LETTER TO THE EDITOR

Prenatal exclusion testing for Huntington's disease

Brock *et al*[1] advocate the use of common sense rather than sticking rigidly to the rules, a position I would normally applaud, but not in the context of prenatal exclusion testing for Huntington's disease. If it is common sense to give people good news they have not asked for because it would save CVS and unnecessary terminations, then it must follow that it is also common sense to give people bad news they have not asked for. If someone *knows* they carry the gene then this will have an effect on their plans to have children and we would be depriving them of the chance to make a properly informed decision. If people have decided not to make use of available information on themselves, then this decision must be respected. It is arrogant and dangerous for any of us to think that we know what is best for them, however tempting that may be. Have Brock *et al*[1] taken into consideration the fact that some people have bad reactions to good news, or feel they would lose some advantages by no longer being at risk? It may be unavoidable in some cases for clinicians to know more than their patients, especially where other family members are coming forward for testing, but at other times they could ensure that the laboratory only gives them the information they ask for, and this could save a number of ethical dilemmas.

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1 Brock DJH, Curtis A, Mennie M, Raeburn JA. Options for prenatal testing for Huntington's disease using linked DNA probes. *J Med Genet* 1990;27:68–9.

BOOK REVIEWS

Garrod's Inborn Factors in Disease. C R Scriver, B Childs. Oxford Monographs on Medical Genetics 16. (Pp 248; £25·00.) Oxford: Oxford University Press. 1989.

The heart of this book is a facsimile reprint of Sir Archibald Garrod's last book originally published in 1931. This is sandwiched between a prologue, 'Garrod in context' and an epilogue 'Genetic predisposition after Garrod' together with two bibliographies, one of Garrod's writings and the other of articles about him. (Gowland Hopkins wrote no fewer than three obituaries.)

The term 'inborn error of metabolism' coined by Archibald Garrod has now become so commonplace that it is difficult for us to remember how startlingly original was his thinking. At the time, disorders such as alkaptonuria were still thought to be caused by a gut infection that disturbed the metabolism of tyrosine. It was Garrod's careful studies of families with patients with alkaptonuria and other metabolic disorders that led him to the concept of an inborn error. These studies also raised the question of why do some people develop certain diseases. Garrod proposed that the predisposition was often genetically determined and he referred to it as chemical individuality. This book is an exposition of his hypotheses and he uses a wide range of diseases to illustrate his ideas, including infections and immune deficiencies, metabolic disorders, atopy, and dysmorphic syndromes. His analysis of the problems and the anticipation of future developments is astonishing. He recognised that all genetic information was encoded on the chromosomes, predicted that the chemical basis of the variation would be expressed in proteins, and that the effect of the genetic defect would either be the failure of a normal process or secondary to the accumulation of toxic metabolites.

In the concluding chapter Garrod casts some doubt on his hypotheses stating that "in fifty years a still more ample knowledge will doubtless displace many of our conclusions". The advances in molecular genetics have, of course, amply confirmed rather than refuted his work. This is a fascinating book, well written in graceful prose, and the extra chapters help to put the whole work into its context. I can warmly recommend it as it provides a remarkable insight into Garrod's genius.

J V LEONARD

Handbook of Normal Physical Measurements. Judith G Hall, Ursula G Froster-Iskenius, Judith E Allanson. (Pp 504; £25·00.) Oxford, New York: Oxford University Press. 1989.

The book is described as an easy to use pocket book, which provides a collection of reference data on physical measurements for use in clinical assessment of children and adults with dysmorphic syndromes. This is a rather dry description of what is a very user friendly book with reference data and much more. Each section begins with a useful definition of the measurement of the particular body part, and the best way to obtain this. Diagrams and helpful hints are included as necessary, followed by appropriate graphs, tables, and references. Many sections include a brief embryological summary.

There is a wealth of information, not just on linear measurements of body parts, but on birth marks, dermatoglyphics, bone age, prenatal measurements, placentation, and growth charts for certain genetic disorders.

My guess is that every clinical geneticist will obtain a copy for personal use, all hospital and community paediatric units ought to have one, and that it will be very widely used as word spreads of its existence.

One criticism—it doesn't fit into my pocket; nevertheless, to borrow a phrase from at least one of the authors "it's a real neat little book".

DIAN DONNAI

Molecular Probes—Technology and Medical Applications. Ed Alberto Albertini, Rodolfo Paoletti, Ralph Reisfeld. (Pp 316; £112·50.) New York: Raven Press. 1989.

This volume contains papers presented at the three day International Sym-

posium BIOTECH RIA '88 held in Florence in April 1988. The text is divided into three sections, the first, 'Molecular probes in genetic diseases', describes new methodologies aimed at improving the detection of genetic defects. Frequent reference is made to the now familiar polymerase chain reaction (PCR) method of DNA amplification, showing the powerful potential of this technique in a range of diagnostic situations from the detection of molecular defects in β thalassaemia (Boehm *et al*) to the detection of hepatitis B viral DNA sequences in the monitoring of antiviral therapy (Larzul *et al*). The paper on Duchenne and Becker muscular dystrophy (Ommen *et al*), although missing PCR deletion detection methods, is a good, brief, but comprehensive overview of developments in DMD and BMD. The genetic analysis of hyperlipidaemia (Humphries *et al*) provides a useful insight into the study of a polygenic disorder at the molecular level.

The second part, 'Molecular probes in infectious diseases', focuses mainly on HIV studies and includes new diagnostic assays and methods of sero-epidemiological characterisation of the infection; again PCR emerges as a detection method of choice. Sensitive antibody screening assays are described, based primarily on the use of viral recombinant antigens produced in quantity and to a high degree of purity by using ingenious high level expression vector systems containing temperature sensitive repressors (Papas *et al*).

The third part, 'Molecular probes on solid tumors', presents data on the diagnostic, prognostic, and therapeutic use of monoclonal antibodies (MAbs) in the treatment of human tumours. Riesfield describes the promising potential use and problems of developing immunoconjugates between MAbs (directed to tumour associated antigens) and chemotherapeutic drugs or toxins, and Kageshita *et al* present encouraging data on the development of specific immunotherapy using anti-idiotypic antibodies made possible largely by recent advances in hybridoma methodology. Optimal combinations of various therapeutic regimens would seem necessary for effective tumour therapy since experiments and clinical trials with single agents, for example, cytokines (Hermann *et al*), have been disappointing. Pierotti and Porta present a useful review of DNA probes and cancer, including examples where analysis with DNA probes has contributed to the diagnosis and/or prognosis of certain solid tumours by showing oncogene alteration, tumour specific oncogene amplification, or allele loss in certain tumours.

Overall, the papers are presented in an easily readable, clear, concise style with good illustrations, and assume only a peripheral knowledge of the areas covered. With the obvious reservation that any publication in the rapidly advancing field of molecular genetics can date with alarming rapidity, this well referenced mini review approach to the molecular probe phenomena provides a useful, broad based source of information for scientists and clinicians alike.

JOHN F HARVEY

NOTICES

The Joint Convention of the 5th International Child Neurology Congress and the 3rd Asian and Oceanian Congress of Child Neurology

This convention will take place at the Keio Plaza Inter-Continental Hotel, Tokyo, Japan on Sunday 4 to Friday 7 November 1990. For further information contact President: Dr Yukio Fukuyama, Department of Pediatrics, Tokyo Women's Medical College, 8–1 Kawadacho, Shinjuku-ku, Tokyo 162, Japan. Tel: 81-3-353-8111, or Secretariat: Dr Yoshiyuki Suzuki, Secretary General, 5th ICNC–3rd AOCCN, The Tokyo Metropolitan Institute of Medical Science, 3-18-22 Honkomagome, Bunkyo-ku, Tokyo 113, Japan. Tel: 81-3-823-2101.

Genetics of Hearing Impairment

The New York Academy of Sciences and The Deafness Research Foundation are holding a meeting on 'Genetics of Hearing Impairment' on 24 to 26 September 1990 at the Sheraton Center, New York City. For further information contact: Conference Department, The New York Academy of Sciences, 2 East 63rd Street, New York, NY 10021, USA. Tel: 212-838-0230.