This edition contains 1142 entries for which the mode of inheritance is certain; an increase of 276 since the 3rd edition which was published in 1971. The number of autosomal recessive phenotypes remains about 20% lower than the number of autosomal dominant entries though, as the author admits, the decision as to whether some autosomal phenotypes are dominant or recessive is often somewhat arbitrary. In addition to the asterisked phenotypes there are another 1194 without asterisks, making a total of 2336. By extrapolating from data on micro-organisms and drosophila the author estimates the expected final number of loci in the human to be near 100,000. The rate of increase of loci in the 9 years (4 editions) since 1966 has remained linear with no acceleration or deceleration.

It is interesting in this edition to see new examples of dominant phenotypes for which the causative mechanisms have been worked out; for example the increased activity of phosphoribosyl pyrophosphate synthetase in some cases of gout and the elegant mechanism which has been postulated for familial hyperbetalipoproteinaemia.

The 'nosologic tables' in the first section of the book have been increased in number since the last edition, and they promise to be particularly helpful in the diagnostic process which is so important in genetic counselling. New tables are given for dental anomalies, sexual abnormalities, and the Ehlers-Danlos syndromes, the tables of skeletal dysplasias and hand malformations are 'updated', and the table on hereditary deafness is discontinued. One small error, perhaps worth mentioning, is that while the table of Ehlers-Danlos syndromes gives the mode of inheritance of type IV as autosomal recessive, the catalogues give it as autosomal dominant and the basic defect, given in the table as deficiency in synthesis of type III collagen, is not mentioned or referenced in the text. New tables are also provided of the ethnic distribution of genetic disease and of allelic series.

Of particular interest is the much expanded section on the human gene map which contains mapping information on at least 105 loci. If the human 'cartographers' continue with their present success one wonders how long it will be before the catalogues are no longer 3 in number but 23 (or 24 if the Y chromosome is included!).

This book is essential to all those giving genetic advice to patients, and it should also be available in all medical libraries.

D. C. SIGGERS


Prenatal diagnosis and the selective abortion of fetuses severely handicapped by such disorders as mongolism, spina bifida, and certain inborn errors, are now available to parents at risk on a limited scale in almost every medical teaching centre in Britain. The introduction and rapid development of prenatal diagnosis in recent years has undoubtedly been one of the most important contributions to medical practice made so far by medical geneticists. It is not surprising that there has been considerable public interest and controversy, particularly over the ethical issues involved.

Professor Harry Harris's thoughtful and timely monograph is to be welcomed on several counts. First, because it gives an excellent up-to-date account of the techniques and scope of prenatal diagnosis in pregnancies at risk of chromosome aberrations, X-linked disease, autosomal recessive metabolic diseases, and open neural tube malformations. Each category is well illustrated with examples (these include an arresting, albeit hypothetical, revision of the Royal houses of Europe, based on the assumption that prenatal sex determination was available to the descendants of Queen Victoria and that sons at risk of being affected with haemophilia had been aborted). Secondly, the monograph is to be welcomed because it discusses in a straightforward manner the long-term implications of prenatal diagnosis, to what extent widespread use of the procedure can be expected to reduce severe handicap among the liveborn, and how it must result in an increase in the number of carriers of X-linked and autosomal recessive diseases among the normal offspring of families in whom abnormal progeny have previously been aborted. The overall effect on the gene frequency in the population is judged to be small for X-linked disorders, and barely worth consideration for autosomal recessive disorders.

The third and perhaps most useful part of the monograph is to be found in the last quarter of the text which deals with the ethical question. All the important arguments are here, thoughtfully considered, emphasizing the responsibility borne by those who offer prenatal diagnosis to their patients. Professor Harris takes us down the slippery slope from selective abortion to selective infanticide, for similar arguments have been used to justify or condemn both procedures. He asks us how severe must the handicap be to warrant termination. What does one recommend when a fetus is found by chance to have a 47,XY karyotype? Should prenatal sex determination be used to allow parents to choose the sex of their children? (It is understood that this is currently the practice in certain parts of India.) Will the widespread use of prenatal diagnosis change our attitude towards handicapped children who have escaped selective abortion? It is good to see these and other questions properly aired, if only to make sure that we continue to use prenatal diagnosis in a responsible fashion.

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