

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

We read with great interest the paper by Woods *et al*¹ 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).²⁻⁴ The physical features, very well illustrated in the paper, as well as the chromosomal breaks, are very suggestive of this diagnosis. Unfortunately, a post- γ irradiation DNA synthesis test has not been performed on the child's cells, nor a serum α 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- γ 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.

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- 1 Woods CG, Leversha M, Rogers JG. Severe intrauterine growth retardation with increased mitomycin C sensitivity: a further chromosomal breakage syndrome. *J Med Genet* 1995;32:301-5.
- 2 Weemaes CMR, Hustinx TWJ, Scheres JMJC, van Munster PJJ, Bakkeren JA, Taalman RDFM. A new chromosomal instability disorder: the Nijmegen breakage syndrome. *Acta Paediatr Scand* 1981;70:557-64.
- 3 Taalman RDFM, Jaspers NGJ, Scheres JMJC, de Wit J, Hustinx TWJ. Hypersensitivity to ionizing radiation, in vitro, in a new chromosomal breakage disorder, the Nijmegen breakage syndrome. *Mutat Res* 1983;112:23-32.

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BOOK REVIEWS

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Prenatal Diagnosis: The Human Side. Editors L Abramsky, J Chapple. (Pp 211; £15.99.) London: Chapman and Hall. 1994.

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 a conference 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 coworkers) 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 placed on radiographers often 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 carers", is too generic and staff concerns are 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 so often feel at a loss as to know 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 psychosocial 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 as well as being stimulated to re-evaluate their practice.

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Friendly Fire: Explaining Autoimmune Disease. D Isenberg, J Morrow. (Pp 155; £17.99.) Oxford: Oxford University Press. 1995. ISBN 0-19-262220-X.

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 to be because of two factors. 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 notions of self tolerance in some practical form. In the twilight years of the 20th century perhaps immunology expresses the ultimate search for self, the search for that which makes us different.

Autoimmune disease in its purest sense is the result of the immune system turning its considerable firepower on its own host tissues. 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 attrition against one's 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 fail 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