Inborn Errors of Metabolism

Edited by Roland Ellis. (Pp 105; figures + tables. £8.95 hardback, £3.50 paperback.) London: Croom Helm. 1980.

This book covers a few topics to illustrate in straightforward language the current state of our understanding of inborn errors of metabolism. It is intended for physicians who wish to keep in touch with this rapidly expanding subject, and will be useful to newcomers who wish to familiarise themselves with the subject.

The genetic basis, written by the Editor, covers the essential aspects in a clear and easily readable style. The section of disorders of lipid metabolism, also by Dr Ellis, summarises relevant enzyme assays, storage substances, and clinical features in some of the sphingolipidoses. An excellent chapter (P Whiteman) on the mucopolysaccharidoses includes clear accounts of the salient clinical and biochemical aspects of these disorders. The speed at which this subject is developing is illustrated by the fact that a form of mucopolysaccharidosis due to glucosamine 6-sulphate sulphohydrolase, tentatively designated by the authors as MPS ?VIII, has since been shown to be a fourth type of Sanfilippo disease (MPS IIID).

A chapter on disorders of amino-acid metabolism (J Seakins) is highly selective, omitting the majority of known diseases, but choosing as models phenylketonuria and disorders of the transsulphuration pathway.

More detailed are the chapters on purine and pyrimidine metabolism (A Harkness) and on disorders of carbohydrate metabolism (I Menzies).

Brief but clear accounts of disorders of trace metals (T Delves) include descriptions of Menkes and Wilson's diseases and acrodermatitis enteropathica.

Finally, there is a masterly overview on possible future developments and research by Professor Cedric Carter, who considers that there are good reasons for screening for heterozygous carriers in the general population, so that when at risk for giving birth to children with metabolic disorders they can be counselled with a view either to avoiding marriage or to providing prenatal diagnosis. Such a programme would be particularly relevant to subjects with European ancestry when tests for carriers of the cystic fibrosis gene are available.

This book does not deal with the subject either comprehensively or in depth. It is suitable for readers wishing to acquire easily an understanding of basic principles of inborn errors of metabolism.

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Genetics of Gastrointestinal Disease

Progress in Medical Genetics vol IV. Edited by A G Steinberg, A G Bearn, and A G Motulsky. (Pp viii + 255; figures + tables. £23.00.) Philadelphia, London, Toronto: Saunders. 1980.

The main theme of this volume is genetic heterogeneity. Until recently much of the progress in splitting diagnostic entities into two or more genetic types has been in biochemical disorders in which different single enzyme defects can result in similar phenotypes. Some of the six authoritative chapters describe work of this type, but in others research on more complex disorders is reviewed.

Peptic ulcer genetics is reviewed by Dr Jerome Rotter. The division of gastric from duodenal ulcer was made long ago but the discovery of the genetic basis of hyperpepsinogaemia I 5 years ago has led to the demonstration of heterogeneity within duodenal ulcer. The 50% of duodenal ulcer patients who have this dominantly inherited character also have hyperchlorhydria but, as yet, no other clinical feature has been found distinguishing them from the normopepsinogaemia I patients. So far the main value of the demonstration of these two genetic types of duodenal ulcer is that in the hyperpepsinogaemic type the genetic predisposition to ulcer in apparently normal sibs can be identified by a raised fasting serum pepsinogen I level.

Familial polyposis coli is reviewed by Edmond Murphy and Ann Krush. Gardner's syndrome has for a long time been considered to be genetically distinct from polyposis without extracolonic manifestations but recent work has cast doubt on their complete genetic separation. Having applied considerable mathematical formulations the authors are not able to draw any firm conclusion about their genetic relationship. There is much useful clinical advice and I found it a useful refresher course on methodology in epidemiology and genetic analysis.

Hereditary hyperbilirubinaemias are described by Gerrard Odell and Barton Childs. They consider the mechanisms of bilirubin conjugation and secretion, and identify the numerous enzymes involved and how a defect of any one of them can result in jaundice. Already three different defects have been shown to result in Gilbert's syndrome, a partial explanation of its high prevalence.

Haemochromatosis and progress in explaining its genetics are reported by Marcel Simon and colleagues who have used refined studies of iron absorption and HLA typing in genetic linkage analysis. They decide in favour of recessive inheritance without much consideration of a polygenic hypothesis. To someone new to HLA, the most useful part of this chapter