Book reviews


Until three or four years ago parents faced with a high risk of producing a child with a serious genetic disorder had no option but to accept the risk and chance of having an affected child, or limit their family completely. The advent of antenatal diagnosis with the possibility of selective abortion of affected fetuses has altered the situation completely. In the case of chromosomal disorders (particularly Down’s syndrome), certain metabolic disorders (but unfortunately not yet fibrocystic disease), and probably anencephaly and open spina bifida a mother can now be offered amniocentesis with confidence that an affected child can be detected in utero.

The author has reviewed the subject of antenatal diagnosis in regard to the risks of amniocentesis (currently being assessed in Britain by an MRC working party), the possibilities of detecting chromosomal and inherited biochemical disorders from the study of cultured amniotic fluid cells, and those disorders which can be diagnosed from studying the composition of amniotic fluid itself. Finally there are chapters on genetic counselling and its relationship to antenatal diagnosis and inevitably a consideration of some of the moral, ethical, and legal problems which surround the subject.

The book is well written and well referenced though some might find the numbering of citations less valuable than using the authors’ names and arranging them alphabetically in the bibliography. By present day standards it is very good value and can be recommended to all those involved in the management of families with genetic disease. It has to be remembered, however, that there have been a number of notable developments since the publication of this book, particularly with regard to the detection of inborn errors of metabolism and congenital CNS malformations.

ALAN E. H. EMERY


Dr Ford’s account of human chromosome cytology is aimed at... the student of medicine, undergraduate or postgraduate, and it is well aimed. The book is informative without being unduly detailed and the balance of subject matter should appeal to those who wish to acquire a sound working knowledge of the subject without becoming too involved in technical complexities. Unfortunately the publication only just caught the flurry of recent activity following the introduction of chromosome banding techniques. Although mention is made of quinacrine and Giemsa banding, it is only of the very beginnings of the publication explosion based on these phenomena.

A brief historical introduction is followed by chapters covering the cell cycle, chromosome morphology, meiosis, and the principal techniques used in the study of chromosomes. Reflecting the author’s own interests, the sections on meiosis and on the ultrastructure of chromosomes are particularly full and well presented. There is a very simple but quite adequate account of nucleic acid chemistry and molecular genetics.

Having established the basic biology of the subject, the second half of the book deals consecutively with the origin and transmission of chromosome abnormalities, incidence of abnormalities in neonates and embryos, abnormalities of sex chromosomes and of autosomes. A separate chapter entitled ‘Skin, brain and the chromosomes’ accepts the fact that almost all chromosomal anomalies are associated with unusual dermatoglyphics and varying degrees of CNS malfunction. Chromosome breakage (spontaneous and induced) and neoplasia are briefly covered and a penultimate chapter then considers the achievements, and speculates on the future, of human cytogenetics. The conclusion, that in purely medical terms the efforts which have gone into the subject have provided rather poor returns, is rather more gloomy than many of those in the field would easily accept, but the case is reasonably argued and raises a whole series of important issues for the consideration of those new to the subject. A final few pages, from Professor J. H. Renwick, summarize knowledge of human gene linkage and assignments of gene loci to particular chromosomes. There are a series of short addenda to the various chapters, an author index, and a good subject index. Selected references are given at the end of each chapter.

The book contains a few errors of note, such as the statement that the cri-du-chat syndrome is due to an abnormality of chromosome 4 (p. 165). The Xg blood group is quite erroneously referred to as ‘haemoglobin Xg’ (pp. 149, 187, 191), which is particularly misleading as it would suggest to the student that a haemoglobin variant is determined by an X-linked locus. The statement (p. 207) that the testicular feminization syndrome is probably inherited as an X-linked dominant would not be easy to justify. These, and a few other inaccuracies, make a generally precise book.