Technical note

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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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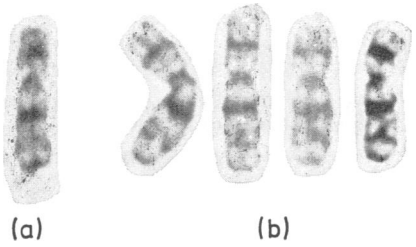

FIGURE *G banded X chromosomes showing (a) the fragile site at q28 and (b) deleted Xs from the same patient.*

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 chromosomes[4] 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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