Fertility in a male with trisomy 21

In 1989, we reported 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 M. Maxwell) and this showed a normal karyotype. DNA samples from the CVS were analysed with the highly polymorphic probes D7S21 and D17S36, 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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BOOK REVIEWS

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Clinical consequences of deletion 1p35

We would like to make a correction to our paper 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(11;12)(p34.3;q32).

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Maxillonasal dysplasia (Binder’s syndrome) and chondrodysplasia punctata

In a recent letter to the editor, Sheffield et al 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 brachytelephalgy. 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 found an intermittent deletion in Xp22.3.

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This book presents an overview of molecular biology in relation to fundamental oral and laboratory. There is a wealth of publications available concerning congenital chromosomal aberrations and cancer predisposition and a number of important 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 unila teral retinoblastoma rather than bilateral tumours. While describing the t(9;22) translocation 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 Mittelman. 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 (Shivelman 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 chromosome. Furthermore, the poor reproduction of figures on pages 52 and 53, depicting induced chromosomal aberrations, makes their interpretation difficult. Finally, the representation of Q, G, and R banding patterns reproduced from the Paper microscope (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.