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BOOK REVIEWS

EARLY DIAGNOSIS OF FETAL DEFECTS. D. J. H. Brock. Churchill Livingstone, Edinburgh. Pp. xi + 165. Price: £7.95. P/B.

Although primarily intended for obstetricians, this short, very readable book will appeal to anyone interested in the prevention of human abnormalities.

Following a general account of amniocentesis and its risks, techniques for the detection of chromosome disorders, neural tube defects and mendelian disorders are described. The author gives a very careful discussion of the limitations of these techniques, including in his discussion a list of technical errors which have occurred in practice. There follows a chapter on antenatal screening. Here the author warns against excessive hopes for programmes of screening. For example, for alphafetoprotein serum screening, he gives figures showing that, in view of the limited sensitivity of the test and limited take-up of the service, the maximum reduction in frequency of spina bifida at birth would be about 60 per cent. The book concludes with a brief chapter on future developments.

Something of the flavour of the book is captured in the author's following calculation (given here in slightly rephrased form). Suppose, as sometimes happens, that an amniotic fluid alphafetoprotein analysis is performed on a pregnant mother with only normal risk of bearing a child with a neural tube defect; in England this risk is 3/1000. The probability that the procedure will identify a fetus with a neural tube defect is about 0·98, whereas the probability that a healthy fetus is misclassified as defective is about 0·005. Then the probability that a fetus, diagnosed as abnormal, is actually defective is only about

$$\frac{0{\cdot}003 \times 0{\cdot}98}{0{\cdot}003 \times 0{\cdot}98 + 0{\cdot}997 \times 0{\cdot}005} = 0{\cdot}37$$

(would that textbooks on statistics included such a fascinating application of Bayes' theorem). Clearly, the alphafetoprotein test, although apparently very reliable, is not good enough in the present context; the author concludes that in such cases the performance of the new acetylcholinesterase test, in addition to the older test, is essential.

Readers will probably be particularly interested in Dr. Brock's views on alternatives to amniocentesis. He gives a cautious welcome to the use of ultrasound for the detection of neural tube defects. While pointing out the enormous variation in skill among individual operators (he says that, while some can diagnose quite small spina bifida lesions, others have difficulty in recognising twin pregnancies) he also states: "where the experts have already gone the less experienced must surely follow". On the vexed question of the effectiveness of vitamin supplements to the mother in preventing neural tube defects, he writes what must surely be one of the most cautious sentences ever to appear in the scientific literature: "though this study had several major design faults its results were sufficiently striking

to suggest that at least among some women vitamin deficiency in the early weeks of pregnancy might be an important factor". Such sceptical comments are a pleasure to read. As Fisher once remarked: 'in *The Design of Experiments* the word *randomisation* did not appear by chance".

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GENETIC COUNSELLING. W. Furhrmann and F. Vogel. Springer-Verlag, 3rd Edition 1983. Pp. x+188. Price: $16.80 P/B.

The authors chose to introduce this book by employing shock tactics, as the reader is confronted on the very first page with a complicated counselling problem featuring no less than three types of diseases segregating within one family, accompanied by the rhetorical question "Should my daughter marry this man?" Despite the reassuring and inevitable outcome of this genetic riddle being "Why not?", the authors do suggest that the untrained reader start at Chapter 2. As Chapter 1 continues with a detailed but out of place list of the mucopolysaccharidoses he is tempted to do just that! A description of genetic counselling and its aims is unfortunately missing. However, the end of Chapter 1 combined with a short Chapter 2 together detail some of the more general principles of counselling with advice on how to obtain and perhaps more importantly how to construct a pedigree.

The next few chapters are devoted to the delineation of the Mendelian laws of heredity in the context of autosomal dominant, autosomal recessive and X-linked human disease. More emphasis could have been placed on the more knotty problems such as penetrance and expressivity in autosomal dominant disease as these are complicating factors within particular pedigrees. More information actually emerges when somatic mutation is discussed with especial reference to retinoblastoma. The Chapter on autosomal recessive disease hardly mentions the metabolic diseases at all, and certainly does not stress the enzyme/biochemical nature of many of these conditions. As a consequence no emphasis was placed on the practical importance of storage of material from affected individuals in order to aid subsequent accurate diagnosis. As a contrast the second half of this chapter, which is devoted to heterozygote testing, is much more detailed and comprehensive. Although this will act as a guide when other family members seek advice, fearing that they may be carriers, the authors do not emphasise that the risk is exceptionally low except in the case of X-linked recessive disease, as both types appear in a single table.

Bayesian risk calculations are very important for the modification of empirical risks and make for better counselling and more accurate prognosis but this Chapter is not intended for the novice as the mathematics are daunting despite the clever use of Christian names to make identification with the problems easier.

A description of chromosomal aberration not unnaturally places its main emphasis on Downs Syndrome with an excellent description of how and how often this condition arises. However, the instruction to compare Tables 8:1 and 8:2 cannot be achieved without the aid of a pocket calculator as one details frequencies and the other percentages. No self-