Letters to the Editor

Lynch syndrome-associated kidney cancer

Key words: Lynch syndrome. Colorectal carcinoma. Surgical treatment.

Dear Editor,

Some 80% of colorectal cancers are sporadic, 10% are familial and the remaining 5% to 10% are hereditary (1). Hereditary colorectal cancer includes familial adenomatous polyposis and hereditary nonpolyposis colorectal cancer, also known as Lynch syndrome (2). The main characteristics of this disease entity are their early presentation (45 years), preference for right colon (70%), high incidence of synchronous colorectal tumours (10%) and metachronous colorectal tumours (30-50%), and an association with extracolorectal tumours. There are 2 subtypes: Lynch syndrome type I, in which the risk of cancer is only associated with colorectal cancer, and type II, where the patients develop colorectal cancer associated with other extracolonic cancers (3).

Case report

We present a 34-year-old patient who was referred to our unit following colonscopy screening for Lynch syndrome. The patient presented protruding lesions in the cecum with biopsy findings of moderately differentiated ulcerated infiltrating adenocarcinoma. Computed axial tomography (CAT) demonstrated a multiseptated cystic lesion (6.7 x 6 x 6.4 cm; Bosniak IV) located on the left kidney (Fig. 1). Following preoperative assessment, right hemicolectomy and left nephrectomy were planned and performed. The patient made a good recovery and was discharged 5 days postintervention. Anatomicopathological study: mucinous carcinoma of the right colon with signet ring cells (pT2, pN0). Clear cell carcinoma of the left kidney (pT1). After case review, the oncology committee decided to forego adjuvant treatment and to adopt a strict follow-up protocol.

Discussion

Lynch syndrome is characterized by autosomal dominant inheritance. It is due to a germline mutation on one of the DNA mismatch repair (MMR) genes, MLH1, MSH2, MSH6 and PMS2, that are located on chromosomes 2 and 3. They are involved in 90% of the mutations and confer a high risk of cancer. This is due to the fact that they are implicated in microsatellite instability by loss of their function as repair genes of the nitrogen bases of DNA (4). The syndrome is normally diagnosed in patients who are around 45 years of age; however, 90% of sporadic
cases are found in patients who are more than 50 years old. Around 70% present tumours in the right colon proximal to the splenic flexure in contrast to that observed in sporadic colorectal cancer which is generally located in the rectosigmoid colon. The most frequently associated extracolonic cancers are as follows: endometrial (63%), stomach (28%) and, less often, urothelial, pancreatic, biliary tract, small bowel, skin and brain (5).

Lynch syndrome tends to be under-diagnosed due to the lack of a clear phenotypic marker, although some patients present a marked family history that enables a clinical diagnosis to be made based on the Amsterdam criteria. These are as follows: a) three or more family members with colon cancer in the absence of familial adenomatous polyposis, 2 of them being at least a first-degree relative; b) more than 1 generation affected; and c) one of the members should be less than 50 years of age. These criteria are restrictive and, thus, very specific; however, they have low sensitivity. Therefore, the Bethesda criteria were subsequently established which screen for patients at risk who should undergo microsatellite instability tests or other genetic tests to confirm the presence of mutations (6).

Surgical options for managing colorectal cancer include segmental resection of the colon, subtotal colectomy and total colectomy. An expert panel from Creighton University’s Hereditary Cancer Center has recommended subtotal colectomy due to the risk of synchronic and metachronic tumours. A recently published article, which compared extended colectomy to limited resection among these patients, found no difference in benefit between the 2 techniques, although there was an increase in the incidence of metachronic colorectal cancer and repeat abdominal surgical procedures among patients who had undergone limited resection. For this reason colonoscopy is recommended every 1 to 2 years. Proctoscopy is recommended in those patients who have undergone extended resection (7).

There is little experience regarding prophylactic colectomy in these patients, and prospective studies are needed to determine its true benefit. Controversy exists concerning the risk/benefit of using nonsteroidal antiinflammatory drugs and aspirin as chemoprevention in patients with Lynch syndrome.

Patients with Lynch syndrome have a 12% risk of cancer of the urinary tract. Little information is available on the follow-up of these patients. Possible diagnostic techniques include urine cytology, immunocytochemistry and the γ-glucuronidase activity test, although all of these have low sensitivity. Regarding imaging techniques, the most sensitive is urography with CAT, although this is not recommended in all patients due to the high radiation rate. Some authors recommend a urine dipstick test for the detection of hematuria (8).

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References