## THREE MEDICAL GENETICS BOOKS

AN INTRODUCTION TO MEDICAL GENETICS. 3rd edition. By J. A. Fraser Roberts. Oxford University Press, London. 1963. Pp. xiii+283. Price 35s.

MEDICAL GENETICS. An Introduction to Medical Genetics for Physicians and Medical Students. By Widukind Lenz. University of Chicago Press, Ill., U.S.A. 1963. Pp. vi+218. Price $6.50.

THE PORPHYRIAS. A Story of Inheritance and Environment. By Geoffrey Dean. Pitman Medical Publishing Co. Ltd., London. 1963. Pp. xi+118. Price 25s.

A third edition, an English translation and a scientific detective story are the latest demonstration of the continuing interest of the medical profession in genetics. Why this interest began rather suddenly is speculative. Perhaps it was because medical men realised that the subject was within their grasp and did not necessarily involve a knowledge of higher mathematics, and it is to writers such as Fraser Roberts and Lenz, with a foot in both camps, that we owe a debt of gratitude for pointing this out.

Fraser Roberts' book is well known and as an introduction to the principles of medical genetics has not been bettered. The clarity of the prose and of the diagrams and the exposition of difficult subjects such as multifactorial inheritance are first class. In the third edition there is an entirely new chapter on the chromosomes and the section on the blood groups has been augmented by a good description of the sex-linked system Xg. However, DNA, inherited biochemical abnormalities and pharmacogenetics only account in all for seven pages. One feels that in 1963 the lessons of the earlier editions had become part of generalised knowledge and that qualified men, anyhow, now require increasing advice on clinical problems. True, Fraser Roberts gives it to them in the important field of counselling, but congenital malformations should receive more attention, and the impact of genetics on epidemiology—motor neurone disease in Guam and kuru in the Fore Islands for example—and the role of inheritance in auto-immune disease and leukæmia should be elaborated. The author would no doubt argue that what he is writing about is *basic* genetics, and its applications should be sought for elsewhere. However, in the reviewer's opinion he should cater more for that not entirely reprehensible character, the clinician who wants to run before he can walk. Notwithstanding the above, the book remains a classic for the beginner and is a pleasure to read.

Lenz's book is considerably more clinical and though much of the basic information is necessarily the same as in Fraser Roberts' there is more the feeling that he is constantly relating the facts to biochemistry and cytology. He gives a good account of DNA (with its application to the hæmoglobins) and raises interesting speculations, for example, as to whether dominant hereditary diseases are due to the presence of a pathological gene product rather than to the lack of a normal one—though if this were so it would be difficult to square with current ideas on the evolution of dominance. There is a very good section on mutations—point, chromosomal and somatic—and in the same chapter the concepts both of biological fitness and natural selection in man are dealt with. The book is particularly good in raising problems and pointing out difficulties and this makes it convincing to clinicians, who are constantly dealing with the inexplicable. It is a pity that the translation has retained the ponderousness of the original German but nevertheless the exposition is clear and there is an excellent glossary of technical terms for the help of the non-medical reader.

The South African porphyria story by Geoffrey Dean is a good example of what an enterprising physician can do, in the course of his work, even though as a geneticist his amateur status is never in doubt.

In 1652 the Dutch government sent out 70 men to colonise a revictualling base for ships en route for India. White women were in short supply and in 1688 eight orphan girls were despatched from Rotterdam with a view to providing wives. The little colony was also augmented by Huguenots after the revocation of the Edict of Nantes and when the orphan girls arrived probably consisted of about 300 people. Today the white population of South Africa is around 3,000,000 of which about a million are Dutch deriving almost entirely from the original few settlers.

Dean emigrated to South Africa in 1947 and soon realised that the Port Elizabethans had a high incidence of porphyria, a *rara avis* in European populations. Here it is usually misdiagnosed, because not thought of, appendices being needlessly removed for the bouts of abdominal pain (which are subsequently labelled " neurosis ") the skin lesions being called eczema and the neuritis " idiopathic " or " viral ". The clue lies in the urine, which is reddish-brown and fluoresces in ultra-violet light. Various forms of the disease are known but there is no doubt that the one which is so common in South Africa is inherited as an autosomal dominant. In earlier years mainly only a nuisance, the disease is now often lethal because of the extensive use of barbiturates, particularly thiopentone anæsthesia which precipitates attacks and may lead to death from respiratory paralysis.

The medical aspects of the disease are well dealt with under the usual headings of diagnosis, differential diagnosis (there is a very good phenocopy in Turkey due to eating bread treated with a fungicide), prophylaxis by means of routine urine screening and treatment of the established condition. In the biochemistry the nature of the block is not known and it is of interest that injection of porphyrin precursors does not cause neurological or gastro-intestinal symptoms.

For readers of *Heredity*, however, the most interesting part of the story is the tracing of the origin of the disease to one of two of the early settlers in 1688. It is only possible to be certain of affected individuals (because of a reported history of fragility of the skin) as far back as 1800 but everyone at about this time with the skin lesion derives from Geritt Jansz or from the imported orphan Ariaantje Jacobs whom he married in 1688. By routine screening and by a study of Dutch family names and hospital, parish and civic records he has estimated the incidence of the disease at the present time and thinks that it is about three per thousand of the white population. (A criticism in passing; " a very high prevalence for a dominant gene " is meaningless.) This high incidence is most impressive, but Dean appears not to realise that the proportion of individuals affected is much the same now as it was in 1688; it is the population which has increased, and what is described is an interesting example of the founder principle at work in man and there appears to have been little selection against the gene until the present time. In a future edition this could profitably be discussed and the arguments for genetic drift versus natural selection set out.

Dr Dean has carried out an excellent piece of research and is to be congratulated on his original observation and on the subsequent detective work.

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