

Medical Genetic Studies of the Amish.

Selected Papers

By V. A. McKusick. (Pp. x + 525; Figures + Tables. £19·25.) Baltimore and London: Johns Hopkins University Press. 1978.

To read this edited collection of papers is both a fascinating experience and, for one already familiar with the Amish, a pleasurable reminder of the persistence and success of their remarkable way of life in 20th century America. A visitor to the Moore Clinic at Johns Hopkins Hospital in the late 1960s might have been excused if he had thought that the Amish formed a majority of the Baltimore population, but this book shows clearly how worthwhile the intensive study of this group has proved in both medical and population genetics.

The papers are reproduced unchanged, but their value is greatly increased by the addition of notes on subsequent developments. They are grouped into those dealing with general and population genetics, studies of previously recognised Mendelian syndromes, and (the largest section) 'new recessively inherited entities, many of which would have been difficult, if not impossible, to recognise outside the setting of a defined, closed, and well documented community as provided by the Amish'. Among these delineations of new disorders, that of 'cartilage hair hypoplasia' still stands out for its clarity and its masterly combination of clinical and genetic information.

The value of the studies in this book extends far beyond the demonstration that the disorders in question are recessively inherited. The range of clinical expression produced by what clearly must be a single gene is something that is difficult to document in the more heterogeneous patients seen in most populations. The studies of autosomal recessive limb-girdle muscular dystrophy and of the 'Troyer syndrome' provide excellent examples of this. Another particularly impressive feature of the Amish studies is the detail and accuracy of the genealogical and demographic work which underlies them, much of it due to the attachment of Amish people themselves to this type of documentation. It is of interest that a number of the main contributors to the book are themselves of Amish origin, and it is fortunate that the Amish antipathy to further education has not prevented the emergence of those individuals to transmit the unique aspects of Amish culture to the world in general.

Anyone working in medical genetics to whom the Amish are unfamiliar should read this book without delay; it will be a revelation as to the value of studying genetic isolates. Those who do know of the Amish will not need encouragement to refresh their memory of the subject and gain new ideas. It should also serve as a stimulus for people to study their own equivalents of the Amish. Though few of us have access to such an ideal population, there are many minority groups which would repay much closer investigation, where studies of genetic diseases have not been attempted. Many of these groups, less robust than the Amish, may soon disappear for ever if the opportunity is not taken soon.

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British Medical Bulletin: The HLA System

Vol. 34, No. 3, September 1978. Scientific Editor, W. F. Bodmer. (Figures + Tables. £5·00.) London: The British Council. 1978.

Medical journals are now replete with articles on HLA and, though weary voices have been heard to exclaim 'not HLA again', we should take heart. Cytogenetics had similarly inauspicious beginnings when, 20 years ago, pictures of 'squashed spiders' began to appear in medical journals. There are many aspects of HLA which are relatively simple, though others seem comprehensible only to experienced travellers in the exotic lands of lymphomania. The latest issue of the British Medical Bulletin provides an excellent overview of the HLA system and will do much to enlighten the weary. It is a convenient introductory package for this fascinating genetic system. I recommend reading 'Evolution and Function of the HLA System' at an early stage as, in it, central issues relating to the system are dealt with succinctly and comprehensibly. The other articles can be divided into several categories. Firstly, the 'technology' of HLA: serology, cellular typing, the chemistry of HLA antigens, and the 'new' HLA-DRW locus are considered in 4 articles. Secondly, the role of HLA as part of the major histocompatibility system (MHS) is discussed in 2 articles devoted to complement genetics and to the immune response. Thirdly, the somewhat variable importance of HLA in human organ transplantation is described objectively. The fourth category, that of the association between HLA and various diseases, takes up a large part of the Bulletin in a total of 8 papers.