Book Reviews

This is a book which clinical geneticists and those interested in malformations will enjoy looking at and reading. It is probably best bought together with the companion volumes rather than in isolation. It supplements but does not replace other illustrated catalogues which have been recently published on the malformation syndromes.

J. INSLEY


Alzheimer’s disease is a moderately infrequent form of presenile dementia, with a well-defined course and clinical picture, and characteristic cerebral pathology. Genetically it is interesting because of the indications of genetic heterogeneity within this clinico-pathologically delimited syndrome. A number of families have been described, in his contribution to the symposium Pratt says ‘more than a dozen’, in which a completely typical form of autosomal dominant inheritance is shown, with equal sex representation, and nearly complete penetrance. In comprehensive series, however, selected on a clinico-pathological basis, these typical families do not appear; and what one sees instead is a moderately raised familial incidence and a female preponderance of about 3:1, i.e. the indications of a polygenic basis. The genetical evidence, therefore, is strongly in favour of regarding the syndrome as being a mixture of at least two different and independent conditions. This is a conclusion which, for the most part, both clinicians and pathologists are unwilling to accept, as is made plain in these papers.

The symposium reported here was predominantly a pathological one, with interesting and original patho logical work. Participants came from Belgium, Germany, the Argentine, Sweden, Hungary, the USA (7) and Britain (10). Active work is going on in histology, histochemistry, and electron-microscopy. Most promising, perhaps, is the study of the ultramicroscopic structure, i.e. the abnormal intracellular microtubules, which differentiate more than one kind of lesion, and may lead to a successful breaking down of the clinico-pathological continuum extending into normal senescence, senile dementia, and cerebrovascular disease, by which most workers are at present both obsessed and bewildered.

The reviewer would suppose that this book is an important one for the neuropathologist. Its genetical interest is not great. Apart from Pratt’s paper reviewing the present state of genetical understanding, the main interest lies in the communication by Jacob (Marburg, Germany) of a family with 6 females in 3 generations showing a form of Alzheimer’s disease with, in every case, the unusual feature of myoclonic twitchings appearing early in the course. The literature is reviewed by Jacob.

The book is well produced, with an abundance of good pathological illustrations, good papers, lively discussions, but rather inconclusive results.

E. SLATER


St. Helena shares with its near neighbour Tristan da Cuhna many of the aspects of an isolated community. It is however an older settlement and has population of some 5000 against the 350 or so of Tristan da Cuhna. A genetical survey of this highly inbred population with a culture of its own and its particular racial amalgam is a worth-while task which Dr. Ian Shine carried out single-handed while acting as medical officer to the island during his national service in the Army. The present book, based on an examination of almost the whole of the population, records his findings.

One study was devoted to hallux valgus. On St. Helena as elsewhere it was common among those who wear shoes and uncommon among the barefooted. Regression analysis showed however that an additional factor, probably genetic, was also operative. Furthermore some families seemed particularly susceptible to the deforming effect of shoes, while others developed the deformity in spite of their barefootedness. An equally detailed study on the frequency of ischaemic heart disease revealed a substantial incidence in the 1500 subjects over the age of 30 examined. This was unexpected, but clear aetiological conclusions were not possible.

In the course of the overall survey a large number of genetic disorders were observed, including some that are distinctly uncommon, such as albinism, retinitis pigmentosa, lithopaeonia, ichthyosiform erythrodermia and Christmas disease. Undescribed disorders such as gross unilateral genu valgum, ‘familial St. Helenian fever’, and dwarfs with pseudobrachydactyly constituted a group of considerable interest. A high incidence of pterygium presented a puzzle, for the commonly accepted exciting factors, such as dust, wind, and dry air, could not be invoked as an explanation.

The genetic problems disclosed in this investigation are discussed in a critical penultimate chapter on in-breeding. Like the rest of the text this is competent, but this useful volume is not well served by its rather flippant title and somewhat jaunty style.

ARNOLD SORSBY


The de Lange syndrome was first described by a Dutch paediatrician from Amsterdam in 1933. Since that time a wealth of data has accumulated on the clinical
aspects of this syndrome, but no facts have emerged regarding its aetiology. In this monograph Professor Berg and his colleagues present an admirable review of the de Lange syndrome, mainly its clinical aspects. Their review is based on 18 personal cases which are carefully documented as regards clinical features (there are photographs of each case), dermatoglyphs, cytogenetics, and family information; and on data from over 200 previously reported cases. On the combined experience they list the most important diagnostic features of the syndrome. These are: low birthweight, retarded mental and physical development, hirsutism, micro-brachycephaly, synphry, anteverted nostrils, prominent philtrum, thin turned-down lips, limitation of elbow extension, single transverse palmar crease, proximally placed thumbs, clinodactyly of fifth fingers, malformed upper limbs, and webbing of second and third toes.

The authors discuss the aetiology of the syndrome in the closing chapters. They estimate the incidence to be between 1 in 30,000 and 1 in 60,000 live births. There is an increased incidence in sibs, raising the possibilities of: polygenic inheritance; recessive inheritance with incomplete penetrance; or a chromosomal abnormality, not detectable by present techniques. The authors favour the latter possibility but not very strongly, and suggest that perhaps there are several causes, and the syndrome is heterogeneous. But whatever the aetiology, it is clear that there is sometimes recurrence in the same sibship, and the authors give, for purposes of genetic counselling, an empirical risk of recurrence of 4 per cent.

The monograph provides excellent and well presented data on this clinical disorder and will be a useful book of reference for those working in the field of childhood syndromes. It is however an expensive book, and much of the contents have been reported previously, in particular by the authors. A review article in a journal would, I think, have been more appropriate than a costly book describing a condition about which there is still much to be learnt.

Finally, I should mention that some of the tables and appendices will be useful for workers in other disciplines of genetics: namely the figures describing birthweight and head circumference of normal and mentally retarded children, and the tables relating parental age to birth order.

SARAH BUNDEY


This book gives a remarkably comprehensive review of the epidemiology of mongolism by an authority in social medicine who has not himself done much work on mongolism and so is able to look at the literature from the outside without personal predilections. A critical survey of this kind is needed since much of the epidemiological work that has been done has been uncritical and a whole series of claims has been made that have not been confirmed. Some of these claims, for example, that maternal anxiety in early pregnancy tends to cause mongolism, were seen to be implausible when it was discovered that G trisomy was present—an abnormality that must almost always have been present at conception. Other claims that may be more plausibly related to non-disjunction in oogenesis, but nevertheless were based on shaky foundations, include the following: that epidemics of infectious hepatitis were related to clusters of mongol births, that the birth of a mongol was preceded by a period of infertility, that late fertilization of the ovum predisposed to mongolism, that intrauterine infection with mycoplasma predisposed to mongolism, that abnormal excretion of dehydroepiandrosterone was related to mongolism. Except in the last case where Dr Lilienfeld and Dr Benesch have missed the non-confirmatory study, claim and counterclaim are set out clearly. Other claims, such as the relation of preconceptional diagnostic radiology and maternal thyroid antibodies to mongolism, are still sub judice. In contrast the most striking epidemiological feature of mongolism, the relation to maternal age is well established, but not yet explained.

Dr Lilienfeld and Dr Benesch are perhaps too ready to accept the indications that there may be racial differences in the aetiology of mongolism beyond those due to differences in maternal age distribution. They are perhaps too doubtful of the association of Hirschsprung's disease and mongolism. On the whole, however, they are a wise guide to the complexities of the great variety of epidemiological studies they consider, and workers in mental subnormality will find it most useful to have the widely scattered literature assembled and evaluated in this book.

C. O. CARTER


Seven short articles published in The Lancet during the summer of 1969 have been assembled with two additional chapters to provide an excellent short account of medical genetics. As the book is primarily concerned with the practical aspects of clinical genetics, including genetic counselling and the control of genetically determined diseases, it will appeal principally to medical practitioners in all specialties who may have little training in formal genetics, but who are anxious to bring themselves up to date and learn what part modern genetics has to play in the practice of clinical medicine. It is also a heaven-sent boon to the hard-pressed medical student who is usually expected to understand genetics after the too short a course of lectures. He will find in this book, which can be read through in two evenings, an excellent introduction to the subject and, moreover, one written by a physician with extensive experience in giving genetic advice to patients. Though some knowledge of clinical medicine is presumed, the non-clinician need not be deterred. In fact, any non-medical biologist charged with teaching genetics to medical students would...
The De Lange Syndrome

Sarah Bundey

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