Fertility in a male with trisomy 21

In 1989, we reported[1] the first fully documented case of an apparently non-mosaic male with Down's syndrome fathering a pregnancy. This pregnancy subsequently miscarried about nine weeks after a CVS procedure.

The same couple returned to the Genetics Centre early in 1991 in the first trimester of pregnancy. A chorion villus sample was again obtained (Mr D Maxwell) and this showed a normal karyotype. DNA samples from the CVS were analysed with the highly polymorphic probes *D7S21*[2] and *D17S30*,[3] the results confirming that paternal alleles for both markers in the CVS were also present in the putative father. This second pregnancy proceeded to term, and a normal boy has now been born to this couple.

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1 Sheridan R, Llerena J Jr, Matkins S, Debenham P, Cawood A, Bobrow M. Fertility in a male with trisomy 21. *J Med Genet* 1989;**26**:294–8.
2 Wong Z, Wilson V, Patel I, Povey S, Jeffreys AJ. Characterization of a panel of highly variable minisatellites cloned from human DNA. *Ann Hum Genet* 1987;**51**:269–88.
3 Horn GT, Richards B, Klinger KW. Amplification of a highly polymorphic VNTR segment by the polymerase chain reaction. *Nucleic Acids Res* 1989;**17**:2140.

Clinical consequences of deletion 1p35

We would like to make a correction to our paper[1] previously published in the journal. Upon analysis of another blood specimen at higher resolution, we found a balanced translocation. The corrected karyotype is 46,XY,t(1;11)(p34.3;q25).

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1 Wenger SL, Steele MW, Becker DJ. Clinical consequences of deletion 1p35. *J Med Genet* 1988;**25**:263.

Maxillonasal dysplasia (Binder's syndrome) and chondrodysplasia punctata

In a recent letter to the editor, Sheffield *et al*[1] discussed the true relationship between maxillonasal dysplasia (Binder's 'syndrome') and chondrodysplasia punctata. The authors noted that the number of patients diagnosed as chondrodysplasia punctata had been underestimated because punctate epiphyses disappear with age, and they suggested looking at old radiographs that had been performed in infancy.

We have another suggestion. If the patient is a male, it would be advisable to examine the hands of his maternal relatives, especially uncles and nephews, to look for brachytelephalangy. Moreover, if the disease seems to be inherited in an X linked recessive manner, an analysis of Xp could be indicated as the gene CDPX 1 has been mapped to Xp22–32. In one familial case, Petit *et al*[2] found an interstitial deletion in Xp22.3.

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1 Sheffield LJ, Halliday JL, Jensen F. Maxillonasal dysplasia (Binder' syndrome) and chondrodysplasia punctata. *J Med Genet* 1991; **28**:503–4.
2 Petit C, Melki J, Levilliers J, Serville F, Weissenbach J, Maroteaux P. An interstitial deletion in Xp22.3 in a family with X-linked recessive chondrodysplasia punctata and short stature. *Hum Genet* 1990;**85**:247–50.

BOOK REVIEWS

All titles reviewed here are available from the BMJ Bookshop, PO Box 295, London WC1H 9TE. Prices include postage in the UK and for members of the British Forces Overseas, but overseas customers should add 15% to the value of the order 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.

Congenital Chromosome Aberrations and Tumour Predisposition. Gyorgy Fekete. (Pp 162; £13.00.) Budapest: Akademiai Kiado. 1990.

In this book, the Hungarian author reviews some of the clinical and scientific publications, published up to 1989, concerning the association between genetic factors and malignant disease, with particular emphasis on congenital chromosome aberrations and tumour development.

In the first chapter the nature and frequency of congenital chromosome aberrations associated with cancer predisposition and associated clinical symptoms are described. The relationship between induced chromosome aberrations, chromosome instability, oncogenes, fragile sites, and malignant disease is discussed in chapter two. Chapter three is very short and deals with the topic of genetic counselling for cancer patients and their families. An attempt is made to give advice concerning screening for malignancy and the author only very briefly covers the subject of risk assessment. The final chapter describes a number of culture and banding techniques for the preparation of chromosomes from various tissues. The classification of structural chromosome aberrations and the nomenclature (ISCN 1985) used to describe them is included.

Much work has been done over the past few years in this fast developing field and a review of this subject is timely. Unfortunately, much of the data presented here are out of date and in places the experimental data derive mainly from the author's own laboratory. There is a wealth of world publications available concerning congenital chromosome aberrations and cancer predisposition and a number of important recent developments have not been included in this volume. For instance, the association between Beckwith-Wiedmann syndrome and Wilms' tumour and the suggestion of two possible loci for Wilms' tumour on chromosome arm 11p (Mannens *et al. Hum Genet* 1988;**8**:41–8) are not mentioned. Also, many of the incidence figures quoted for particular disorders relate only to Hungary and therefore may not have much relevance for a wider readership. It must be mentioned, however, that there may be problems in obtaining some current journals in eastern Europe which may have contributed to this lack of up to date references.

A number of serious scientific inaccuracies are also present in this book. The author states incorrectly that carriers of constitutional 13q14 deletions usually develop unilateral retinoblastoma rather than bilateral tumours. When describing the t(9;22) observed in chronic myeloid leukaemia (CML) the author states that the breakpoint on 9q34 is located close to the c-abl oncogene. However, it is known that the breakpoints on 9q34 are scattered and can be located both 5' of the first c-abl exon as well as located within the c-abl oncogene (Heim and Mitelman. *Cancer cytogenetics*. New York: Liss, 1987). It is also stated that in CML the c-abl oncogene is associated with the immunoglobulin heavy chain locus. This statement is incorrect as the t(9;22) translocation involved in CML places c-abl next to the breakpoint cluster region on chromosome 22 (Shtivelman *et al. Nature* 1985; **315**:550–4) and not in association with the immunoglobulin heavy chain locus.

The translation into English is poor and consequently the book is difficult to read in places. Also, the quality of many of the figures, including those reproduced from other publications together with ones from the author's own laboratory, is very poor. For example, in a figure showing deletions of chromosome 13 involved in retinoblastoma it is very difficult to see the GTG banding pattern on the chromosomes. Similarly, the poor reproduction of figures on pages 52 and 53, depicting induced chromosome aberrations, makes their interpretation difficult. Finally, the representation of Q, G, and R bands reproduced from the Paris Conference (1971) has been altered by the addition of extra and incorrectly positioned light G bands on the long arms of chromosomes 13,14,15, and 16.

The book is primarily aimed at paediatricians, oncologists, and medical geneticists. However, as it is sadly out of date, it will only be useful as a general introduction to the topic. If greater detail is required by the reader then it will be necessary to read more up to date publications.

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Aspects of Oral Molecular Biology. Frontiers of Oral Physiology, volume 8. Ed DB Ferguson. (Pp 144; £67.40.) Basel: Karger. 1991.

This book presents an overview of molecular biology in relation to fundamental oral and