

*Chromosome Abnormalities and Genetic Counselling.*

By R. J. M. GARDNER and G. R. SUTHERLAND. Oxford Monographs in Medical Genetics No. 17. Oxford University Press. 1989. 255 pages. £24.00. ISBN 0 19 504932 2.

Over the last 30 years cytogenetics has played a cardinal role in genetic counselling. The technology has established the cause of many congenital abnormalities and has provided the means of diagnosing such disorders *in utero*. Down's syndrome and the fragile X syndrome still remain the most frequent indications for these studies, but the recent introduction of various staining and banding techniques has increased the range of conditions in which microscopic chromosomal abnormalities can be demonstrated. The latter now includes, for example, several so-called 'microdeletion' syndromes (retinoblastoma, aniridia-Wilms complex, Prader-Willi syndrome, etc.). This book is addressed primarily to genetic counsellors: those who have to explain and interpret a chromosomal problem to the individual or family in whom it has occurred. Though at pains to explain the theoretical basis of such abnormalities, the authors present an essentially practical approach. Both are internationally recognized experts in the field, and they therefore write with authority. Sutherland in fact was responsible for 'rediscovering' the fragile X in the late 1970s, the original observation of Lubs in 1969 remaining unconfirmed for several years.

The text is divided into 5 sections: basic concepts, the parent with a chromosomal abnormality, normal parents with a chromosomally abnormal child, reproductive failure, and finally prenatal diagnosis. A great deal of useful information is summarized in tabular form and the text is neatly divided into relevant sections. There is a very extensive bibliography.

I would recommend this book unreservedly to anyone involved in counselling and managing a family with a chromosomal abnormality as being both scholarly and practical.

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*Genetics of Neuropsychiatric Diseases.* Edited by L. WETTERBERG. London: Macmillan Press. 1989. 363 pages. £50.00. ISBN 0 935859 65 9.

After the flurry of activity over the last few years into mapping and isolating gene loci for rare genetic diseases, the attention of molecular biologists is now beginning to turn to common disorders. The problems are more intractable, not least that of identifying 'candidate' genes, but the rewards are greater. This is nowhere more evident than in the case of the common neuropsychiatric disorders. The current text, which represents the edited proceedings of an international

meeting held in Sweden in 1988, addresses recent research developments into this group of diseases.

It is divided into four sections: research methods, research models, genetic studies specifically in neuropsychiatric disorders and possibilities for future research. Particularly valuable in the first section are summary tables of the locations of gene loci for important neuroreceptors and peptides, and outlines of the technologies used in molecular studies (DNA methods and linkage analysis). The applications of these techniques are illustrated in several well-researched disorders: phenylketonuria, the fragile X syndrome, and Huntington's chorea. However, to me the most fascinating discussions in the book centre on Tourette syndrome, affective disorders, schizophrenia, Alzheimer's disease, narcolepsy and alcoholism.

Tourette syndrome is a bizarre condition characterized by tics and a wide range of compulsive behaviour including grunting and obscene outbursts. It is inherited as an autosomal semi-dominant trait which affects perhaps as many as 1 in 200 children. Blood serotonin and tryptophan levels are low, but the basic biochemical defect is unknown and the responsible gene has not yet been identified. Clearly an understanding of the molecular pathology of the disorder might well throw light on our understanding of related behaviour patterns in otherwise normal individuals.

Gene loci for bipolar affective disorders (on the X chromosome and chromosome 11) and schizophrenia (on chromosome 5) have been identified in some rare affected families but not in others. Perhaps this is a reflection of genetic heterogeneity. Alzheimer's disease also now seems to be heterogeneous, with linkage to markers on chromosome 21 only being evident in some families.

Narcolepsy is a rare autosomal dominant disorder characterized by a periodic uncontrollable tendency to fall asleep. The cause is unknown, but almost all affected individuals type as HLA DR2, which must be of pathogenetic significance but in what way remains a mystery. Again our understanding of this rare disorder might help us understand more of normal behaviour.

Finally a detailed and critical review of family, twin and adoption studies clearly indicates that alcoholism is familial: 'a significant proportion of which can be attributed to genetic factors'. But the nature of these factors is still unknown. They may reside in individual variations in the metabolism of alcohol (alcohol dehydrogenase and acetaldehyde dehydrogenase) or in the effects of alcohol on the brain. However, in contrast to nicotine, opiates and catecholamines, no specific alcohol receptor has so far been found in the central nervous system.

It has been estimated that possibly 10% of our 100000 or so genes may be specific to the brain. Their identification and characterization is a mammoth task, but this text clearly indicates that a start has