Book reviews

The Prenatal Diagnosis of Hereditary Disorders.

The author, from the Birth Defects and Genetic Clinic at Harvard, has addressed this book to those who may want to refer patients for prenatal diagnosis. The conditions for which prenatal diagnosis are possible, the dangers and limitations of the procedure, the interrelation with screening for heterozygous carriers of genes for recessive disorders, and the moral dilemmas posed are all well described. Technical details of clinical and laboratory procedures are not given. In order to justify writing a book rather than a paper the author has, perhaps, included unnecessary material. Accounts of the clinical features of the neurolipidoses and mucopolysaccharidoses, for example, are available elsewhere. Statements on the principles of genetic counselling are also available elsewhere. The ethical issues appear to be more straightforward in the more homogeneous British society than in the United States. The main difficulty in dealing with the situation in which, for example, an XXY fetus is found is that we do not yet know what prognosis to give the parents for the future child, and so cannot provide the information which they need in order to make a decision about termination themselves. The author includes some useful tables, for example of the conditions for which prenatal diagnosis has already been achieved, of those for which it is likely to be feasible with present techniques, and of the findings on amniocentesis so far in a number of co-operating centres in the United States.

Overall the book is valuable and opportune, though it could perhaps have been pruned to half its length.


Modern work on human chromosomes, which can be dated from 1966 but which in practice started in 1959, has an intrinsic biological interest and has found immediate and practical application to diagnosis and prognosis as a guide to treatment and as a means of often simplifying and making more accurate clinical investigation. A more recent application is to genetic counselling for the prenatal diagnosis of chromosome disorders by diagnostic amniocentesis, which has allowed the counsellor to move, often, from probabilities to almost certainties. Nuclear sexing (or Barr body sexing) of amniotic cells, which predated chromosome studies, has now been expanded and made more accurate by the complementary method of fluorescence Y-sexing (sometimes called F-body sexing), while the various forms of chromosome identification by banding are also useful in practice. In addition, methods have been and are being developed for the identification of generally recessively inherited inborn metabolic errors caused by gene mutation, while dominant disorders are still a problem. Here, awkwardly, diagnosis might for some time have to rest on linkage information still to be developed. Congenital morphological anomalies, other than those attributable to the above, are beginning to be diagnosable in early pregnancy.

It is to the prenatal diagnosis of conditions resulting from abnormalities of the genetic material that this slim volume, edited by Emery, is devoted. It naturally pivots around the central problem of selective abortion of the fetus 'that is likely to be substantially handicapped' (Abortion Act, 1967), and by implication the consequent chance of some couples to have normal children, and also by implication the changes which some of the available procedures are bringing to the practice of genetic counselling. The book is largely about facts and techniques and deals not with ethical or other related issues. While other approaches to prenatal diagnosis are also considered (by Scrimgeour), most of the book is devoted to amniocentesis and to the uses of the cells and fluid for diagnostic purposes.

The results and prospects for the detection of inborn metabolic errors are informatively discussed (Brock), as are the more strictly cytological techniques (Nelson). There is a good summary of what is found and can be expected from the biochemical study of the fluid itself (Emery). The importance of a raised α-fetoprotein level in the amniotic fluid in neural tube defects was discovered by members of the team of contributors to this book. We now have the great value of this technique in identifying 'open' neural anomalies. The final chapter (Smith) on the implication of amniocentesis deals with the burden of genetic disease, with an assessment of cost-benefit to society of prenatal diagnosis, with the difference which prospective as opposed to retrospective diagnosis obviously makes, and with the 'beneficial' (i.e. eugenic) as against the dysgenic effects of the ability to diagnose prenatally and to terminate the pregnancy when the results show abnormality. The relative importance of these effects on gene frequency is considered in relation to the type of disorder, the reproductive efficiency of the affected, and the reproductive compensation by the parents who will be substituting nonaffected for affected children, if they have not already tended to compensate for the affected.

The book is clear and succinct and is a good introduction to a moving and growing aspect of applied genetics.