testicular atrophy, and the affected females had a high incidence of obstetric abnormalities. This family also shows that obstetric abnormalities (other than hydramnios) are the result of the maternal and not the fetal genotype, and that these abnormalities are not responsible for the features of congenital myotonic dystrophy.

Dr Harper was well chosen to write this monograph which can be highly recommended for its very thorough cover of a most curious disease. An extra bonus is that the style is lucid and easy to read. My own feeling is that when the conundrum of congenital myotonic dystrophy is solved the following editions of the book will be even more fascinating than the first. In the meantime, this first edition should be available to all paediatricians and neurologists and hopefully will stimulate further research.

SARAH BUNDEY

Advances in Human Genetics

Volume 9. Edited by H Harris and K Hirschhorn. (Pp xiv + 379; figures + tables. \$35.00.) New York, London: Plenum Press. 1979.

Advances in Human Genetics, now in volume 9, is the most important annual publication in human genetics. This year Opitz, Herrmann, Adinolfi, Harnden, *et al* have contributed reviews of α -fetoprotein, chromosomes and neoplasia, malformations in man, aspects of the molecular biology of haemoglobin, and the metabolism of folate.

In an informative chapter on malformation in man Opitz and his colleagues discuss terminology. They denigrate, with justification, the clinician's ignorance and lack of epistemological skill in assigning birth defects to their natural biological subgroups. They reject with emphasis "the deplorable and redundant term anomalad . . . on linguistic, biological and historical grounds", but whether their preferred "developmental field complex" will fare any better is uncertain. Their concern for correct usage of terms is without doubt justified, but the chapter will inhibit most people from describing any new syndrome and perhaps that is as it should be; there are too many old syndromes masquerading under a new name.

In the review of α -fetoprotein, Adinolfi comprehensively covers the physicochemical properties of the substance, as well as its clinical usefulness in the detection of neoplasia and neural tube defects. He lists other conditions where raised levels have been found and although uncomplicated hydrocephalus heads the list, the significance of raised

levels in this latter condition must be doubted. It is traditional to review one metabolic condition, and this year it is folic acid. For the clinician, the main interest is the emphasis the author places on defects of folate metabolism as a cause of neurodegenerative disease in the absence of severe anaemia.

The relationship between chromosomal anomalies and neoplasia is intriguing. There are those genetic diseases which predispose to the development of neoplasia and these are discussed by Harnden, but it is the genetic change within the neoplastic cell that is his main concern. Finally, the chapter on the expression of human haemoglobin loci is an update which substantiates the many claims that haemoglobin is in advance of any other substance in its contribution to the understanding of molecular genetics. Volume 9 is highly recommended.

M BARAITSER

Inherited Disorders of Carbohydrate Metabolism

Edited by D Burman, J B Holton, and C A Pennock. (Pp xiv + 433; figures + tables. £15.95.) Lancaster: MTP Press. 1979.

This monograph is a report of the proceedings of the annual meeting of the Society for the Study of Inborn Errors of Metabolism (SSIEM) in Bristol in July 1978, and the contributors form a most distinguished international cast. The membership of the society is open to workers in many disciplines, including biochemists and clinicians, and the papers reflect this heterogeneity.

Professor Hers in the Milner lecture gives a masterly account of the control of carbohydrate metabolism in the liver to which he has contributed so much.

Galactosaemia is reviewed in detail. The biochemistry, clinical aspects, problems of pregnancy, prenatal diagnosis, and screening are all covered. However the discussion failed to resolve why patients in Los Angeles have a mean IQ of 95 whereas those treated in the UK have a mean IQ that is approximately 30 points lower even when it is known that dietary control has been 'satisfactory'.

Fructosaemia has a frequency of about 1 in 20 000 in Switzerland but I doubt that the diagnosis is made with frequency in the UK. Similarly, patients with fructose 1-6 diphosphatase deficiency probably remain undiagnosed and the chapters on fructose metabolism are a useful summary of both clinical and biochemical aspects of the disorders.

Congenital lactic acidosis remains one of the most