LETTERS TO THE EDITOR

X linked α thalassaemia/mental retardation (ATR-X) syndrome

We read with interest the series of reports in the journal on the ATR-X syndrome.1,2,7 The facial features are similar to affected subjects with another X linked MR syndrome in a Canadian family described by us a few years ago.3 Cole et al suggested that our family may have the ATR-X syndrome. We studied the three living affected males in the family again. All showed normal haematological indices, no detectable haemoglobin H on electrophoresis, and no cells containing haemoglobin H inclusions using 1% brilliant cresyl blue preparations of peripheral blood smears.

In addition, our patients appear to be less severely retarded than the British families described. The possibilities are that these two disorders may (1) be the result of different mutations, (2) be allelic, or (3) represent an X chromosome contiguous gene syndrome with different sized deletions. In respect to the last hypothesis, high resolution chromosome analysis in our laboratories (Winnipeg and Calgary) and in Houston (courtesy of David H Liedbetter) showed no deletions. Identification of the gene locus and molecular studies will ultimately resolve the issue.

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49,XXXY syndrome: behavioural and developmental profiles

I read with great interest the recent paper by Lomelino and Reiss,1 reporting behavioural, psychological, and cognitive profiles of two cases of 49,XXXY and one of 48,XXXXY/49,XXXY mosaicism. In 19642 I reported two cases of severely mentally handicapped patients with XXXXY syndrome, who suffered from similar developmental profiles. Case 1 improved very little in cognition, behaviour, and language development since 1964 and died from bronchopneumonia at the age of 23 years.

Case 2 is now 44 years of age and intellectually he has improved slightly since 1964. Assessed on the Merrill Palmer Scale, his MA is 5-11 years but he still cannot read or write. His very limited comprehension and expression has shown some improvement. Results of testing on the Reynell Development Language Scales were: raw scores 42, equivalent age 5-03 years. The main characteristics which make him difficult to understand are the very low volume of his voice (he mostly speaks only in a whisper) and his shyness.

However, there are increasing problems with his behaviour, so temper tantrums, kleptomania, and self-injury, particularly to his hands. He constantly picks a chronic hypostatic ulcer3 on his leg. He is often untruthful and mischievous. The patient has been caught setting fire on many occasions and therefore requires constant supervision.

Sometimes he becomes verbally abusive, swearing and threatening violence, and will resort to throwing things.

Physically he is prone to falls and sustained a fracture of the neck of the right femur and fracture of the left patella, which is adding to his frustrations.

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BOOK REVIEWS

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An academic paediatrician who recently reviewed advances in genetics commented that the subject had become much too important to be left to geneticists. His realisation that only came late in his obstetric practice should congratulate themselves on being far-sighted enough to have established (with perhaps much more than a little prompting from a few pioneering geneticists) a multidisciplinary subspeciality which, paradoxically for a subspeciality, has grown to touch upon the lives of nearly everyone in the general population. This, of course, means that workers in the field carry a very heavy burden of responsibility, so it is timely that the Royal College of Obstetricians and Gynaecologists should, in its 23rd study group meeting, examine the state of antenatal diagnosis.

This book contains papers presented at the meeting and edited transcripts of the resulting discussions. Its title is brief but do not be misled into anticipating that its contents will merely be another account of the latest clinical and laboratory techniques, although there are certainly many expert and critical expositions of these. One only has to run through the list of the meeting’s participants, noting a philosopher, an economist, a psychologist, a civil servant, and several epidemiologists, to realise that a wide brief is being addressed. The breadth of the contribution is reflected in a list of 19 conclusions and recommendations at the end of the book. As well as statements on clinical procedures and laboratory tests, such as ultrasound examination, fetal blood sampling, and maternal serum screening, there are more general statements supporting the organisation of regional genetic centres and fetal medicine centres, calling for improved professional and lay education in genetics and recommending that ethicists should be involved in the introduction of new techniques. Two other recommendations are, firstly, each district general hospital should have a consultant obstetrician with a special interest in and responsibility for the various aspects of prenatal diagnosis and, secondly, that appropriate support, obstetric follow up, and genetic counselling should be available to women who undergo termination of pregnancy for fetal abnormality. I thought there might be a statement on the relative merits of amniocentesis and chorionic villus sampling, given that there is discussion of this at several points in the text, but perhaps the study group did not wish to prejudice the then unpublished MRC trial comparing these procedures.

The book’s 25 chapters, which embrace almost the whole subject, are arranged in seven sections and comprise reviews in the...