PostScript

LETTER TO THE EDITOR

Adult transmural intestinal ganglioneuromatosis is not always associated with multiple endocrine neoplasia or neurofibromatosis: a case report

A 45-year-old woman presented with a 6-week history of anorexia, diarrhoea and weight loss. Barium enema examination showed a stricturing lesion in the caecum consistent with the radiological features of a colonic carcinoma. A computed tomography scan of the abdomen confirmed the presence of circumferential mural thickening of the caecum, with extensive surrounding pericolic infiltration. There was no evidence of metastasis.

At laparotomy, there was a palpable caecal mass extending to the ileocaecal valve which was adherent to the right pelvic side wall and to the proximal transverse colon. An extended right hemicolectomy was performed.

The surgical specimen contained an annular mass in the caecum (fig 1), extending to the terminal ileum. There was no luminal surface ulceration of the lesion. Histological examination showed a diffuse infiltration of the muscularis propria by fine nerve fibres and larger nerve bundles occasionally accompanied by ganglia, with frequent extension into the submucosa and subserosal adipose tissue (fig 2A). Immunohistochemical staining showed strong immunoreactivity for S100 protein (fig 2B). Immunostaining of the tumour with anti-CD117 for interstitial cells of Cajal was negative. Despite widespread infiltration, the tumour cells lacked other features of malignancy, showing no mitoses and only mild nuclear pleomorphism. In the caecum and ileum, patchy infiltration of lymphocytes and histiocytes was seen, with the formation of lymphoid aggregates and occasional granulomas. The caecal mucosa also contained several crypt abscesses. Twelve lymph nodes retrieved from mesenteric fat showed only reactive changes. The histological features were typical of intestinal ganglioneuromatosis.

Intestinal ganglioneuromas occur in three forms: as an isolated polyp, as multiple polyps (ganglioneuromatous polyposis) and as diffuse involvement of the bowel wall (intestinal ganglioneuromatosis).¹ Intestinal ganglioneuromas occurring as isolated lesions are not typically associated with neurofibromatosis type 1 (NF1) or other genetic syndromes. However, ganglioneuromatous polyposis and diffuse ganglioneuromatosis have been described in association with NF1, multiple endocrine neoplasia type 2b (MEN2b), juvenile polyposis and non-familial adenomatous polyposis. Further clinical examination and biochemical screening in this case showed no features of NF1 or MEN2b.

Diffuse ganglioneuromatosis has two variants, transmural and mucosal. Both variants are described in children, but in adults the disease is most often limited to the mucosa.² This contrasts with the case reported here which is transmural. In children there is an

Figure 1 Right hemicolectomy specimen. There is diffuse thickening of the wall of the caecum.

association with RET proto-oncogene abnormalities: either loss of RET function associated with Hirschsprung's disease or RET activation associated with MEN2.³ In adults, lack of association with neural crest diseases in most cases would suggest an alternative molecular pathway. A number of cases reported in the literature also presented with colonic adenomas or adenocarcinomas; the underlying molecular mechanisms remain unexplained, although it has been suggested that some sporadic colorectal cancers may be linked to a point mutation of the NF1 gene.⁴

This unusual case reports an adult patient with transmural diffuse intestinal ganglioneuromatosis not associated with MEN2b or NF1. Preoperative investigations suggested a colonic carcinoma, yet the final diagnosis depended on histological examination of the resected specimen, which showed characteristic features.

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Figure 2 (A) Histological section stained with haematoxylin and eosin showing diffuse infiltration of muscularis propria by nerve fibres and large nerve bundles ($\times 10$ objective). (B) Immunohistochemical section of caecal ganglioneuromatosis stained for S100 protein ($\times 10$ objective).

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BOOK REVIEW**Tumour diagnosis: practical approach and pattern analysis, 2nd edn**

Awatif Al-nafussi. London: Hodder Arnold, 2005, \$340.75 (hard cover), pp 1338. ISBN 0-340-8094-42

This book contains the most recent information on tumours, is comprehensive in a most practical

fashion and is in accordance with the World Health Organization classification of tumours. It has 1338 pages of text and more than 1600 high-quality colour images providing clear and straightforward examples for comparison.

The book is composed of three different parts. The first part is a detailed introduction to pattern analysis, mainly dedicated to trainee pathologists. Here, ten histological patterns and related subpatterns of growth and distinctive cell types are described with simple terminology, and illustrations.

The second part considers tumours by system or organ. In each chapter, tumours are classified by cell of origin. The book describes each tumour in a concise but complete manner, and the essential approach in brief paragraphs: clinical and pathological features, often with a separate short-cell morphological description and differential diagnosis. Special techniques, such as immunohistochemical staining, are considered with diagnostic applicability. A summary is presented at the end of each chapter with the World Health Organization classification. The references are adequately organised for each category of tumour.

To recapitulate, the third part consists of a System Index, "according to various microscopic patterns and cell types of tumours", divided by systems or organs, where tumours are classified according to the cell type that they are composed of. Readers may rapidly refer to this section to get an overall impression.

I found this book to be valuable for both the inexperienced reader, seeking an understanding of tumours (it provides basic knowledge useful to achieve diagnostic skills), and also for the practising and experienced pathologist, using the book with an in-depth diagnostic approach while benefiting from subtle differences among tumour patterns and cell types. Lovers of morphology will be happy to see their dream realised.

Tumour diagnosis: practical approach and pattern analysis deserves a space on the shelf next to the microscope as it provides an excellent rapid consultation for either the experienced pathologist or the novice, and is a great source of study for the trainee.

S Serra**CALENDAR OF EVENTS****Diagnostic histopathology of breast disease**

14–17 May 2007, Hammersmith Hospital (Imperial College), London.

A 4-day course on the Hammersmith Campus designed for pathologists at Consultant and Senior Trainee level and suitable for the final examination for MRCPath. The course will provide a comprehensive coverage of the histopathology of breast disease, with special emphasis on areas which pose diagnostic difficulties. The course is based historically on the extensive referral collection of Professor JG Azzopardi, to which has been added new cases from Charing Cross, Nottingham City and Addenbrooke's Hospitals. The course will be slanted towards practical diagnostic pathology with a specific session dealing with problems encountered in core biopsies and a special workshop dealing with the interpretation of HER2 immunostaining. The participants will be given ample time to study histological preparations, followed by illustrated discussions of the cases. In addition, there will be several daily talks on specific topics, given by eminent breast pathologists and followed by discussions. The topics will include: new issues in the interpretation of breast biopsies, breast immunohistochemistry, problematic breast lesions, dealing with the gross specimen, fibro-epithelial, spindle cell and papillary lesions, proliferative lesions/carcinoma in-situ, sentinel lymph node biopsy and the impact of molecular genetics on diagnostic breast pathology. The faculty will include Anthony Douglas-Jones, Ian Ellis, Bharat Jassani, Andrew Lee, Sarah Pinder, Jorge Reis-Filho and Sami Shousha.

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