

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

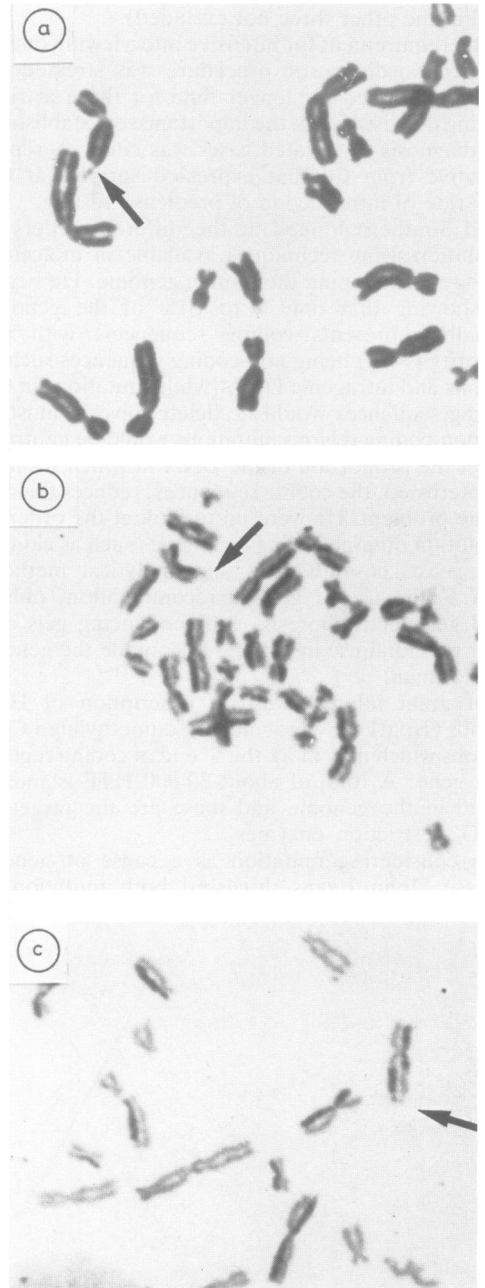


FIGURE *Fra(Xq)* shown in first (a), second (b), and third (c) divisions in peripheral blood culture.

commissures and unaccompanied by a systolic click. Of patients with a dysplastic pulmonic valve, many will have Noonan syndrome. Conversely, many patients with Noonan syndrome and pulmonic stenosis will have a dysplastic pulmonic valve. This 'atypical' and less common type of valvar pulmonic stenosis is distinctive for Noonan syndrome and unlike typical pulmonic stenosis is not amenable to balloon dilatation valvuloplasty.

The precise diagnosis of a highly specific congenital heart defect in a malformation syndrome is of equal importance as the identification of craniofacial dysmorphism.

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Reference

¹ Allanson JE. Noonan syndrome. *J Med Genet* 1987;24:9-13.

The population genetics of Duchenne muscular dystrophy

SIR,

I am indebted to Dr A O M Wilkie for pointing out an error in equation 14 (p 523) in a recent paper,¹ due to my overlooking the contribution from new mutations of grandmaternal origin. An additional 'u' in this equation leads to the paternal:maternal ratio of mutational rate being equal to the $4g/(1-2g)$, and a sixth, not a fifth, of mutations being from the maternal grandfather if these rates are equal.

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Reference

¹ Edwards JH. The population genetics of Duchenne: natural and artificial selection in Duchenne muscular dystrophy. *J Med Genet* 1986;23:521-30.