or in their offspring. It is quite possible that the material that has become available in this way accounts for only part of a much wider spectrum of clinical variation spreading into the normal range. The patients in the present study are no exception to the general rule of ascertainment through abnormality, though they were not necessarily discovered because a diagnosis of mongolism was suspected. Five of the 10 trisomy G/normal mosaic patients in this study were detected during karyotyping of 38 mentally retarded patients with multiple congenital anomalies and retarded growth; 3 were discovered during investigation on the basis of a dubious diagnosis of mongolism, and the chromosomes of 2 had been studied because they were the first mongoloid child of a young mother.

The group of 10 trisomy G/normal mosaic patients were studied together with a group of trisomy G patients and a group of normal controls. Clinical parameters studied included body height, cranial circumference, upper/lower body segment ratio, and 10 physical signs of diagnostic value in mongolism. Special investigations were made of dermatoglyphs and the radiology of pelvic bones. Maternal ages and birth weights were compared. Histochemical and biochemical assay of alkaline phosphatase activity of leucocytes was carried out, also fine-structural studies of the granules of the neutrophil granulocytes. Unfortunately by virtue of their means of
selection the patients in this study were mentally retarded and were not considered to be sufficiently representative sample for psychometric studies, and the interesting question of intelligence and possible clinical and cytogenetical correlates could not be pursued.

The chromosome studies of blood and skin showed a varying ratio of trisomic cells in both. The degree of clinical similarity between the mosaic patients and the trisomic patients was discussed, and the importance of selection of clinical signs according to age was stressed, especially with respect to neonates. The presence of more than three of the 10 selected cardinal signs was considered highly suggestive of either trisomy G/normal mosaicism or mongolism. A dermatoglyphic difference was shown between patients with more than 30% trisomic cells in either skin or blood, and those with a smaller percentage, the former group having positive dermatoglyphic log indices (Ford Walker) and the latter negative values. In both mosaic and trisomic patients the mean alkaline phosphatase levels of leucocytes were higher than in the controls, but no significant difference between the 2 groups (mosaic and trisomic) was found; an intermediate value between mosaic and trisomic subjects was found however with respect to the mean lobe count. Overall, the results supported the view that the extra autosomal G chromosome in the mosaic subjects in this study was the same as that found in mongolism.

This is an unpretentious study, methodically planned, and carried out with thoroughness. It is a solid contribution to the body of knowledge on the subject. In the monograph, the literature is reviewed comprehensively, and it may be regarded as a compact small book of reference, useful both to the cytogeneticist and to the clinician.

VALERIE COWIE


This is the second study in the series begun by the publication of 'A Psychiatric-Psychological Study of 50 Severely Hypogonadal Male Patients, including 34 with Klinefelter's Syndrome, 47,XXY' by Johannes Nielsen and the same colleagues (Copenhagen: Munksgaard, 1969). That volume was not reviewed in this Journal. Part of the motivation for these studies was to find out whether 47,XXY individuals derived their characteristic psychiatric and psychological traits directly from their genetic constitution or through the mediation of their hypogonadality. Hence the comparison of Klinefelter cases with a control series of 16 hypogonadal 46,XY males of the same mean age. The results have been to show that while the XXY men did not differ from the XY men in respect of father's occupation, childhood milieu, size of sibship, and parental age, they did differ significantly in having much more in the way of psychiatric symptoms, and on the somatic side having smaller testes and more frequently a gynecomastia.

This second volume gives more detail about the psychiatric scoring, and reports more mental illness and more criminality in the XXY men. The results of examination by a multiplicity of psychological tests (WAIS, Memory, Draw-a-person, Rorschach, Word Association, TAT) are presented. The XXY men are reported as less intelligent, more psycho-infantile, and generally psychologically disadvantaged to a greater extent than the XY men in respect of many psychological traits. A good deal of the material of the first volume is repeated in this one, including a set of case histories, here abbreviated; this is a sensible step, since psychologists, to whom this volume is of particular interest, can study it without reference to the first. Medical men and human geneticists will concern themselves with the first volume, and will have less interest in this. Other studies are to follow the same team on the same series, including a report on anthropological findings.

The main importance of this series of Klinefelter XXY cases resides in the fact that they were ascertained at subfertility clinics, and were therefore not subject to a selective bias in favour of mental subnormality, mental illness, psychopathy or criminality. However, the comparison series are not strictly comparable, and it is doubtful how far deductions can be safely based on the features in which the two series differ. The work done on both series of subjects is clearly very painstaking, and should be reliable.

ELIOT SLATER


The increasing number of rare inborn errors of metabolism, many with ocular abnormalities, presents the ophthalmologist with the formidable task of searching the literature in order to keep abreast of advances in this difficult but important branch of medicine. The author has presented, in a simple manner, the ocular and systemic manifestations of these metabolic abnormalities and has included notes on their pathology, biochemical defect, genetics and treatment, as well as a few recent key references. The disorders are presented according to the ocular structure primarily involved, thus making this small volume of particular value to ophthalmologists but also of interest to other specialists.

BARRIE JAY


Six hundred references are included and grouped under 4 headings: an author index, a subject index under 37 different headings, reference books, and journals on human genetics. Titles and basic bibliographical details are given in the language of publication. The introduction, the headings, and other bibliographical details are in English, French, and Dutch.