syndrome (PWS) at 1 in 10 000, comparable to the frequency of phenylketonuria, and PWS is certainly the commonest chromosomal deletion syndrome. The hypotonia and failure to thrive as a neonate, the hyperphagia and obesity from childhood onwards, the hypogonadism, and the learning difficulties pose many problems in terms of pathogenesis, diagnosis, and management, making the syndrome an appropriate subject for a monograph. The editors emphasise that the “book focuses on selected research and management issues related to Prader-Willi syndrome. It is not intended to be a comprehensive review of the syndrome, nor a ‘how to’ book on treatment and management”. However, the book is written with clinical practice in mind and also assumes little previous knowledge of the various subjects discussed. It is not just a collection of research papers.

It starts with a first rate overview of the diagnosis, characteristics, and management of PWS by Luisselli et al, followed by an excellent discussion of the clinical implications of the chromosome 15 abnormalities that occur in about 60% of cases by Ledbetter and Cassidy. They put the recurrence risk (in the absence of a familial translocation) as <0-1%. Taylor then gives a good review of the cognitive and behavioural characteristics, including a useful discussion of food related behaviour. He draws attention to the discrepancy between earlier reports in which up to 97% of people with PWS were mentally retarded and a more recent study of his with Caldwell in which they “administered the Weschler Adult Intelligence Scale to 12 individuals with the syndrome and found 50% had IQs outside the retarded range (average IQ = 70-25)”. While he does discuss possible explanations he does not consider ascertainment bias, which may well account for both extremes. (If his referencing is correct, his study was actually on 11 subjects who came from five states to participate in a five week residential summer programme.)

Chapter 4 is a useful short account of the management of the problems of hypotonia, developmental delay, and feeding problems in infancy by Cassidy. I found chapter 5 a less useful, long account of behaviour management and intervention.

A lot of space is taken up explaining the technical jargon of behaviour analysis and therapy, which is not very relevant to a description of five studies of weight loss and food stealing involving a total of 10 patients with PWS. In six subjects the weight loss was not maintained and Luisselli himself emphasises the need for “deliberate programming in extra therapy settings”. His conclusion that “clearly, behaviour methods have much to offer in designing comprehensive care programs for the individual with PWS” is perhaps a little too optimistic. The more direct approach of surgery for obesity control is discussed by Wagner in his account of surgical considerations in PWS. The final chapter summarises the responses of 12 families to a questionnaire and provides no great surprises.

All in all this is a useful, well referenced monograph. I was disappointed in the lack of research into the neuroendocrine basis of the hyperphagia, the understanding of which might allow more rational intervention. This is a book for most clinical medical libraries and for those with a special interest in this puzzling syndrome.

M Pembrey

Genetics and Alzheimer's Disease

This volume contains the proceedings of a meeting held in Paris, in March 1988, by the Fondation Ipsen pour la Recherche Therapeutique. In recent years work on genetics, and particularly molecular genetics, has come to the forefront of research into Alzheimer's disease (AD). The contents of this book reflect this situation and chapters have been contributed by many of the leading groups in this field.

Molecular genetic studies of AD can be divided into two groups. First, there have been a number of RFLP linkage studies of families multiply affected by AD. By March 1988 there had been one report suggesting linkage between AD and markers on chromosome 21 from St George-Hyslop and colleagues. This group has contributed a thoughtful chapter to the present volume considering some of the methodological problems faced by such research, which may account for negative findings such as those reported in the chapter by Percik-Vance and colleagues. The role of genes on chromosome 21 in AD is also dealt with in the two chapters that consider the link between Down's syndrome and AD. The issues are clearly discussed by Epstein who also describes work on trisomic and transgenic mice. Such experimental models are likely to become increasingly important as assay systems for genes of possible importance in the aetiology of AD, and will subsequently play a crucial role in studies of pathogenic mechanisms and therapeutic strategies.

The second line of research into the molecular genetics of AD has focused upon the pathological structures found in the brains of sufferers. A number of chapters in this volume describe recent
studies of the amyloid A4 protein precursor (APP) gene. Although this is also located on chromosome 21, it does not contain the primary genetic defect in familial AD. However, the abnormal deposition of this protein is clearly a prominent feature of the neuropathology of AD. Studies of the several different forms of mRNA coded for by this gene and its regulation are described in a number of chapters. This work is likely to lead to an increased understanding of the pathogenesis of AD. However, a major unresolved issue remains that of how early in the chain of pathogenic events amyloid deposition is located.

Overall this book is of a higher standard than is usually encountered in conference proceedings and is exceedingly well presented. It will be of value to those working in this area but most of the chapters are probably too technical for those less well acquainted with the subject.

M J Owen

Practical Genetic Counselling

It is good to see the third edition of this book, which is unique in its comprehensive cover of most of the genetic problems encountered in clinical practice. It contains major changes and additions. There is a new chapter on congenital malformation syndromes and dysmorphology, while the rapidly advancing area of molecular genetics is covered in a much revised chapter.

The book is organised in three parts. Part 1 covers general aspects of genetic counselling in its broadest sense. The basics of mendelian, chromosomal, and molecular genetics are well described, as are the applications to carrier detection, prenatal diagnosis, risk determination, and segregation analysis. The final chapter in this section is devoted to ‘The genetic counselling clinic’ and reflects the author’s great experience and practical approach and will be very valuable to those who are interested in setting up or understanding clinics of this sort.

Part 2 deals systematically with specific organ systems and their inherited disorders and has excellent references and sources of further information.

Part 3 deals with ‘Genetic counselling and society’. There is much valuable information in an appendix on sources of information, including the addresses of genetic centres in Britain.

This book is probably not designed for clinical geneticists, although I suspect that many always have a copy near by. Its main attraction will be for perceptive clinicians in other specialties who realise how profoundly genetics is impinging on their practice. It is possible to dip into this book and find something about almost any genetic disorder, while the general chapters provide an excellent revision course for those whose clinical genetics is either very rusty or non-existent.

I suppose there is a danger that this book will encourage people to believe that they can cope with any genetic problem, but as the general awareness of the importance of genetics increases, most clinicians will appreciate that there are many pitfalls, not least of which are the problems created by genetic heterogeneity and the need for precise diagnosis of rare disorders.

The style and presentation of the book are very attractive although even this paragon among books contains occasional errors. However, one has to search hard for even minor mistakes, for example, figure 5.5 (a) is not a fully informative pedigree.

This book is without doubt one of the most useful currently available. It can be thoroughly recommended to any clinician or scientist interested in this rapidly advancing field.

Rodney Harris

Advances in Human Genetics

As in previous volumes in this series, five review articles are presented, one largely clinical, one largely cytogenetic, and three which straddle the interface of molecular biology and clinical medicine.

In a clear and concise update of HLA disease association Bell, Todd, and McDevitt describe the structure and function of the major histocompatibility complex (MHC). The chapter concentrates on the class II region, with which the strongest disease associations have been found, associations which are being refined through sequencing of the class II loci. Although other factors, both genetic and environmental, play a role in the genesis of the diseases showing HLA association, unravelling the molecular basis of the associations will provide insight into a key step in their aetiopathology. Furthermore, the hope expressed by the authors, that peptide-MHC interactions underlying these diseases might in the future be modified directly, does not seem too remote.

Cohen and Levy review the major chromosome
Genetics and Alzheimer's Disease

M J Owen

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