LETTER AND REPLY

FAP and Marfanoid habitus

We read with interest the report of two unrelated patients with FAP and Marfanoid habitus. Notably, a point mutation was detected in Case 2, in the donor splice of exon 4 of the *APC* gene. This ruled out a gross deletion or rearrangement involving the nearby fibrillin 2 gene locus on 5q23–31 in this case. However, direct FISH of the FBN_2 locus was not performed. We are not told whether the mother who died of abdominal malignancy had a Marfanoid habitus although the sister was tall and thin.

We would draw attention to our published case (*J Med Genet* 1992; **30**: 369-375) of FAP in an individual who had a similar Marfanoid habitus but without cardiovascular abnormalities. We were able to demonstrate a deletion in chromosome 5q in this individual (46XY, del (5), (q15q22.3 or 23.1)) *de novo*. In this case we were able to confirm the presence of two copies of the fibrillin 2 gene. The Marfanoid habitus cannot thus be caused by deletion of the fibrillin gene in this case.

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Reply

Reply to letter from ML Wilkinson and SH Hodgson

We have read with great interest the letter from Drs Mark L Wilkinson and Shirley H Hodgson, relative to their previous report on a patient with Marfanoid habitus and FAP characterised by an interstitial deletion of chromosome 5q not encompassing the fibrillin 2 gene. Their findings support our conclusion that the Marfanoid phenotype could be a direct consequence of the mutation/deletion at APC and not an in-*cis* defect of both the *APC* and the *FBN2* genes. We sincerely apologise for having missed Dr Wilkinson's and Dr Hodgson's important contribution.

We would like to take this opportunity to add to our previous clinical report that proband's mother was also characterised by a Marfanoid habitus.

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