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Correspondence

Sir,

Regarding the articles by A. J. McMahon[1] and M. G. Wyatt *et al.*[2] in Vol. 17, No. 4, the following case report may be of interest.

A 1 cm pT1 well-differentiated transitional cell carcinoma (TCC) was endoscopically excised from the bladder of a 61-year-old male following an episode of painless haematuria. Three months later, this had degenerated to a pT2 poorly-differentiated TCC with areas of non-keratinising squamous carcinoma. Since there was no evidence of synchronous or metastatic disease, he underwent external beam radiotherapy. A barium enema 5 months later, for iron deficient anaemia, showed a hepatic flexure lesion. At laparotomy he underwent an extended right hemicolectomy and an ileal resection for tumours of the hepatic flexure and ileum. There was no palpable disease in the liver or para-aortic glands. Histologically these tumours were squamous cell carcinomata consistent with that of the bladder.

We believe that colonic metastases of urothelial squamous cell carcinoma have not previously been described, and should be considered where primary squamous cell carcinoma of the colon is suspected.

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Sir,

Von Recklinghausen's neurofibromatosis is a relatively common (approximate frequency, 1/3000) autosomal dominant trait.[1] In addition to the classical signs (neurofibromas, cafe-au-lait spots, Lish nodules), increased incidence of some malignancies is of paramount clinical importance in this disease. However, to our knowledge, mammary tumors have not been reported to be increased thus far.[2] Similarly, some disturbances of endocrine function may manifest themselves in this disease, but Addison's disease has not been associated previously with neurofibromatosis.[3]

We report here the association of Von Recklinghausen's disease with a primary ductal mammary adenocarcinoma and with primary adrenal cortical insufficiency in a female who, in addition, was a likely carrier for haemophilia B. The patient, a women of 34 years of age, was diagnosed more recently to have Addison's disease of unknown etiology (non-tuberculous), which was treated with replacement therapy. She is the mother of a male child diagnosed and treated for haemophilia B, who presents no signs of neurofibromatosis. There is no evidence for haemophilia B or neurofibromatosis in other relatives in her family. She was referred to our breast tumor unit because of a suspicious lump in her right breast of 2-months duration. A biopsy and histological analysis of the mastectomy specimen confirmed an infiltrating ductal mammary carcinoma, with lymphatic spread to the axillary area (six of 20 nodes were positive) and with no evidence of metastatic spread to bone or other organs. The tumor was positive for estrogen receptors and was staged as pT2N1(6/20)M0, according to the TNM classification. It was treated with radical mastectomy followed by radiotherapy (5000 rads) and oral tamoxifen. Chariotyping and G-banding of the chromosomes obtained from peripheral blood and from tissue culture of an excised neurofibroma yielded a 46 XX patter with no abnormalities.

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To our knowledge this is the first time that neurofibromatosis, breast cancer, Addison's disease and a probable carrier state for haemophilia B have been found associated. It is conceivable that, in our patient, cortical insufficiency is the result of neurofibromatous destruction of suprarenal tissue. Whether the mammary tumor reflects the increased susceptibility to cancer of these patients,[2] or whether it is just the result of casual association cannot be ascertained. From our case, a study to test for the possible association of breast cancer and neurofibromatosis might be justified, particularly since clinical diagnosis of a breast cancer in these patients is hampered by the presence of multiple neurofibroma. As in half of the patients with neurofibromatosis,[1] this disease arose in our patient by new mutation. Haemophilia B also seems to have arisen from a new mutation either in the proband or in her son, suggesting a high mutational frequency in this family.

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