INBORN ERRORS OF METABOLISM

By D. Y. HSIA

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It is questionable whether the title of a classical work should be used afresh. In this case there is no question of fraud; nevertheless the synonymy may perplex students of the history of science in future centuries. Garrod's book was profoundly original, and, when written, difficult to follow. We can now see that it contained the germs of advances in several branches of science.

The book before us is quite different. It shows no signs of originality, but is easily read, and incomparably the completest book available on the subject. It will be indispensable for all students of human genetics, and desirable reading for animal, plant, and microbial geneticists. Nowhere else is all the information summarized in it available in one volume, or even in several.

But the book is open to severe criticism. No attempt is made to explain the special methods of human genetics, for example those used in testing whether the numbers autosomal recessives agree with Mendelian expectation, though numbering over a quarter of families including at least one recessive. Whereas 56 pages are devoted to analytical methods. European and Asian work is systematically neglected. For example the monumental research of Montalenti and his colleagues on thalassemia (microcitemia) is completely ignored, with the statement that "the incidence is probably higher in Mediterranean countries". The account of sickle cell anaemia is incomplete. Ingram's work is ignored on p. 30, though described on p. 42. The account of its adaptive value on p. 40 is misleading. It is only claimed that haemoglobin S confers resistance against malignant tertian malaria, due to *Plasmodium falciparum*. No effect on other kinds of malaria is claimed. It is therefore not at all surprising "that malaria occurs in certain regions where sickle cell disease is unknown". Similarly the pedigree on p. 89 contains at least three mistakes, apart from the omission of most of the normal brothers and sisters of the haemophilics. As I compiled the pedigree in question with some effort, I could wish that it might be copied accurately, even if my name is not mentioned.

The choice of metabolic "errors" may be thought rather arbitrary. Such rare conditions as xyloketosuria are included, while gamma-amino-isobutyric aciduria is not. Again cystinosis is very doubtfully included under "disturbances in renal transport mechanisms," since cystine is deposited in the liver, marrow, and eyes. Nor is it obvious why myotonia dystrophia is described since the pathogenesis "is completely unknown at the present time". Misprints are not rare, and the price seems very high.

Nevertheless the book is unique and there is no substitute for it. I hope that in future editions some of the errors mentioned may be corrected. If a number of diagrams which convey little information (for example the "pedigrees of causes" on pp. 92, 93 and 95) were omitted, the price could be reduced.

(J. B. S. HALDANE)

Inborn errors of metabolism are a group of inherited disorders characterized by enzyme defects. Clinical manifestations are usually due to the accumulation of toxic substances in the body. While in many cases the disorder cannot be cured, disease outcomes and life expectancy can be improved with supportive care and the appropriate diet. Alpha-1 antitrypsin deficiency (AAT deficiency). Definition: a congenital disorder characterized by the accumulation of defective alpha-1 antitrypsin enzyme. Epidemiology: more common in individuals of European descent [1]. Etiology: mutations in SERPINA1 gene Inborn errors of metabolism (IEMs) are a large group of rare genetic diseases that generally result from a defect in an enzyme or transport protein which results in a block in a metabolic pathway. Effects are due to toxic accumulations of substrates before the block, intermediates from alternative metabolic pathways, defects in energy production and use caused by a deficiency of products beyond the block, or a combination of these metabolic deviations. Inborn errors of metabolism (IEM) are a group of inherited metabolic disorders leading to enzymatic defects in the human metabolism. As its name implies, inborn errors means birth defects in newborn infants which passed down from family and affecting metabolism. Hence, it is called Inborn errors of metabolism or inherited metabolic disorders. IEM can appear at birth or later in life such as phenylketonuria, albinism, lactose intolerance, gaucher disease, fabry disease etc. IEM refers a condition where in bodyâ€™s metabolism is affected due to genetic disorders. The cause of IEM is mutations in a Many of the inborn errors of metabolism, including urea cycle defects, organic acidemias, and certain disorders of amino acid metabolism, present in the young infant with symptoms of an acute or chronic metabolic encephalopathy. Typical symptoms include lethargy, poor feeding, apnea or tachypnea, and recurrent vomiting.