Book reviews

Journal of Medical Genetics (1975). **12**, 116–117.

Selected Screening Tests for Genetic Metabolic Diseases. By G. H. Thomas and R. Rodney Howell. (Pp. vii+101; figures+tables. £3·50.) Chicago: Year Book Medical Publishers; London: Lloyd-Luke. 1973.

This is a remarkable book both in its conception and its execution. Few would have thought the subject could have justified a volume of this size and few would have expected it to turn out so well. The book is saved by the considerable practical laboratory experience of the authors, and it will find a ready place on the shelves of any paediatric, genetics, or mental subnormality unit, and if it is not there it will be on someone's laboratory bench.

Simple observation by smell and vision are rightly placed first in the examination of urine, and then the apparently simple ferric chloride test is treated in all its variations in its own chapter of 10 pages and 44 references! Reagents for keto acids, phenols, sulphur compounds, and reducing substances and their significance in relation to metabolic disease are described. Other sections detail the favoured method of screening for mucopolysaccharidoses, methylmalonic aciduria, metachromatic leukodystrophy, GM_1 gangliosidosis, and oxaluria.

Throughout it is emphasised that the tests are simple screens and require more specialized tests for definitive diagnosis. But in many instances those described take the investigator very close to a conclusion. Indeed sometimes the screen is no simpler or more rapid than a definitive test; for example, assay of plasma or urine enzymes instead of those in leucocytes for the neurodegenerative disorders. The section on mucopolysaccharidoses is remarkably full and accurate, but it is surprising that while the lack of specificity of the toluidine blue spot test is recognized, no mention is made of the much more specific dye alcian blue which has been in use for over 10 years.

The value of the test for sulphite oxidase deficiency (part of the differential diagnosis for ectopia lentis, with lysinuria and homocystinuria) must be unestablished since it was developed after the only known case had been diagnosed. However, it is regularly used when urine from patients with dislocated lens give a negative reaction with cyanide-nitroprusside.

The book is well produced and very good value for the price and, provided the warnings that more definitive diagnostic tests may be required are heeded, it will do much to improve the standard of laboratory testing in the field of inherited metabolic disease.

D. N. RAINE

Personality Differences and Biological Variations. A Study of Twins. By Gordon Claridge, Sandra Canter, and W. I. Hume. (Pp. viii+175; tables. £3·80.) Pergamon Press, Oxford. 1973.

The project described in this book consists of a series of studies of different aspects of the behaviour of twins. Its main interest lies in the senior author's attempt to bring the observations into the context of the theoretical model which he has been working on for a number of years. The approach is derived from the work of Pavlov, as developed in this country mainly by Eysenck. Like Eysenck, Dr Claridge is concerned with three continuously variable dimensions of personality, introversion–extraversion, neuroticism, and psychoticism which are assumed to run through the general population and to account for normal personality differences. In Dr Claridge's model, however, the last two dimensions, instead of ranging from the abnormal to the normal, stretch respectively from dysthymia to hysteria, and from active psychosis to retarded psychosis. Furthermore, the different dimensions of personality are determined by differences in interaction between the underlying psychophysiological processes, called 'tonic arousal' and 'arousal modulation'. The former is measured by autonomic activity and sedation threshold and is manifested as high and low anxiety drive. The latter has a CNS regulating function, and is concerned with sensory input and attention; it is associated mainly with EEG parameters and with introversion–extraversion. High tonic arousal is associated with active psychosis and dysthymia, low tonic arousal with retarded psychosis and hysteria. In respect of arousal modulation, however, the relationships are reversed; low arousal modulation (and extraversion) is associated with active psychosis and hysteria, and high arousal modulation (and introversion) with retarded psychosis and dysthmia. This model seems to make some clinical sense—though it is not clear how it encompasses the differences between manic-depressive psychosis and schizophrenia.

Hitherto the authors' researches were mostly confined to psychotic patients, and the dimensional aspect of their hypothesis remained conjectural. The unexpected windfall of a sizeable sample of twins collected by another group of workers for a completely different purpose, has now made it possible for them to test out the model on a group of supposedly normal subjects, and to investigate its genetic basis. On the whole, the results support the dimensional model of both neurosis and psychosis. Interestingly, the twin analyses suggest that some parameters are much more under genetic influences than others.

Much painstaking work and thought have evidently been put into the collection and analysis of the data, and several of the individual findings are of interest. As a whole the research, of which this is only part, represents a well-sustained and imaginative effort. It could eventually bring about radical changes in our understanding of the nature of psychotic illness, and its relationship to neurosis and normality.

D. W. K. KAY

Huntington's Chorea, 1872–1972. Edited by A. Barbeau, T. N. Chase, G. W. Paulson. (Advances in Neurology, vol. 1.) (Pp. xxii + 826; figures + tables. Dfl.150.) Amsterdam: North Holland. 1973.

During the middle decades of the 19th century the familial form of progressive adult chorea associated with dementia was recognized but, for reasons that are not altogether apparent, it was George Huntington's comparatively late account of the disorder which was first to be widely publicized. To celebrate the centenary of this description, a symposium was held during March 1972, near where Huntington used to live, in Columbus, Ohio and the various reviews and papers read at this meeting make up the present volume. As it took a long time for the disease to be widely recognized, so knowledge concerning the condition has accumulated gradually, and indeed if the symposium had been held 15 to 20 years ago, there would have been little to report that would have surprised even Huntington himself. The considerable amount of work done in recent years, and the rather more modest advances in knowledge, justify the publication of a large work on the disease, although it may be somewhat unexpected to those not directly concerned with the condition, that it has been possible to produce 826 pages on this comparatively obscure disorder.

In this book most aspects of the disease are covered in detail, few are treated in an incomplete or unbalanced manner, and virtually everything that is worth saying about the condition is said. As there are many authors there is inevitably a lot of repetition which, whilst it is sometimes annoying, is not really the bad thing that it is sometimes said to be, for occasionally a better perspective can be obtained by reading several accounts of the same subject. The book is divided into a number of sections, each containing one or more review articles as well as several smaller papers. The subjects dealt with include 'History', 'Clinical variants and differential diagnosis', 'Genetics and epidemiology', 'Early detection', 'Pathology', 'Biochemistry', 'Experimental models', 'Behaviour and social aspects', and 'Management'.

The section on history contains some of the other early descriptions, as well as that by Huntington and it also contains several detailed references to one aspect of the history of the disease which is now in serious doubt, without there being any acknowledgment of the existance of these doubts. This concerns the story of the emigration from Essex in 1630 of some of the alleged ancestors of a number of American choreics, a story that is based on genealogical work which has recently been called into question. The subject of variants is dealt with in the appropriate section and is also touched upon in the section on pathology. The clinical and pathological features of these variants are outlined several times and their genetic characteristics are described. The question as to whether all of these variants are really distinctive is also raised. The section on preclinical diagnosis is headed by a thoughtful and provocative review and amongst the papers that follow are two on the laevodopa load test, this being a test in which subjects at risk are given oral laevodopa to establish whether or not involuntary movements can be provoked, the assumption being that those with the abnormal gene will develop such movements whereas those without will not. Contained in the lead review is a plea for answers to the many difficult questions posed by the possible preclinical diagnosis of such a grave disease, but this aspect is either ignored or at best only mentioned in passing in subsequent papers. The section on pathology contains several papers on the variety of changes that can occur in the classical form of the disease as well as in some of the variants. The value of cerebral biopsies is also discussed. The lead review on biochemistry by Barbeau is immensely detailed and contains 268 references, most of which are to studies that have yielded negative or equivocal results. Included also is the paper by Perry and colleagues in which they first report their finding of low levels of gamma aminobutyric acid in the brains of choreics, an observation that has since been followed up by the finding of changes in the amounts of glutamic acid decarboxylase activity in similar brains. This is one of the very few occasions when the book can be seen to be already out of date. The section on experimental models contains a wealth of information on a number of drug induced and naturally occurring animal disorders; amongst the latter being the endearing but, in this context, probably irrelevant acrobatic rabbits and the quaint hyperkinetic episodes of scottie dogs. The section on management touches on many aspects including the management of the family as well as the care of the choreic individual, but the parts dealing with the drug treatment of chorea are a little eccentric to British eyes, for the two drugs that are used widely in this country—tetrabenazine and thiopropazate—are either mentioned very briefly or, in the case of the latter drug, not at all.

Notwithstanding the criticisms, this book is the best and most detailed account of this disorder that is currently available and it should be on the shelves of every major library used by neurologists, geneticists and psychiatrists. The problem is that it is enormous, and as it is hardly practical to read it from cover to cover, a newcomer to the disease would find it difficult to obtain an overall view of the disease without a lot of effort. It is also very expensive, the United Kingdom price being well over £20, which means that it will be purchased by comparatively few private individuals and, one suspects, few small medical libraries either. Thus, the two major contemporary sources of detailed information on the condition—this book and the even more expensive sixth volume 'Handbook of clinical neurology'—will not be available to most of those who might wish to consult them. Something smaller and cheaper is needed.

DAVID L. STEVENS