Book Reviews


This volume comprises the Proceedings of the Ninth Symposium of the Society for the Study of Inborn Errors of Metabolism held in Leeds in the summer of 1972. Twenty papers are presented with introductory remarks, synopses of experimental techniques, and results in tables and clear figures. Most have a brief summary and all have valuable references and group discussion.

Just as paper chromatography led to the discovery of individual amino-acidurias and metabolic disorders, so gas chromatography and mass spectrophotography elucidate organic acidurias and a new family of inborn errors. Similarly, these phenomena are produced by enzyme deficiencies as demonstrated in ketotic hyperglycaemia and propionic acidemia (propionic CoA carboxylase), β-OH isovaleric acidemia and p-methyl crotonyl glycinuria (p-methyl crotonyl CoA carboxylase), Leigh’s encephalomyelopathy (pyruvate carboxylase), methylmalonic aciduria (leucocyte methyl malonyl CoA mutase), and in branched chain acidurias. Whether tyrosinosis is caused by lack of inhibition of p-OH phenylalanine hydroxylase (seemingly a Cu/Fe enzyme inhibited by Hg²⁺ but not by other –SH reagents) or the cirrhosis, leading to lowered prothrombin ratios and bleeding disorders (in contrast to tyrosinaemia) by peroxide formation; is still debated.

Thermodynamic considerations in calcium oxalate precipitation suggest that calcium and oxalate are probably separate or stabilized in the normal renal papilla whilst in hyperoxaluria, now divided into Type I (glycolic aciduria with glyoxylate: 2-keto-glutarate-carboxi- lase deficiency) and Type II (glycric aciduria with glycric acid dehydrogenase deficiency) magnesium still seems the best therapy.

Deuterated phenylalanine confirms the lack of alternative paths from phenylalanine to tyrosine in phenylketonuria (PKU) and hyperphenylalaninaemia (HPE) and the excess mandelic acid seems to come from the phenylalanine whereas the hippurate probably comes from intestinal benzoate. Why the plasma phenylalanine and the response to a load should be lower in HPE is not yet clear. Other investigators notice that changes induced in the kynurenine and indole paths by a tryptophan load is related to resting levels of more or less than 20 mg%. Are these further differences between PKU and HPE?

There is description of the elegant linking of GLC to mass spectrography and computer data storage in the characterization of body fluid abnormalities and silica thin layer chromatography of keto acid hydrazones demonstrating differences between adult and juvenile cystinosis.

Other works include the four childhood lactic acidoses contrasted to the more benign adult form, benzoate excretion in psychosis, and the independence of pyroglutamate to N metabolism and its possible implication in amino-acid transport. Treatment is mentioned when noteworthy, eg, dietary leucine restriction in β-methyl crotonyl glycinuria and thiamine in Leigh’s encephalomyelopathy.

It is fascinating to see modern biochemistry and biochemical techniques unravel the complexities and subtleties of metabolic disease and here, there is much stimulus for similar applications in other directions. Although clinical indices for suspicion (eg, persistent acidosis in a hydrated child with glycine/alanine ratio 1) are mentioned, the bias is essentially biochemical with limited clinical description and brief mention of genetic mechanisms. Perhaps the volume will have limited use in general post- or undergraduate medicine but it will be of interest especially to the academic clinician and considerable value for the researcher. The presentation is clear although the chromatograms might have been better displayed with keys rather than descriptive text. Overall this is a most interesting and valuable book of high quality in a new branch of medicine.

Christopher Owens


The appearance of this book will be very welcome to radiologists, paediatricians, and medical geneticists. The delineation of the bone dysplasias has much improved over the past 10 years. The literature is widely scattered over paediatric and radiological journals and it is invaluable to have it summarized in this book by contributors, about equally divided between radiologists and paediatricians, who include many of those who contributed to the new developments. While many of the disorders are single gene determined, only a minority are as yet defined in terms of biochemical defect, their histology is often non-specific, and the radiological appearances are still in most instances the most reliable methods of diagnosis. However, Rimoin and colleagues report new studies in this volume showing that the histological appearances in achondroplasia and hypochondro-
plasia, in which both resting cartilage and enchondral ossification are relatively normal, are distinguished from those in thanatophoric dwarfism and metaphyseal dysostosis, in which resting cartilage is normal and enchondral ossification is disrupted, and from those in achondrogenesis and metatropic dwarfism type II, in which resting cartilage is abnormal and the defect in enchondral ossification is apparently secondary to the defect in epiphyseal cartilage.

In the chapters on specific disorders the main emphasis is on the radiological appearances, including the changes with age and the radiological differential diagnosis. The radiographs are of excellent quality. The clinical features, including course and prognosis, are also described briefly. The genetics of each condition are also didactically given. Full references are listed for the radiological appearance and one or two key references for the clinical aspects.

C. O. CARTER


This book opens with a quotation by C. D. Darlington: ‘...the difference between the two sexes, man and woman, is one of the most fundamental facts of life, physiologically, and socially, intellectually, and historically. It is a totally genetic one...’ Nevertheless, only two chapters of this book are written by geneticists while the other nine are contributed by 11 psychologists and psychiatrists.

The first chapter, by C. O. Carter deals with sex linkage and sex limitation and thus hardly touches on the problem of sex differentiation. This is followed by a more exhaustive treatment of ‘Errors of Sex Determination and Sex Chromosome Anomalies’ by P. E. Polani, which makes it clear that the human Y chromosome is strongly male determining.

The other chapters seem to be centred round the mind, the body receiving only secondary treatment. Although there is a section on ‘Embryological Development and Sexual Differentiation’, by Corinne Hutt, which deals with many pertinent problems concerning the physical basis of sex differentiation, the treatment is altogether too general. For example, it is not sufficient to state that, following the administration of exogenous androgens ‘the female is masculinized to the extent that male genitalia develop’ (p. 76); a more precise description of the degree of masculinization achieved in these circumstances is needed.

Some of the biological examples have been chosen unwisely. Thus, although sex chromatin studies on spontaneously aborted fetuses seemed at one time to suggest that these contained a marked excess of males, subsequent results of chromosome studies have cast serious doubts on the accuracy of the earlier reports. Again, a mass of data on environmental factors reputed to be affecting the sex ratio at birth are bedevilled by the fact that if there is any effect at all, this is very small, so that problems of sampling loom large. On the other hand, the development of a male newborn ensues in the vast majority of surviving zygotes containing a Y chromosome, and conversely, a zygote lacking a Y chromosome will develop into a female. The mechanism linking cause and effect is unlikely to be unravelled by studies of sex ratios, whatever their merits.

Birth weights, as studied by Margaret Ounsted, are clearly far more relevant, even though the weight at birth can no longer be accepted as a direct effect of the chromosome constitution. In the concluding chapter, Christopher Ounsted and David Taylor propose that sex differentiation may be due to differences in developmental rates. The authors are of the opinion that transcription in males proceeds at a slower pace than in females, notwithstanding the fact that male fetuses grow faster than female ones.

The topics raised in this book are of great general interest and also very complicated. In order to satisfy geneticists, however, they require an altogether more rigorous treatment than the one which has been adopted.

Ursula MITTWOCH


The first edition of this dictionary appeared in 1968 and was reviewed in this Journal in 1969 (6, 230). This second edition contains 700 new entries and the appendices, which include an historical survey and a list of relevant periodicals, are updated to 1971.