Severe intrauterine growth retardation with increased mitomycin C sensitivity, or Nijmegen breakage syndrome?

We read with great interest the paper by Woods et al entitled "Severe intrauterine growth retardation with increased mitomycin C sensitivity: a further chromosome breakage syndrome." We believe that this is an important paper. However, we do not agree that this patient has "a further chromosome breakage syndrome.

The reported infant had pre-and postnatal microcephaly and growth retardation, a distinctive facies, and developmental delay. He became pancytopenic at 16 months and died soon after. Increased spontaneous random chromosome breakage was seen in blood and fibroblast cultures. Mitomycin C induced chromosome damage was increased and comparable to that seen in Fanconi anaemia.

The authors hypothesise that this entity of severe intrauterine growth retardation and increased mitomycin C sensitivity may be a distinct chromosome breakage syndrome. We suspect that the patient of Woods et al most probably has the Nijmegen breakage syndrome (NBS). The physical features, very well illustrated in the paper, as well as the chromosomal breaks, are very suggestive of this diagnosis. Unfortunately, a post-irradiation DNA synthesis test has not been performed on the child's cells, nor a serum fetoprotein determination to differentiate from ataxia telangiectasia (AT).

The hypospadias described in the patient of Woods et al has not been previously reported in NBS patients. However, we have followed up a boy of Yugoslavian origin affected with NBS who presented with hypospadias and thus suspect that the child reported by Woods et al and our patient are affected with a new clinical variant of NBS. At the end of their paper the authors inform us that fibroblast cell line MI-C445 from their patient is available from the Murdoch Institute for additional studies. We suggest that a post-irradiation DNA synthesis test be performed on these cells to rule out or confirm the diagnosis of NBS.

In case this diagnosis is confirmed, the cells of their patient and ours should have complementation studies with cell lines of other patients diagnosed as NBS, to determine whether they represent a separate and new complementation group.

VAZKEN M DER KALOUSTIAN
ALISON M ELLIOTT
PATRICE EYDoux

Divisions of Medical Genetics andCytopathology,
Montreal Children's Hospital and
McGill University,
2000 Tupper,
Montreal, Quebec H3H 1P3,
Canada.


3 Taalman RDFM, Jaspers NGJ, Scheres JMJC, de Wit J, Hruska TWJ. Hypersensitivity to ionizing radiation in vitro, in a new chro-mosomal breakage disorder, the Nijmegen breakage syndrome. Mutat Res 1983;112:33-32.


BOOK REVIEWS

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This is the first book which devotes itself entirely to the psychosocial implications of prenatal diagnosis and screening. The multidisciplinary array of chapter authors participated in conferences at the Institute of Obstetrics and Gynaecology in London in 1992, which showed the increasing awareness among health care professionals of the emotional sequelae to antenatal testing for abnormality. The chapters, on the whole, complement one another rather than going over the same ground, and are enhanced by being approached from so many different perspectives (clinicians, genetic counsellors, midwives, ultrasonographers, research psychologists, parents). The only chapter which does not sit comfortably with the rest is the one on preimplantation diagnosis, as it mainly describes the procedure rather than focusing on acceptability and psychological aspects.

Jo Green and Lenore Abramsky's chapters clearly set out the case for improved pretest counselling, based both on research data (useful comprehensive list of references) and clinical experience, although they focus mostly on routine population screening tests rather than prenatal diagnosis for genetic indications. Christine Garrett and Lyn Charlton's excellent chapter on difficult decisions will be of particular interest to genetic counsellors (clinicians and coworkeurs) as they focus on those situations where the results of prenatal diagnosis are less than clear. Their discussion of decision making models is practically focused.

Although prenatal screening is provided routinely in ultrasound departments, the pressures and dilemmas this poses for radiographers has previously been given little attention, so the chapter by an ultrasonographer is an important contribution. Although the dilemmas outlined will no doubt be familiar to radiographers, it is also informative for the rest of us to appreciate the constraints under which these professionals often work, because of historical protocol. The "human side" of prenatal diagnosis must include the emotional impact on staff as well as on parents, but the chapter devoted to this, "Caring for the care-takers," is often good and staff consider themselves in fact better dealt with in the chapter on late prenatal diagnosis (written by Lucy Turner, a midwife). It would also have been interesting to include the impact on laboratory staff.

Helen Statham, writing from her own personal point of view as a mother, as well as through her contacts with members of SAFTA, describes how each diagnosis of abnormality impacts on a family, even when the diagnosis and decision are "clear". The chapter I liked most of all was that written by Ray Hall, the father of a baby terminated after the diagnosis of spina bifida. As he states, so little is written about the father's perspective, and as genetic counsellors we often feel at a loss as to how to help fathers, that I avidly read his candid account and views on fathers' ways of coping.

Overall this volume is admirably comprehensive, with the omission of chapters on (1) support for couples who choose not to terminate, through the pregnancy and after, and (2) parents at high genetic risk, who may be facing a series of pregnancies and prenatal tests. The editors state in their introduction that the book is not intended to pass on a large body of information, but rather to draw attention to the extent of emotional sequelae stemming from prenatal screening and diagnosis. This is in fact overly modest, as I am sure that most people involved in providing prenatal diagnostic services will learn a great deal from this book and being stimulated to re-evaluate their practice.

LAUREN KERZIN-STORRAR


Together with single gene disorders and multifactorial diseases, such as cardiovascular disease and cancer, autoimmune diseases are currently the object of much investigative attention. As far as autoimmune disease is concerned I believe this book is somewhat parochial, but the first is the intense contemporary intellectual ferment which is the study of immunology, while the second is the strong desire to exploit our emerging understanding of the world in which we live and to feel how we might make the most of it.

Autoimmune disease in its purest sense is the result of the immune system turning on itself, for its own purposes. In this book the authors attempt to explain the devastating impact of autoimmune disease on the unfortunate victim by invoking the concept of "Friendly Fire". The term was coined during the Gulf War to describe accidental fire from our own side (which operationally meant the anti-Iraqi coalition). The analogy is useful in that autoimmune disease is a relatively infrequent consequence of that most potent of defense mechanisms, the host immune system. However, the authors enjoy stretching their metaphor to the point where it becomes irritating. I find that I want to see how neutrophils can be seen as the equivalent of the SAS or Delta Force as claimed on p 20. The latter are not numerous and are reputed to be highly selective killers whereas neutrophil invasion of an inflamed tissue exhibits quite different characteristics. There are also problems with the Friendly...
Secure by Peter Tumpey. (Pp 226; £12.95.) UK: British Agencies

As genetic knowledge increases and becomes more widely applied it becomes more imp-
portant for the adopted person to have inform-
ation about his or her genetic parents and family history. Where this is unavailable he or she may feel particularly vulnerable when starting their own family and request "genetic testing" in general, a request difficult to discuss in a vacuum. Reasons for this anxiety will become clearer after reading this book.

It is multidisciplinary with contributions from clinicians, social workers, and a lawyer all involved in British Adoption Societies, together with clinical and laboratory gen-
eticists and research psychologists. The only omission is that of the individual person himself or herself but a chapter of illustrative case studies and other case reports and quotes to some extent fill this gap.

The early sections outline medical aspects and stress the importance of obtaining ade-
quate details of the birth parents' family history, although inevitably this may prove difficult. A number of chapters describe rel-
levant genetic disorders with special emphasis on neurological, developmental, cardiac, and psychiatric disorders, and malignant disease. Inevitably repetition occurs as neuro-

Familial Adenomatous Polyposis and Other Polyposis Syndromes, Editors Philip PAS, Spigelman AD, Thompson JPS. (Pp 234; £65.00.) London: Edward Arnold. 1995.

This is an extensive book about FAP and associated disorders by a group of experts in this field. The book inevitably has a surgical bias, but in some senses this makes it even more applicable to a clinical genetic readers.

This volume provides a useful body of experience to geneticists contemplating a similar exercise on their favoured syndrome. Other papers discuss the characterisation of various syndromes to better understand the basis of the way affection is approached, as well as the possibilities for future genetic approaches as well as discussing mod-
els of tumour development and progression.

The other chapters are much more heavily based on basic biological research into cell regulation. In this regard, they do not dis-
appoint, providing handy reviews of the cut-
ing edge in this burgeoning area of investiga-
tion and thereby offering a useful backdrop into the processes that might be active in cancer. They may also supply a ready source of candidate genes for further investigation by medical geneticists wishing to characterise the aetiology of human cancer.

A basic understanding of the underlying genetics must be assumed in the reviews and less well prepared readers would be advised to have read the relevant chunks of the current edition of a decent undergraduate molecular biology text, say, The molecular biology of the cell.

In summary, the content of this volume is heavily biased toward the biology of cancer and although mostly not of direct relevance to medical geneticists, it is a useful reference text to consult when attempting to develop a fresh approach to genetic research in cancer and as such may be usefully acquired by medical libraries.

DAVID HUEN


This is the latest offering in a distin-
guished series of symposium proceedings emanating from the Cold Spring Harbor Laboratory Press. It presents a collection of "well written and participative" papers to be found in the symposium of the same name held at Cold Spring Harbor Laboratory a year ago.

The volume is organised around five subjects: control of cell cycle and cell growth, checkpoint and genome stability, apoptosis, gen-

etic models, human cancer genes and their products, and genetic methods for diagnosis and cancer therapy.

Given that the papers presented in the final category were focused primarily on gene therapy, it is the penultimate category that would of immediate relevance to medical geneticists. Two retrospectives review the genetic approaches deployed in the iden-
tification and cloning of the BRCA1 gene, which should prove a useful body of ex-
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David Huen


This is an extensive book about FAP and associated disorders by a group of experts in this field. The book inevitably has a surgical bias, but in some senses this makes it even more applicable to a clinical genetic readers.

It contains strong chapters on in-
trduction, history and registry, pathology, endoscopy, screening including CHIRPE iden-
tification, and the various surgical op-
tions. The St Mark's experience with FAP is such that they of all those centres are best placed to offer surgical guidelines on the management of this condition. Inevitably there will be a slight bias in their as-
certainment with more severely affected cases and families being referred. Nonetheless, their recommendations are broadly ap-
plicable, particularly to FAP itself.

There are, nonetheless, areas in which surgeons rather arbitrarily put forward for various aspects of screening in FAP and other polyposis conditions. For ex-
ample, while the experience of St Mark's with screening of the upper GL tract for duodenal polyps and cancer is undeniable, they are yet to show a benefit for this form of screening, and in some senses this should be treated in the same way as screening in other cancer settings, such as that of the ovari with ovarian