We report here a female patient with Williams syndrome who showed typical dysmorphic features, a characteristic personality, and bilateral renal artery stenosis. This patient has an apparently balanced reciprocal translocation involving chromosomes 1 and 18: 46,XX,t(1;18)(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.*⁵ The father of the present 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 phenocopy of Williams 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 anomaly of genomic imprinting might be involved.

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BOOK REVIEWS

All titles reviewed here are available from the BMJ Bookshop, PO Box 295, London WC1H 9TE. Prices include postage in the UK and for members of the British Forces Overseas, but overseas customers should add 15% to the value of the order for postage and packing. Payment can be made by cheque in sterling drawn on a UK bank, or by credit card (Mastercard, Visa, or American Express) stating card number, expiry date, and full name.

Analysis of Human Genetic Linkage. Jurg Ott. (Pp 302; \$34.00.) Baltimore: Johns Hopkins University Press. 1991.

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 strictly concerned with human linkage analysis program packages 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 underpins the construction 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 to mosaicism. His aim is to help people produce valid lod scores for mendelian characters.

Guidance in this area is extremely welcome. 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 penetrance to use on a sample of families showing apparently dominant schizophrenia? How does one set up the liability classes for fragile X or adenomatous polyposis? What is a reasonable 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 experience 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 problem of locus heterogeneity. There is much useful discussion of locus ordering and of the meaning of multilocus 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' (Morton NE. *Ann Hum Genet* 1988;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 breakpoints, 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 what the book is 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. Problems, 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 unique and indispensable 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 Genetics. R N Jones, G K Rickards. (Pp 228; £13.50.) Milton Keynes: Open University Press. 1991.

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 experiments is easy to follow and well illustrated and there is a particularly useful emphasis on the interpretation of results and observations. The authors claim that each experiment is known to work well repeatedly and to be within the means of a modestly equipped teaching laboratory.

In contrast, practical work in human genetics 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 illustrate classic genetic principles but the lack of emphasis on molecular genetics was surprising 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, population genetic studies of DNA polymorphisms, DNA sequence analysis, etc).

J M CONNOR

Principles of Genetics. 8th edition. E J Gardner, M J Simmons, D P Snustad. (Pp 649; £21.50.) New York: John Wiley. 1991.

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 information on gene structure and function (especially in relation to development) and new chapters on transposable elements and methodologies of genetic engineering. The text is clear and accurate and the multicolour illustrations 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 included but, surprisingly, other areas with general implications for genetics such as unstable mutations, imprinting, tumour suppressor genes, microsatellite repeats, and mitochondrial 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 indicating the general population relevance of genetics.

J M CONNOR