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Case Report

Multiple colon polyposis associated with hereditary colorectal cancer

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ABSTRACT

Currently, colorectal cancer ranks third in presentation worldwide and second in terms of mortality. Being its developed by a multifactorial process, by non-modifiable factors such as age, history of inflammatory bowel disease, family history of colorectal cancer, adenomatous polyps or inherited syndromes as well as the modifiable ones. Consumption of fats in more than 30% of the daily diet, consumption of red meat in more than 100 grams per day, consumption of sausage in more than 50 grams per day, consumption of tobacco in 2 to 5 cigarettes day, alcohol consumption at more than 40 g/d in women and more than 60 g/d in men/day, coffee consumption >300 mg daily, stress and lack of physical activity. Likewise, a 27% higher risk of present colorectal cancer in patients with type 2 diabetes than in non-diabetic controls, suggestively due to the proinflammatory state. This condition has been associated with several intestinal and extraintestinal conditions, but especially with intestinal polyposis. The impact that modifiable risk factors have on the development of the disease becomes important due to its natural evolution. Below we presented the case of a woman diagnosed with multiple colon polyposis associated with hereditary colorectal cancer.

Keywords: Familial adenomatous polyposis, Hereditary colorectal cancer, Colonoscopy

INTRODUCTION

Familial adenomatous polyposis is a hereditary disease with autosomal dominant characteristics which occurs approximately in 1:10,000 of the general population, being associated with various extracolonic manifestations such as gastric and duodenal polyposis, desmoid tumors, among others.[1-4] Familiar adenomatous polyposis will develop colon cancer at 50 years, compared to hereditary non-polyposis colon, which have a risk of progression nonpolyposis colon, which have a risk of progression of malignancy of 70% to 82%.[5] According to a study carried out in 2019 in Cuba by Trujillo-Pérez et al the most common tumor distribution is: rectus sigmoid: 41.37%; ascending colon 37.92%; cecum colon: 12.64%; transverse colon: 9.19%; and descending colon: 5.74%.[6]

The most important diagnostic approach for this condition is screening for patients with a family history, although one-third of all cases are thought to be due to de novo mutations.[2] Patients without a family history represent a great diagnostic challenge with negative prognostic estimates. Surgical treatment aimed at preventing the development of colorectal carcinoma is indicated when familial adenomatous polyposis is diagnosed, especially when the risk of its appearance is very high.[5]

If treatment is not provided, an average individual with classic familial adenomatous polyposis will develop colorectal carcinoma around the age of 40 years.[3] The development of colorectal carcinoma is inevitable when the disease runs its natural course in patients who do not undergo treatment which basically consists of surgery and lifestyle changes.

We reported a case of familial adenomatous polyposis and discussed its relationship with hereditary colorectal cancer.

CASE REPORT

A 66 years old female with a first-degree family history diagnosed with colon cancer, diagnosis of arterial hypertension for 30 years, multiple gastric polyps diagnosed in 2019 by biopsy with hyperplastic characteristics, colonoscopy in November 2019 with colonic polyp with hyperplastic characteristics by histopathology; three hospitalizations for anemia and gastrointestinal bleeding requiring blood products, smoking from 29 to 64 years of age with consumption of 5 cigarettes a day with an TI of 9. Being admitted for severe anemia (admission laboratories: leukocytes: 3,840 10×3 /ul, hemoglobin: 6.4 mg/l, hematocrit: 24.7%, platelets: 270,300 10×3 /ul, neutrophils: 5, 900 10×3 /ul).

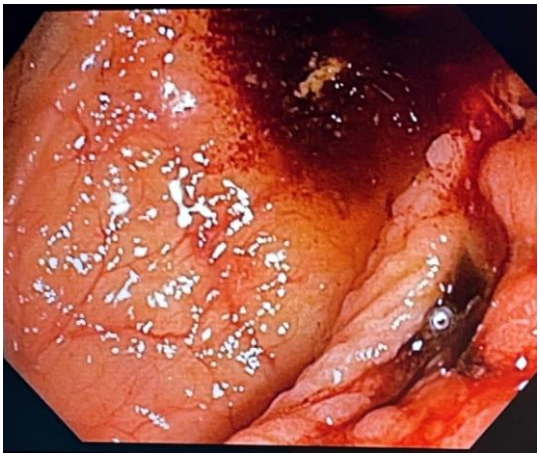

Figure 1: Rectosigmoidoscopy shows multiple sessile villous polyps of various diameters. Multiple colonic adenomatous polyposis and stenosing exophytic neoplasm of the hepatic flexure, compatible with adenocarcinoma.

An endoscopy was performed with the following results: multiple gastric polyposis, multiple colonic adenomatous polyposis, stenosing exophytic neoplasm of the hepatic flexure, compatible with adenocarcinoma biopsies were taken of polyps in the anal margin, rectum and neoplasia of the hepatic flexure of the colon (Figure 1). Tumor markers were elevated like carcinoembryonic antigen: 22.54 ng/fl, alphafetoprotein: less than 2, ca125: 18.1 U/ml, Ca 19-9: 6.92 U/ml and a simple and contrast-enhanced thoracoabdominal computed tomography showed activity colon tumor at the level of the hepatic flexure with wall thickening of 12-15 mm with increased density in the surrounding fat, without evidence of lesions compatible with metastasis. Histopathological study of biopsies reported colon adenocarcinoma in exophytic tumor of the hepatic flexure (Figure 2).

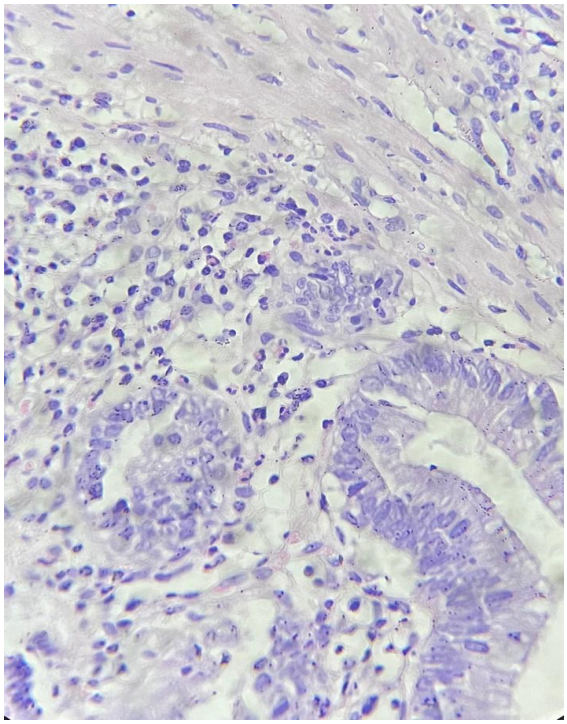

Figure 2: Histopathological study of biopsy from stenosing exophytic tumor of the hepatic flexure shows mucosa of the sigmoid colon with tubular adenoma and high-grade focal dysplasia (10%).

DISCUSSION

Colon cancer currently occupies the 3rd place worldwide, presenting an increase in its prevalence due to the greater presentation of risk factors in daily life as well as its genetical load.[7]

According to the case presented, the patient had factors as first grade relatives who died of colon cancer at an early age as background recognized risk factors for colon cancer such as work consumption, obesity, high-fat diet, stress and mainly a history of first-degree relatives with polypomatosis and colon cancer at an early age.[8]

Presenting as personal pathological history 2 years of diagnosis of multiple colonic polyposis, the main route of degeneration towards neoplasia being the activation of KRAS and the MYC oncogene through the chromosomal instability pathway.[9]

In addition to this, the patient presented the most common type of colon cancer at an early stage of the disease, which represented a good prognostic factor for disease remission.

Presenting the second most common site of location, being in the ascending colon, exactly in the hepatic angle of the colon.[6-10]

The genetic study and the correct protocolization of the patients who had a first-degree hereditary family history with colon cancer was important as well as the correct follow-up of patients diagnosed with polyposis with annual colonoscopy, seeking an early diagnosis of degeneration towards neoplasia.

CONCLUSION

Colorectal cancer is a pathology in which there is an increase in its incidence due to the persistence and increase in modifiable risk factors as well as the persistence of its high heritability. Our patient presents both modifiable risk factors such as smoking with compatible consumption for increased risk of colon cancer according to the bibliography as well as her history of first-degree relatives who died at middle ages due to colon cancer, her personal history of multiple polyposis, coupled with being at an age above which there is an increase in the prevalence of the disease, even though it is found in a good stage, which is why, with adequate therapy, a good prognosis for short-term survival is expected compared to more advance stages of the disease.

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