Letters to the Editor

Gastrointestinal bleeding secondary to intestinal neurofibromatosis

Key words: Neurofibromatosis type 1. Intestinal neurofibromas. Gastrointestinal bleeding.

Dear Editor:

We report the case of a 54-year-old male patient diagnosed with neurofibromatosis type 1 (NF1). The patient consulted with a 12-hour history of asthenia and rectorrhagia. He had pale skin and mucosa, was haemodynamically unstable and showed progressive anaemia. An emergency upper gastrointestinal endoscopy was performed, showing several acute lesions in the gastric antrum mucosa with no signs of bleeding. No lesions were found in the colonoscopy but fresh bleeding was detected at the ileocaecal valve, coming from upper intestinal segments. We decided to perform a selective arteriography of the caeliac trunk, superior mesenteric artery and associated branches, finding bleeding along straight jejunal branches in the left iliac fossa. Embolisation was not possible due to significant colonic distension as a result of the colonoscopy. The patient was referred to the surgical department. An exploratory laparotomy showed multiple neurofibromas affecting the proximal and mid-jejunum (Fig. 1). A jejunal enterotomy was performed and an endoscope was inserted to examine the proximal section of the small intestine as far as the duodeno-jejunal flexure and distally as far as the ileocaecal valve. No haemorrhagic lesions were observed. There was some wall thickening about 140 cm beyond the ligament of Treitz, and 15 cm of the jejunum was resected at this point (Fig. 2). Anatomical-pathological findings: submucosal haemorrhage affecting 90% of the circumference.

The patient progressed well post-operatively, with no signs of rebleeding.

Fig. 1. Neurofibromas of various sizes located in the duodenal wall.

Discussion

NF1 or von Recklinghausen's disease is an autosomal dominant genetic disorder involving mutations of the NF1 gene located on chromosome 17q11.2 (1). Diagnosis requires two or more of the following criteria: at least six "café-au-lait" maculae over 5 mm in prepubertal individuals or over 15 mm after puberty, at least two neurofibromas of any type or one plexiform neurofibroma, freckling in the axillary or inguinal region (Crowe sign), optic nerve glioma, at least two iris hamartomas (Lisch nodules), characteristic bone lesions (sphenoid dysplasia, thinning of long bone cortex with or without pseudoarthrosis), history of a first-degree relative with NF1 (2).

There is a 12-60% incidence of gastrointestinal complications in patients with NF1 and this figure increases between 40 and 60 years of age. The three main manifestations of gastrointestinal complications are: intestinal neurofibromas, gastrointestinal stromal tumours (GISTs) and periampullary neuroendocrine tumours, sometimes associated with pheochromocytomas (3). The jejunum

Fig. 2. Jejunal submucosal haematoma beside the enteroscope port of entry.

is the commonest location for intestinal neurofibromas, followed by the stomach, ileum, duodenum and colon (4). They originate in Meissner's plexus in the submucosa, in Auerbach's plexus in the muscularis propria and even in the serous membrane (5). Most intestinal neurofibromas are asymptomatic, although they may cause abdominal pain, intestinal obstruction, perforation, diarrhoea, upper or lower gastrointestinal bleeding, a palpable mass or microcytic anaemia (6).

They undergo malignant transformation in up to 5-15% of patients, especially in individuals over the age of 40. Surgery is therefore the treatment of choice in symptomatic intestinal

tumours, although asymptomatic patients may be monitored conservatively (7-9).

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