Book reviews

Journal of Medical Genetics, 1978, **15**, 408

Biochemical Methods in Medical Genetics

By Sally Kelly. (Pp. xii + 346; 14 Figures + 13 Tables. $17·50). Springfield, Illinois: Charles C. Thomas. 1977.

This is a remarkable collection of analytical procedures, described in practical detail, necessary for the diagnosis of inherited metabolic disease. The book is well produced, with colour plates where these are helpful, and is very reasonably priced. It would be tedious to count the tests described (sometimes there are alternatives for a given disorder) or the diagnoses to which they would lead. Suffice it to say that the list is as comprehensive as a single author could make it and quite up to date. Most geneticists, and those in laboratories serving them, will wish to have this book at their elbow.

The work has both its strengths and its weaknesses and these are illustrated by the treatment of glycogen storage disease. This section contains a Table of clinical features of types 1 to 7 and 9, with types 2 and 6 subdivided. Analytical procedures are given for liver glucose phosphatase, glucosidase in urine, liver, and leucocytes, brancher and debrancher enzymes, and muscle and leucocyte phosphorylases. However, there are no tests to show whether the patient might have one of the glycogen disorders or not: no glucagon before and after a carbohydrate feed, no ischaemic exercise test for type 5, and no real recognition that a an apparent liver phosphorylase deficiency might be a deficiency of phosphorylase, phosphorylase kinase, or a more subtle deficiency involving cyclic AMP. Determination of red cell glycogen is another simple and useful discriminant that is not described. Under the heading of the mucopolysaccharidoses, the toluidine blue spot test is preferred to the alcian blue test which has been considered for the past 15 years to be more specific. The cetylpyridinium chloride turbidity test, abandoned by many because of the high proportion of false-positives, is described without warning, as is the notoriously unreliable metachromatic staining of urinary sediment. Similarly, after describing the assay of hexosaminidase A in cultured amniotic cells, the same assay is described for uncultured cells for prenatal demonstration of Tay-Sachs disease. True, the warning is given that for enzyme activities measured in uncultured cells the 'values are lower and less reliable than those for cultured cells', but it is stated quite definitely that between gestational ages 16 to 25 weeks, hexosaminidase A is 23 to 40% of the total hexosaminidase activity in normal homozygotes; heterozygotes between 20 and 28 weeks have 22 to 27% hexosaminidase A, whereas abnormal homozygotes have less than 6%. For a subject in which even the best methods have built-in uncertainty, one feels that in offering such a 'less reliable' test, the object of this particular exercise has been obscured.

For the discriminating and already well informed this book is valuable and will save much literature searching, but the lack of more critical guidance could lead the less experienced reader into deeply troubled waters.

D. N. RAINE

Errata

In the February 1978 issue of *Journal of Medical Genetics*, an error appeared on page 68 in the article 'Waardenburg-like features with cataracts, small head size, joint abnormalities, hypogonadism, and osteosarcoma'. The first line of the second paragraph of the Discussion 'Premature greying, cataracts, and hypogonadism . . .' should read 'Prematurely grey hair, medially flaring eyebrows, . . .'.

On page 34 of the same issue, in Dr Naomi Fitch's address for reprint requests and at the head of the paper, 3755 Chemin Cote Street, Catherine Road, should read 3755 Cote St. Catherine Road.

On page 111 of the April 1978 issue, in the article by Dr Bonaïti on 'Genetic counselling of consanguineous families', the numbers in the Table under the heading $F = 1/16$ should read downwards: 4·2, 13·8, 33·9, 5·9, 1·0, 0·3, 2·1, 1·6.