Book Reviews


The first papers on anaemias due to recognizable enzymic defects in red cells deal with many aspects in an encyclopaedic manner, but the article by W. S. Schroter on the neonatal and paediatric aspects is very helpful and readable and the general discussion valuable.

The next section deals with lysosomes and the opening paper by A. C. Allison shows how far recent research has gone since the lysosome was identified in 1955. The subsequent papers go still further in showing how important the lysosomes are, and how the absence of certain specific enzymes in the collection of lytic enzymes in the lysosome is the cause of most of the so-called storage diseases. The next five papers mainly deal with specific examples of storage diseases, while one on Leigh's encephalopathy attempts to deal with a specific enzyme defect of gluconeogenesis. Six papers on disorders of the urea cycle report various enzymic errors, many of which cause hyperammonaemia, and A. Russell suggests they can cause cyclic vomiting and migraine. Finally, eight unconnected papers describe recent work on various disorders, including a study of diurnal variation in the serum phenylalanine in phenylketonuria, in which the normal pattern is reversed; a new error—saccharopinuria; selective malabsorption of magnesium causing tetany in the neonate; and two papers on histidinaemia.

This is a rapidly growing field and the papers given were full of unreported observations, many of which point the way to further study.

To anyone interested in inborn errors of metabolism this is an important book. It is well produced, of convenient size, and is easily read.


This book presents the proceedings of an international conference held in November 1967, and it must suffer by being nearly 2 years out of date. The conference dealt with the whole picture of galactosaemia—the clinical pattern, the enzymic defects, the genetic factors, and results of treatment. It is inevitable in a series of individual papers that there is much repetition especially on the technical aspects, but this in some cases is of benefit as different results and conclusions are presented and clarified in the discussions which follow groups of papers. For the more clinically minded the opening paper by G. Komrower sets the scene and puts the reader au fait with the main problems. This is followed by 9 mainly technical studies of the way the enzymic error affects the body metabolism and on methods of assessing the enzymic defect. A clinical paper on the effects of treatment on the IQ raises doubts of the value of diet in preventing mental retardation, and a further group of more technical papers follows. In these the detection of the heterozygotic carrier of the defective gene is dealt with and again the methods to be used are well discussed. Some very interesting studies of in vitro cell cultures are reported—the enzymic defect being perpetuated in the cultures. Two variants are reported—the Duarte and the Negro. In both there is a less severe loss of enzyme, and in the Negro it only shows clinically in infancy: Duarte variation is only of importance in genetic studies.

An excellent paper by Donnell and co-workers studies the results of treatment, and again doubt is expressed of the value of strict diet after the infant has survived the first 24 months.

The final papers present studies on the technique and results of various screening tests.

In general the book is a good but somewhat lengthy study of galactosaemia, which would repay reading by anyone wishing to know all about the condition.