

A Rectocolonic Adenomatous Polyposis Discovered by an Acute Liver Colic

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Abstract

Familial adenomatous polyposis (PAF) is usually revealed by digestive signs such as gastrointestinal bleeding; and transit disorder or diagnosed by family screening in the event of an index case. In all cases; if there is not a prophylactic colectomy; PAF degenerate. We report the case of a patient who doesn't have symptoms of colonic diseases and have a rectocolonic adenomatous polyposis discovered by an acute liver colic

Keywords: *Familial Adenomatous Polyposis; Degenerate*

Introduction

Liver colic is a sign which have a variable etiologies. His link with PAF isn't common. We report here the case of a patient who doesn't have symptoms of colonic diseases and have a rectocolonic adenomatous polyposis discovered by an acute liver colic.

Presentation of the Case

A 43 years old man was referred from the urgency department for a persistent liver colic appeared 10 days ago. He had no history. He had no fever and no jaundice.

The clinical examination of the abdomen noticed an hepatomegaly: liver size at 17 cm.

An abdominal ultrasound was done and revealed several hepatic masses suspected to be metastases; ascites; and mesenteric and celiac lymphadenopathy.

The patient had normal gastroscopy.

Colonoscopy showed multiple sessile and pedicled polyps from the rectum to the left colon. Some of them were suspected of malignancy because of their aspects (Figures 1).



Figure 1: PAF.

Progression on the left colon was not possible because of a risk of perforation; Biopsies were performed from different sites of suspected polyps.

The pathological examination was in favor of low grade tubular adenomas

We supposed that was due to the biopsies and uncompleted colonoscopy.

Liver biopsy of hepatic masses confirmed the diagnostic of liver metastasis of a colonic adenocarcinoma.

A body scan showed left colonic cancer with liver metastases and peritoneal carcinomatosis.

Unfortunately, the patient died few days after his hospitalisation.

Discussion

Familial adenomatous polyposis (FAP) is an inherited disease characterized by an autosomal dominant transmission. Its prevalence is evaluated between 1 - 8 per 10,000 individuals [1] and is due to a germline mutation in the adenomatous polyposis coli (APC) gene. *De novo* gene mutation in the APC gene can be noted in 30% of patients without a history of FAP. This mutation is responsible of the early development in the second decade of hundreds to thousands colorectal adenomas polyps (> 100) and can be associated with several extra-colonic manifestations. Adenomatous polyps are usually discovered during endoscopic evaluation for symptoms such as gastrointestinal bleeding or during routine screening in individuals with a known family history of FAP [2].

FAP is considered as the second most common inherited colorectal cancer syndrome. It is the cause of 1% of colorectal cancers.

The risk of colorectal cancer at the age of 40 years old is evaluated at 100% [2]. It was the same case of our patient. However he doesn't have any digestive sign such transit trouble; melena or rectal bleeding.

In addition, he has no family history of FAP, so he most likely had a *de novo* gene mutation in the APC gene because he had one hundred to several thousand polyps since the other FAP mutations are responsible for the appearance of a moderate number of polyps. Arrived at his forties; the polyps degenerated and came into a stage of carcinomatosis with liver metastases.

Conclusion

FAP degenerates into cancer which encourages family screening if presence of index cases. However when it's due to *de novo* mutation, the diagnostic can be delayed and expose to the risk of an invasive colonic cancer.

Conflict of Interest

No conflicts of interest.

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