Surveys, Symposia, and Transactions

Research in Muscular Dystrophy. Proceedings of the Fourth Symposium on Current Research in Muscular Dystrophy held at the National Hospital, Queen Square, London, W.C.1, January 1968. Edited by the Members of the Research Committee of the Muscular Dystrophy Group. (Pp. 472; illustrated +tables. 60s.) London: Pitman Medical Publishing Co. 1968.

The aims of the Muscular Dystrophy Group of Great Britain are first to form a friendly link between sufferers and their families, and second to make available funds for medical research in muscular dystrophy and other muscle diseases. Recently the Research Committee decided to support all disorders affecting muscles, whether of the muscle cell itself or of its innervation, and such conditions as myasthenia gravis and spinal muscular atrophy are now also its concern.

This volume contains the 33 papers (collected into 5 chapters) presented at the Fourth Symposium held in January 1968, and also an edited version of the discussion which followed the individual papers. Chapter I consists of the Joan Vincent Memorial Lecture, established in memory of the first secretary of the Group and given by F. J. Nattrass, who classifies the 'pure' muscular dystrophies, reviews modern developments, and gives some fascinating biographical details about Duchenne—'a strange, sauntering mariner-like figure' from Boulogne.

In Chapter II (clinical and genetic studies) there are four papers of particular interest to the reviewer. (1) The correlations between myasthenia gravis and disorders of the thyroid gland (J. A. Simpson)—and these are by no means only confined to thyrotoxicosis; (2) second thoughts on classification of the muscular dystrophies (J. N. Walton and D. Gardner-Medwin) in which the non-progressive and ill-understood congenital myopathies (such as central core disease and nemaline myopathy with its affinities with Marfan's syndrome) are discussed in some detail and compared with the better known progressive types; (3) a description of the important benign X-linked muscular dystrophy (A. E. H. Emery); and (4) the exciting discovery that by cross-innervation of the two fibre types (fast and slow), the histochemical pattern of human skeletal muscle can be changed, suggesting that the dystrophies may prove eventually to be an aberration of neural influences rather than primary muscle disorders (Dubowitz).

In Chapter III (anatomical and pathological studies), two papers are selected for comment. The first questions whether the two major types of nerve lesion, Wällerian degeneration (as exemplified by isoniazid neuropathy) and segmental demyelination without axon damage (e.g. alloxan diabetic neuropathy), are in fact distinct entities, and the authors (Hildebrand, Joffroy, and Coërs) suggest that the two forms may merely be the result of differences in the severity of the disease process. The second deals with the structure and fine structure of the intramuscular nerve endings in dystrophia myotonica, and the authors (Allen, Johnson, and Woolf) show that though the architecture of the motor end plates is largely normal, yet there is a tendency for them to be unusually elaborate, suggesting that the number of available binding sites for acetylcholine and the amount of acetylcholinesterase present would be increased. It is of great interest that somewhat similar findings are found in two quite different conditions, namely amyotrophic lateral sclerosis and myasthenia gravis, the latter in some ways being the reciprocal of dystrophia myotonica.

In Chapter IV (biochemical studies) there is a paper (W. H. S. Thomson) critically assessing the reliability of the biochemical identification of the carrier-state in X-linked muscular dystrophy, and there is emphasis on the importance of bearing in mind the diurnal variation of the enzyme creatine phosphokinase. When this is done the tests are satisfactory, for of 9 undoubted heterozygotes only 1 gave normal serum enzyme values (despite a suggestive EMG) in every circumstance. One of the heterozygotes investigated was found to have micro-symptoms shown by incapacity to sustain rope skipping as a schoolgirl.

Finally in Chapter V (neurophysiological studies) there are two papers (D. Gardner-Medwin and R. G. Willison) on the problems encountered when using electromyography to try and detect carriers of the Duchenne type of dystrophy. These pitfalls should be widely known by registrars and others who ask for 'EMG studies, please' and feel disappointed when the reports are equivocal.

The book is in the usual good value Pitman style and is strongly recommended, being of use both to general physicians and also to neurologists, biochemists, and neurophysiologists—and the discussions following the papers round off the topics particularly well.

C. A. CLARKE

Screening in Medical Care. Reviewing the Evidence. A Collection of Essays. Prepared by the members of a group brought together by the Nuffield Provincial Hospitals Trust. (Pp. 173; tables. 35s.) London: Oxford University Press for the Nuffield Provincial Hospitals Trust. 1968.

Since the war ended both the benefits and the hazards of medical treatment have been extended, under the National Health Service, to all persons with symptoms without reference to the cost to the individual. As some diseases are not responsive to therapy by the time they are manifest through symptoms (it is convenient to follow the classical usage of *symptoms*, by which a disease is manifest to a patient, and *signs*, by which it is manifest to others), various attempts have been made to increase the scope of treatment by inverting the normal sequence of discovery and seeking signs rather than symptoms, in the hope that earlier treatment would make life both longer and more pleasant.

In some organs, such as the lung, which can be displayed by *x*-ray to reveal early stages of tuberculosis

which respond to treatment, screening procedures were rapidly brought into use on both economic and humane grounds. As they were safe, painless, intelligible to most persons, and claimed to help eliminate an illness within the personal experience of most adults the technique spread rapidly. Since cancers must also start somewhere, similar attempts were made to detect presymptomatic signs, an attempt partly based on the obsolete concept that the lymphatics carried the lethal cells in leisured cohorts beyond reach of the knife. The veins are now known to offer quicker travel. This procedure was particularly attractive in the breast which was palpable, and the cervix which could be scraped. Both procedures became widely recommended, and liberally financed, without any clear evaluation of the relative risks and costs of removing what was healthy as opposed to not removing an organ which would show suggestive signs of cancer if examined.

The slow and steady extension of medical curiosity from that minority, first defined by the rich and ill, and then by the ill, to the whole population, has changed the basis of medical responsibility, since the doctor goes to the patient, and, in consequence, must be even more careful to do more good than harm. In order to explore some of these problems a series of essays was written under the chairmanship of Professor McKeown, with the sponsorship of the Nuffield Provincial Hospitals Trust, both to discuss the general problem and to evaluate some of the screening procedures in use.

Of the 10 disorders, or groups of disorders, currently exposed to enforced scrutiny only 4 are said, on the dust jacket, to be based on valid evidence. One of these appears to be phenylketonuria, though the lucid article by Dr. J. M. G. Wilson shows the weakness of even this evidence. For human geneticists this disorder is of prime interest, not because of its physical impact, for it is rare and only partially curable by a diet whose unpalatability is itself a threat to normal development, but because, in the excitement of finding a potentially curable disorder which could only be diagnosed by chemical methods, it led to such enthusiasms—mainly among physicians interested in novel therapeutic advances and geneticists inexperienced in medicine but exasperated by the ignorance and complacency they found there in relation to genetics—that vast projects were imposed with a rapidity appropriate to an epidemic disease and without clear costing or open discussion. (In some American states emergency legislation was introduced.)

By diverting midwives from midwifery and hospital biochemists from hospital biochemistry a vast network is being extended to catch some 50 children a year in Britain who may or may not need a diet they may or may not get. This diversion may or may not be justified, but the decision has been taken, and it will be difficult to stop a steady growth in demand for more and more blood for detection of rarer and rarer diseases unless medical geneticists take an active and informed part in the orderly and harmless allocation of resources.

In this disorder, and in some other disorders which contain a subgroup of mendelian conditions, such as deafness, or show intense familial concentrations which are doubtless in part genetic, such as diabetes and glaucoma, or which are due to genetically determined antipathies, as in rhesus sensitization, the authors have attempted to evaluate the humane and economic consequences of investigating those who do not, or cannot, complain. No one who has read the book, or its introduction by Lord Cohen, will discard it as a mere numerical exercise by those ignorant of clinical medicine. The remarkable point is that a book of this nature should not have been written earlier, and that, for 20 years, neither the Medical Research Council nor the National Health Service should have noticed the vast and fertile no man's land lying between their more obvious responsibilities.

The book cannot be recommended too highly. It is appropriate that Lord Nuffield, who was introduced to medicine by the erratic performance of Osler's car, and who exploited the inspection of apparently working parts in the motor industry in order to provide a guaranteed product by economic methods, should have endowed the organization responsible for one of the first discussions of the orderly and beneficial deployment of inspection procedures to man.

J. H. EDWARDS

Shorter Notices

Forerunners of Darwin, 1745–1859. Edited by Bentley Glass, Owsei Temkin, and William L. Straus, Jr., under the auspices of the Johns Hopkins History of Ideas Club. Johns Hopkins Paperback 41. (Pp. xxii+471; 6 figures. 28s. 6d.) Baltimore: Johns Hopkins Press; London: Oxford University Press. 1968.

Not the least of the services of the History of Ideas Club at Johns Hopkins was the publication in 1959 of a series of outstanding historical and philosophical essays brought together on the centenary of the *Origin of Species*. This welcome paperback edition—the third reprint of the collection—carries a valuable bibliographical note by Richard Macksey on the literature from 1959 to 1967.

Genetische Familienberatung. Ein Leitfaden für den Artz. By Walter Fuhrmann and Friedrich Vogel. (Pp. viii+98; 27 figures+14 tables. DM. 8.30. $2.20.) Berlin: Springer. 1968.

This little book on genetic counselling is one of the series of authoritative Heidelberg Pocket Monographs. It is intended for the general practitioner and covers adequately the elementary human genetics necessary in such an exposition. The empirical aspect of much of the advice that can be given is stressed, as are the avoidable difficulties. The text is lucid and is greatly helped by well-constructed Tables.