
Before the second world war the statistical content of general medical publications was minimal. Over the course of half a century there has been a dramatic change in presentation. There is now the expectation that results will be presented in statistical language, which has generally come to mean significance testing.

During the 1980s it has repeatedly been pointed out by medical statisticians that significance testing does not usually provide the ideal framework for expressing results. Many medical journals, including the British Medical Journal and the Lancet, now favour a confidence interval approach when this is appropriate.

Statistics with Confidence is a handbook rather than a textbook. It is based on a series of articles by several medical statisticians, originally published in the British Medical Journal. The reasons for preferring a confidence interval approach are explained thoroughly. The methods of construction of confidence intervals for the most frequently encountered situations are set out clearly and are accompanied by simple worked examples. The methods included in the book do not necessitate the use of a computer, although many of the calculations can be performed by statistical packages such as Minitab. Furthermore, the authors have made available a program called CIA which can perform the calculations. There are also sections on statistical guidelines for authors and on check lists, and tables required for the calculations described in the book.

The book can be thoroughly recommended to a general medical readership. Medical genetics publications include straightforward descriptive and comparative material, to which the methods described in the book apply directly. However, the book would not provide a great deal of specific help to those who wish to assign confidence intervals to the parameters specific to the highly developed, specialised, statistical methodology of medical genetics. Moreover, in the genetic counselling situation, in which a proband is assigned a point estimate of his/her risk of carrying or transmitting a gene, it is not usually appropriate to give a confidence interval as well: the risk itself, a number strictly between 0 and 1, expresses the uncertainty aspect as well as the probability. Nevertheless, when advances in the understanding of human DNA at the molecular level lead to the development of potentially effective corrective treatments for inherited disorders, medical geneticists will need to evaluate these treatments in the same way that other drugs are evaluated. When this situation is reached, this little book will assume as great an importance for medical genetics as for other specialities.


This is an account of a workshop held in Berlin immediately following the 7th International Congress of Human Genetics in 1986. It includes information on the prevalence of epilepsy, the inheritance of EEG patterns, the definitions of epileptic syndromes, and accounts of genetic studies. Epilepsy as defined by 'recurrent unprovoked seizures' has a lifetime cumulative incidence of about 4%, but there are two peaks of incidence, one before 20 years and one after 60 years. The aetiology is likely to be different for the two age groups; the epilepsy that families are concerned about is that which occurs in childhood, with an incidence in the population of about 2%. However, the useful chapter here on incidence and prevalence by Hauser and Annegers is marred by the absence of a reference list.

The debate on the genetic basis of the epileptic syndromes centres on a rather artificial distinction as to whether an autosomal dominant gene with modifiers is mainly responsible, or whether the aetiology is polygenic. EEG patterns appear to be dominantly inherited, and many are age dependent. There is a useful discussion by Greenberg et al on whether two genes (one dominant and the other recessive) are responsible for juvenile myoclonic epilepsy; their models can be useful for looking at other disorders. The study on the genetics of partial epilepsies was disappointing as the number of affected relatives out of the total was not given, but was instead described by the number of index patients who had a positive family history. In general, the empirical recurrence risks for sibs of index patients with most types of epilepsy lies between 4 and 8%, although a few types (such as febrile convulsions and the progressive myoclonic epilepsies) have a higher risk. A useful source for genetic risks in epilepsy is a review article by Blandford et al in Human Genetics (1987;76:303), which is not even referred to in this book.

The four chapters on recurrence of epilepsy in the offspring of patients will be useful to clinical geneticists. Three of these studies come from clinics at Heidelberg and Berlin, where adult patients with children were enrolled in a prospective follow up between 1969 and 1982; the observations of epilepsy or EEG abnormalities or both in children have been correlated with type and onset of seizure in, and with sex of, the affected parent. The overall risk to offspring is 4 to 5%, and this is nonsignificantly greater for the offspring of affected females, and is also greater if the parent had generalised epilepsy, particularly if associated with absences. Useful data are being collected through these prospective studies on the occurrence of epilepsy, and any triggering
mechanism, in young people who have generalised spikes and waves on their EEGs.

This book is interesting and will be a useful addition to any library. However, as it arises from a workshop it does not deal comprehensively with the subject of epilepsy and is therefore rather a luxury. It gives the impression that epilepsy is difficult to classify and that a predisposition to it is difficult to measure. Further advances in understanding the genetic basis of epilepsy await the finding of some specific predisposing markers, other than EEG patterns.

SARAH BUNDEY

NOTICES

2nd European Meeting on Psychosocial Aspects of Genetics

This meeting will be held in Leuven, Belgium, on 24 to 26 September 1990. Topics will include: Psychological and social aspects of genetic counselling, prenatal diagnosis, carrier identification, and predictive testing; Psychological profile of patients with specific genetic diseases; Follow up studies on the impact of genetic diseases; and genetic risk perception and decision making. For further information, contact Dr Gerry Evers-Kiebooms, Centre for Human Genetics, Herestraat 49, B-300 Leuven, Belgium. Tel: 00-32-16-21.58.67.

Bloom’s Syndrome Registry

The Bloom's Syndrome Registry has accumulated genetic and clinical information concerning bona fide instances of Bloom’s syndrome (BS) since the early 1960s. Almost 150 persons with BS have been accessioned to the Registry, probably the vast majority of those ever diagnosed anywhere in the world. Progress reports from the Registry are published periodically (see Clinical Genetics 1989;35:57-69). Besides defining this very rare clinical syndrome, this project has documented the enormous cancer predisposition of affected homozygotes, and has shown that the cancers are of diverse types and sites. Samples of serum, erythrocytes, lymphocytes, and various sources of DNA from affected families have been collected and are stored frozen at The New York Blood Center, as are cultured cell lines.

The accession of new cases to the Registry will cease as of 1 January, 1991. Thereafter, the families in the Registry as of that date will be followed, as in the past, either through the referring physician/geneticist or with the family directly. In order to make the cohort of affected persons to be followed as large as possible, the Registry requests information during 1990 about any persons known to have BS but who are not already in the Registry. (Note. If confirmation of the diagnosis is required, blood samples from patients will be accepted without charge for sister chromatid exchange (SCE) analysis. Please contact the following: Laboratory of Human Genetics, The New York Blood Center, 310 East 67th Street, New York, NY 10021, USA. Tel: 212/570-3075; Fax 212/570-3195.)

Workshop on ‘Towards the identification of the CF gene: progress and strategies’

The International School of Pediatric Sciences of the Istituto G Gaslini and the Fondazione Menarini announce a Workshop on 'Towards the identification of the CF gene: progress and strategies', 9 to 11 April 1990, at Sestri Levante, Genoa, Italy. The Workshop, directed by Professor Romeo (Genoa), Dr Devoto (Genoa), Dr Galietta (Genoa), Dr Rugolo (Bologna), and Professor Greger (Freiburg), will consist of morning lectures, afternoon lectures, and general discussion. The second afternoon consists of a poster session. Applications (CV, letter of presentation) will be considered until the course is full. The official language will be English and 100 participants will be admitted. Inquiries to Silvia D'Angostino, Secretary of the Workshop, Lab Genetica Molecolare, Istituto G Gaslini, 16148 Genoa, Italy. Tel: +39 (10) 5636370/400; Fax 391254.

Some notes for contributors on nomenclature

Nomenclature. Authors should refer to the following publications.


Genetics of the Epilepsies

Sarah Bundey

*J Med Genet* 1990 27: 71-72
doi: 10.1136/jmg.27.1.71-a

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