Correspondence

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Expression of fragile X chromosome and possible deletion in successive cell divisions

SIR,

Fitchett and Seabright¹ have shown that deletion of the Xq distal segment occurs in a number of cells from patients with fra(Xq). To explain this finding we put forward the hypothesis that the fragile site is manifested in vitro only in the first cell division, whereas deletions may occur during the second and third divisions, thus masking the expression of the fragility.

To test this possibility we studied the patterns of SCE using a low concentration of BrdU (2 µg/ml instead of the standard 10 µg/ml, which inhibits the expression of the fragile site). Our hypothesis was not confirmed, however, because the fra(X) was manifested in the first, second, and third cell divisions (figure).

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Reference

- ¹ Fitchett M, Seabright M. Deleted X chromosomes in patients with the fragile X syndrome. *J Med Genet* 1984;21:373.

Noonan syndrome

SIR,

I would like to add a word to Dr Allanson's excellent and concise review of Noonan syndrome.¹ The adjective 'dysplastic' should precede 'pulmonic valve stenosis'. Most stenotic pulmonic valves have a domed appearance from commissural fusion and are readily detected on auscultation by the presence of a systolic ejection click. In contrast, the dysplastic valve in Noonan syndrome has thickened, immobile, non-doming leaflets without clearly defined

FIGURE *Fra(Xq)* shown in first (a), second (b), and third (c) divisions in peripheral blood culture.