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FAMILIAL ADENOMATOUS POLYPOSIS

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Clinical History

A 41-year-old female was admitted into Nan Fang Hospital for severe abdominal pain with bloody-mucoid stool for a month. The symptoms started a year ago without obvious causes and she did not have any systemic treatment. The patient felt fatigue and loss of weight for the last three months and increased frequency of bloody-mucoid discharge from 2-4 times/day to 10 times/day for the last month. Two weeks ago the patient had a proctoscope with biopsy in Pan Yu people's Hospital. The pathological diagnosis was rectal villous adenoma with focal malignant changes. Rectal examination in this hospital found a rectal mass, 4 cm from the anus, longitudinal growing and occupying a quarter of the circumference. Further colonofiberscope diagnosis was familial polyposis of colon. Family history showed that her father died of lung cancer, her mother died of colonic cancer and her brother and sister were healthy. A total colo-rectectomy with ileostomy was performed.

Pathology Report

The specimen was a segment of colon measuring 79 cm in length with multiple polypus varying in size, from 0.1 mm to 30 mm in diameter. Situated at 15 mm from the pectinate line there was a polypoid mass, 45×40×18 mm, situated at 75 mm and 140 mm from the pectinate line there are two ulcerated masses measuring 40×30 mm and 30×25 mm respectively. The surrounding mucosa was elevated.

The pathological diagnoses was polyposis; moderately colo-rectal multiple polyps (>100) consistent with familial differentiated adeno-carcinoma infiltrating through full thickness of the bowel wall; sigmoid flexure tubulovillous adenoma with superficial malignant changes; lymph node metastasis carcinoma (6/33).

Discussion

Familial adenomatous polyposis (FAP) also called familial adenomatous is an autosomal dominant genetic disease. About 1/10,000 population carries this abnormal gene. Polypus is its surface-type mark. The mutation of APC is relation with FAP tumorigenesis. APC is a suppressor gene, it is reported the APC germ-line mutation may be response to 67% FAP. Theoretically, if a patient carries this gene, the half of the children can be involved. In fact, only 8% clinically present with FAP. Pathologically, the entire colonic mucosa can be involved by multiple, various polypus from 150 to 5000, or even more. Most patients have 500-2500, average 1000 polypus. Usually the minimal number to diagnose FAP is 100 polypus; if less 100, it is called multiple adenoma. FAP involves the rectum the most, but never involves the small bowel. Small polypus in FAP is just mucosa millet papules. FAP has a tendency of malignant changes. Usually, 2/3 of the patients are associated with carcinoma in their the first visit. Malignancy always starts from adenoma (polyp) itself but not the mucosa in between. It takes about 10 year from adenoma to develop carcinoma. Cautery or surgery is the treatment. FAP with malignant changes should be treated as carcinoma.