Book Reviews

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 special 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 pyridoxal 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.

Graham Serjeant


The phenotypical effects of trisomy G/normal mosaicism are of great interest in the phenomenology 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