cancers (pancreaticoduodenectomy).

Although risk factors for cholangiocarcinoma include primary sclerosing cholangitis, congenital biliary malformations, and parasitic liver disease, and hepatolithiasis, none of those was present in the case herein reported. This is the first report the occurrence of a SRC bile duct cancer in a Western patient, and this might be relevant since cholangiocarcinomas in the East possess epidemiology and etiology that are diverse to those bile duct cancers occurring in Western countries. A higher incidence of cholangiocarcinoma in the East has been largely credited to colonization by liver *Opisthorchis viverrini* and *Clonorchis sinensis*, liver flukes that may induce inflammatory changes in the biliary tree and are endemic in Asian countries.

Although one could hypothesize that SCR hilar cholangiocarcinomas would arise from gastric ectopic or metaplastic mucosa[4], no evidence of gastric mucosa was found in the surgical specimen of the case reported herein. It has also been demonstrated that bile duct epithelium may undergo a sequence hyperplasia-metaplasia-dysplasia-carcinoma similarly to the one of intestinal-type gastric adenocarcinomas. Nevertheless, no evidence for such sequence was found in this case. In fact, biliary SRC carcinomas are poorly-differentiated aggressive tumors that may arise de novo as it is thought to occur in SRC gastric carcinomas. As happened in this case, most cholangiocarcinomas share expression of markers of progenitor cells such as CK7 and CK19. Thus, it is suggested that cholangiocarcinomas are monoclonal tumors that may arise from hepatobiliary pluripotent stem cells[9].

Another uncommon finding in the case presented here is the association between cholangiocarcinoma and clear cell renal cancer[6,8]. Although bile duct obstruction could have been attributed to the occurrence of a biliary metastasis from renal cell carcinoma[10], immunohistochemical analysis confirmed the biliary epithelial origin in this case (positivity for CK19, CK07, CAM 5.2 and AE1+AE3)[5]. Renal cell clear carcinomas do not usually stain for all these markers. Although Levy *et al.*[6] described the occurrence of simultaneous intrahepatic cholangiocarcinoma and a left kidney cancer in a patient on long-term use of methotrexate, there was no previous chronic use of any medications in this case[8]. Thus, the association of a renal cell carcinoma with a SRC Klatskin tumor is likely to be fortuitous in this case.

Cholangiocarcinoma is an aggressive malignant tumor and it is possible that the presence of signet-ring cells could confer additional aggressiveness to this tumor. Further studies will be necessary to confirm or refute this hypothesis.

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ABCDDV/1110
ABCD Arq Bras Cir Dig 2015;28(2):149
DOI:http://dx.doi.org/10.1590/ S0102-67202015000200017

Letter to the Editor

HEREDITARY DIFFUSE GASTRIC CANCER: LAPAROSCOPIC SURGICAL APPROACH ASSOCIATED TO RARE MUTATTION OF CDH1 GENE

síndrome do câncer gástrico hereditário difuso: abordagem cirúrgica radical laparoscópica associada a mutação rara do gene CDH1

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Conflicts of interest: none

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Received for publication: 08/05/2014
Accepted for publication: 20/01/2015

INTRODUCTION

Gastric cancer sets among the five most prevalent cancer in Brazil. For the year 2014, according to the National Cancer Institute (INCA), were expected 20,390 new cases with 13,328 deaths from the disease[3].

Despite the identification of risk factors for the occurrence of neoplasia[1], approximately 80% of cases are sporadic and not associated with risk factors. Even less frequent, it is the hereditary cases and associated with mutation of the HRC 1 gene, determining the occurrence of Cancer Hereditary Diffuse Gastric Syndrome. It corresponds to 3% of cases of gastric cancer[7,10].

Patients mutation CDH1 gene carriers have shown gastric cancer with more adverse prognosis, as the presence of signet ring cells, poorly differentiated tumors and diffuse histological pattern, making also important to identify families suffering from chromosomal gene changes and syndrome determinants to allow early diagnosis and preventive treatment[7,10].

As for the surgical treatment of hereditary or not gastric cancer, laparoscopy has demonstrated oncologic results similar to laparotomy approach, and lower

morbidity, with patients returning earlier to their activities[4].

This study aims to report a case diagnosed clinically and genetically, with treatment done through laparoscopic radical gastrectomy.

CASE REPORT

Man with 33y was admitted in July 2012 at the General Surgery Service of the Hospital of the Military Police of Paraná, asymptomatic and without comorbidities, but with a family history of gastric cancer in reporting two first-degree relatives with a diagnosis of cancer, one of them younger than 50 years (Figure 1A). After the endoscopy (Figure 1B), was identified ulcerated lesion in the gastric body with positive pathology for adenocarcinoma with histological pattern of signet ring cells (Figure 1C).

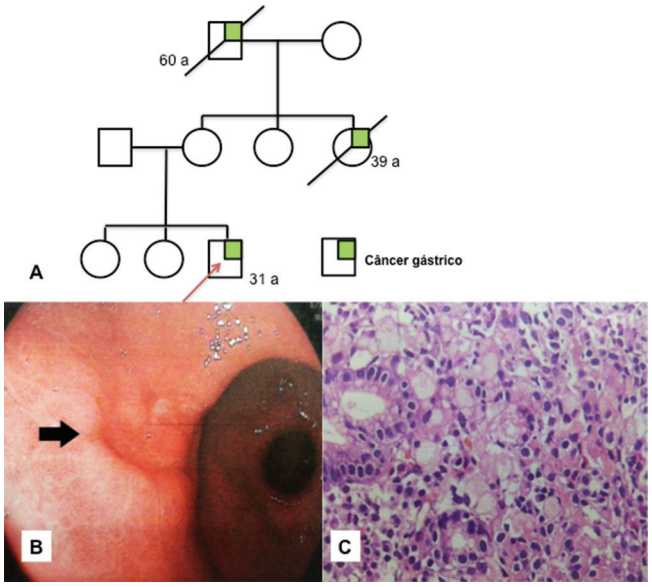

FIGURE 1 – A) Heredogram showing three cases of gastric cancer in three generations of the same family; B) videoendoscopy demonstrating gastric ulcer; C) gastric adenocarcinoma (H & E)

Staging showed no metastatic or locally advanced disease. The patient underwent laparoscopic radical gastrectomy with derivation in Roux-en-Y and lymphadenectomy D2. The pathology of the surgical piece showed the presence of poorly differentiated adenocarcinoma, diffuse pattern, with cells in signet ring, free surgical margins and no lymph node compromised histologically in 29 dissected.

After the operation, according to guidelines of the International Gastric Cancer Linkage Consortium, was collected the blood and carried the mutation research of the HRC gene 1. It was analyzed by technique of polymerase chain reaction (PCR) with subsequent bidirectional sequencing of all coding exons and introns-exons junctions CDH1 gene. It was identified as heterozygous deletion of two pairs of bases 1763-1764 in the nucleotide, resulting in frameshift mutation at codon 588 (c.1763-176delTG; pVal588Glufs * 2) in CDH1 gene.

DISCUSSION

This syndrome affects individuals of the same family at young age[7,10]. It is defined by the presence of germline mutations of CDH1 gene, acquired by autosomal dominant, reaching penetration rates of up to 80% for both genders[4,7,10].

The CDH1 gene is located at 16p22.1 gene. It consists of 16 exons encoding a transmembrane protein called E-cadherin. It holds cells together in intra and extracellular domains that communicate with neighboring cells by other proteins called catetines[4].

The genetic alterations detected in the CDH1 gene are mutations, frameshift mutations and deletions. Mutations are more related to the occurrence of hereditary diffuse gastric cancer syndrome, the codon 1003, located in exon 7 the most frequent site of changes. In the present study, a frameshift mutation was detected in codon 588, the first of its kind to be described in the literature. The real impact of this mutation on the phenotype is unknown[6,7].

Clinical diagnostic criteria are established by the "International Gastric Cancer Linkage Consortium" that defines as the presence of two or more cases of diffuse gastric cancer in first or second degree relatives being one of them diagnosed before age 50, or three or more cases of diffuse gastric cancer diagnosed in first or second degree relatives, regardless of age[7,10].

Once established the clinical and genetic diagnosis of the syndrome, it is important to extend the research to the direct family members, and women crawl them for lobular breast carcinoma. Other syndromes may be present at diagnosis, such as colorectal cancer not polypoid, Li-Fraumeni syndrome, familial adenomatous polyposis, Peutz-Jeghers syndrome and Cowden[5].

Prognosis is dependent on early diagnosis and total gastrectomy indication even in the absence of invasive disease[7,9,10].

Total gastrectomy is the most efficient way to eliminate the risk of developing gastric cancer, including how to limit proximal surgical resection along the distal esophagus and eliminating any risk of residual mucosa tissue on cardia[9,10].

Endoscopic surveillance of patients at risk is not the first choice, considering the high penetration rates of the gene, as well as the tendency to multifocality of neoplastic cells and sometimes more initial stages, and the neoplasia below the mucosa. The diffuse histological pattern also carries a worse prognosis due to higher frequency of histologically positive lymph nodes and peritoneal implants[6,9,10].

Only in patients who refuse surgical treatment is indicated follow-up with serial endoscopies, added to chromoscopy and immunohistochemistry in biopsies, targeting respectively the largest detection of early gastric cancer and signet ring cells[6].

Norton et. al.[8] demonstrated the occurrence of early gastric cancer associated with the syndrome, indicating early radical gastrectomy in six patients. In all surgical specimens had multifocal adenocarcinoma, invasive and histological pattern of signet ring.

Despite the risks of surgery, such as fistulas, infections, dehiscence and anesthetic complications may reach up to 22% of morbidity and mortality of 4%, the benefits of early operation are superior to endoscopic surveillance or the diagnosis of neoplasia in symptomatic patients. Laparoscopy becomes the major route of surgical approach, since the syndrome is prevalent in young patients and its early identification enables the provision of prophylactic operation in the absence of advanced disease, with faster surgical recovery and earlier return to daily activities[2,10].

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ABCDDV/1111
ABCD Arq Bras Cir Dig 2015;28(2):151
DOI:http://dx.doi.org/10.1590/ S0102-67202015000200018

Letter to the Editor

RETRORECTAL TUMOR: A CASE REPORT OF A PATIENT WITH "SCHWANNOMA"

Tumor retrorretal: relato de caso de um paciente com "schwanoma"

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Conflicts of interest: none

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Received for publication: 22/04/2014
Accepted for publication: 24/02/2015

INTRODUCTION

Injuries that affect the presacral space are rare, and are part of a heterogeneous group of entities of primary concern to colorectal surgeons. These lesions are located in anatomically difficult area to be addressed, hence the need for prior surgical planning, as well as knowledge of the major diseases that affect this region.

The incidence of these lesions varies from 1: 40.000 to 1: 630,000[7], being more common in females between 40 and 60 years[8].

CASE REPORT

Man of 94 years showed changes in bowel habits few months ago. Digital rectal examination showed solid pelvic mass with no apparent invasion of the rectal mucosa; it is not possible to predict its upper limit. Computed tomography of the pelvis showed solid-cystic lesion in proximity to the sacrum bone and rectum, with apparent cleavage plane between adjacent structures (Figure 1). He underwent transperitoneal resection of the lesion. Major bleeding occurred during surgery, which was controlled, requiring blood transfusion with four blood cell concentrate units.

Postoperative was without morbidity and discharged on the 7th day after surgery. The pathology confirmed it was schwannoma of low-grade without evidence of malignancy (Figure 2).

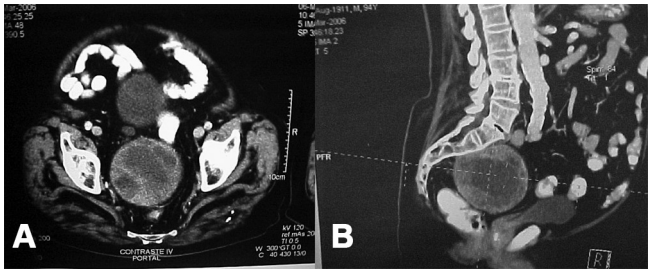

FIGURE 1 - Tumor images located in the presacral space

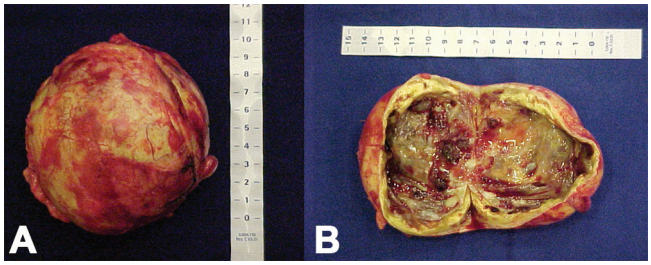

FIGURE 2 - Encapsulated tumor (10 cm in diameter - A) and open aspect (B) showing mucoid material inside

DISCUSSION

Although most retrorectal lesions are congenital, most patients do not have a previous positive family history; the most common clinical presentation is asymptomatic mass found to proctologic examination[5]. The retrorectal masses are palpable on rectal exam by up to 97% of cases[7].

Due to the angular change caused by the puborectalis muscle mass patients often have changes in bowel habits with a tendency to constipation, feeling of incomplete evacuation or thin stools.

A flexible sigmoidoscopy is useful for viewing the invasion of the rectal mucosa by the tumor and its upper limit, for correct surgical management.

Imaging tests are essential for accurate diagnosis, and the MRI is superior to CT in the characterization of pelvic masses for the presence of bone invasion or neural involvement[2].

Treatment is mainly surgical. Many of these injuries,

In the article “HEREDITARY DIFFUSE GASTRIC CANCER: LAPAROSCOPIC SURGICAL APPROACH ASSOCIATED TO RARE MUTATTION OF CDH1 GENE”, with the number of DOI: /10.1590/S0102-67202015000200017 published in the periodical Arquivos Brasileiros de Cirurgia Digestiva, 28 (2): 149 -151, on page 149:

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