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 pancypopenic 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 are surprised 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 hypoplasias 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 hypoplasias 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 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


BOOK REVIEWS


If you wish to order or require further information regarding the titles reviewed here, please write to or telephone the BMJ Bookshop, PO Box 295, London WC1H 9JR. Tel 0171 383 6244. Fax 0171 383 6662. Books are supplied post free in the UK and for BFPO addresses. Overseas customers should add 15% for postage and packing. Payment can be made by cheque in sterling drawn on a UK bank or by credit card (Mastercard, Visa, or American Express) stating card number, expiry date, and full name. (The price and availability are occasionally subject to revision by the Publishers.)

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 it is dominated by 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 knowledge of the immune system and its 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 fire from 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 infected tissue exhibits quite different characteristics. There are also problems with the Friendly...