We report here a female patient with Williams syndrome who showed typical dysmorphic features, a characteristic personality, and a bilateral renal artery stenosis. This patient, however, showed no clinical translocation involving chromosomes 1 and 18: 46,X,Y(1)(q11.2)(q12;q23). Thus the breakpoint of chromosome 18 in band q23 is in the same band as in the patient reported by Colley et al. The father of the presently reported patient carries the same balanced translocation and is apparently healthy. Studies of this chromosome region at DNA level are currently being performed.

We conclude that there are now two patients with Williams syndrome showing a chromosomal rearrangement involving 18q23. On one hand a gene defect at a locus at 18q23 might lead to a phenoctopy of Wil- liams syndrome. Alternatively, patients with the Williams syndrome not showing any chromosomal abnormality might have a submicroscopic deletion or a mutation located at 18q23, or an anatomico genomic imprinting might be involved.

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BOOK REVIEWS

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It is curious, when the Human Genome Project engages the energies of so very many scientists, that there is only this one book on human linkage analysis. Perhaps Ott is so pre-eminent in this field that nobody else would presume to compete. The field Ott addresses is, however, quite narrow. He is concerned with maximising the output of chromosomal linkage analysis programs such as LIPED or LINKAGE. He does not discuss the interplay of genetic and physical (especially FISH) methods, or of human and mouse genetics, which underpin the construction of the map of the human gene map. He does not even give much space to the various forms of extended sib-pair analysis or to exclusion mapping or mosaicism. His aim is to help people produce valid lod scores for mendelian characters.

Guidance in this area is extremely wel- come. It is relatively easy to master setting up the input files for LIPED or MLINK: after a few crashes and some hair tearing, most of us can get the computer to churn out lod scores. The real problem is to know what the scores mean. What is a valid penetration to use on a sample of families showing appar- ently dominant schizophrenia? How does one set up the liability classes for fragile X or autosomal dominant conditions or at random threshold lod score to aim for if a collection of disease families is tested for 50 random markers? If you read, learn, and inwardly digest Ott’s book you will be able to think through such problems (though I still don’t agree with him about multiple markers). He offers a wealth of useful formulae and references, and draws on his unrivalled ex- perience to discuss innumerable practical issues encountered in reducing a genetic problem to a lod score.

Compared to the first edition, this revised version puts much more stress on multilocus mapping and on the practical aspects of locus heterogeneity. There is much useful discussion of locus ordering and of the meaning of multi- locus lod scores. It is a pity that no reference is made to some fundamental arguments about the validity of multilocus mapping. When the man who set this whole industry in motion has written a paper titled ‘Multipoint mapping and the Emperor’s Clothes’ (Mor- ton NE. Ann Hum Genet 1990;52:309-18), a little discussion would have been in order. Believable multilocus maps usually depend not just on lod scores but on results with chromosomal breakpoint in situ hybridisation, and pulsed field mapping. If there is a weakness in the book, it is that Ott concentrates on statistical approaches, even when molecular methods can give a surer answer. But of course that is not the book about: likelihoods, not biochemistry.

It is not always easy reading. Ott’s motto might well be “Listen carefully, I shall say this only once”. He doesn’t waste words, and many sections require close reading. Pro- blems, some quite difficult but with solutions provided, help. The mathematics of human linkage analysis can be formidable. Ott gives plenty of mathematical formulae, but always concentrates on what they mean. Probably very few of us have much idea how the Elston-Stewart algorithm works, or why it is so clever. This doesn’t matter – you can drive a car safely without knowing anything about tappets, just as long as you know your Highway Code. Ott provides a Highway Code for linkage projects. It is a valuable and indispens- able book, and everybody who generates or uses lod scores needs a copy. But they must remember that this is only one facet of human gene mapping.

ANDREW P READ


Practical work is an essential component of any genetics course and thus it is surprising to find few books devoted to this area. The authors aim to help rectify this deficiency with Practical Genetics which is aimed at school/college/university undergraduate courses in genetics. It is based on the authors’ teaching experience and includes chromosome analysis in mitosis and meiosis, Drosophila breeding experiments, complementation of yeast strains, meiotic analysis in Sordaria and Aspergillus fungi, and population genetics studies on white clover. Each set of experi- ments is easy to follow and well illustrated and there is a particularly useful emphasis on the interpretation of results and observa- tions. The authors claim that each experi- ment is known to work well repeatedly and to be within the means of a modestly equipped teaching laboratory.

In contrast, practical work in human gen- etics has marked limitations and the authors emphasise the hazards of handling blood samples, the need to avoid both family studies which might reveal non-paternity, and chromosome analysis on students. Overall the experiments were well chosen to illus- trate classic genetic principles but the lack of emphasis on molecular genetics was surpris- ing given the central role of this field for genetics and other biological sciences. This might in part be explained by the desire to contain costs for ‘wet’ practicals and this might be circumvented in a future edition by the inclusion of analysis of data sets (linkage analysis using DNA polymorphisms, popu- lation genetic studies of DLA polymorphisms, DNA sequence analysis, etc).

J M CONNOR


This is a new edition of a well respected textbook which was written for undergraduate courses in genetics. Each chapter has been revised and expanded with new infor- mation on gene structure and function (es- pecially in relation to development) and new chapters on transposable elements and meth- odologies of genetic engineering. The text is clear and accurate and the multicolour illus- trations are excellent aids to understanding. Key points and principles are highlighted in the text and each chapter is followed by a summary and sets of problems.

Some aspects of human genetics are in- cluded but, surprisingly, other areas with general implications for genetics such as un- stable mutations, imprinting, tumour sup- pressor genes, microsatellite repeats, and mitochon- drial heteroplasmy are not. The sections on gene therapy and diagnosis of genetic disease by linkage with RFLPs and mutational analysis could also be expanded to help emphasise the authors’ goal of indi- catino the general population relevance of genetics.