Deleted X chromosomes in patients with the fragile X syndrome

It is now well recognised that the fragile X chromosome anomaly is associated with certain cases of X linked mental retardation.¹⁻³ The fragile site is usually seen as an isochromatid gap at the interface of the long arm bands Xq27 and Xq28.

However, we have observed that the fragile X is also frequently manifested as a deleted X chromosome with loss of the distal long arm band, q28 (figure). These deleted Xs may have arisen in culture as a result of mitotic non-disjunction of the segment q28.1→qter at a previous division. This event would also result in the production of X chromosomes with triradial configurations (sometimes described as 'double satellites') which are not uncommonly found in cells from fragile X patients. Alternatively the deleted Xs may result from the dissociation and loss of the segment q28.1→qter during spreading when slides are made. (Acentric fragments, morphologically similar to the deleted segment, have been observed at some distance from the deleted X chromosome in metaphase spreads.)

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FIGURE G banded X chromosomes showing (a) the fragile site at q28 and (b) deleted Xs from the same patient.

We have observed this deletion in several patients with the fragile X syndrome. It was particularly evident in one case in which the deleted X was identified in 11 out of the 50 cells analysed. Four cells showed triradial configurations and a further 21 showed the fragile locus at q28, giving an overall incidence of 72%.

Chromosome preparations for fragile X detection are routinely G banded without previous scanning of conventionally stained slides. This allows the unequivocal identification of the X chromosomes⁴ and precludes the need for a two stage analysis. The deleted X chromosomes could not be detected without the application of banding techniques as the lost segment is too small to make a noticeable difference to the overall length of the X chromosome in unbanded preparations.

References

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MARGARET FITCHETT AND
MARINA SEABRIGHT

Wessex Regional Cytogenetics Unit,
Salisbury General Hospital, Salisbury,
Wiltshire SP2 7SX.

Correspondence and requests for reprints to
Dr Marina Seabright, Wessex Regional Cytogenetics
Unit, The General Hospital, Salisbury, Wiltshire
SP2 7SX.