FAMILIAL ADENOMATOUS POLYPOSIS COLI

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Key-word: Intestinal neoplasms

Background: A 53-year-old male patient with a familial history of colon carcinoma presented with stool changes and abdominal cramps. There was no weight loss nor signs of blood in the stool. An endoscopic colonoscopy procedure showed multiple rectocolic polyps but was incomplete due to a stenosing mass at the splenic flexure. Biopsies showed multiple adenomata and an invasive adenocarcinoma at the splenic flexure.
Work-up

On CT colonography (TRM (translucency-rendering-mode) using CAD (computer aided-detection) (Fig. 1) an astonishing number of 44 pedunculated and sessile colorectal polyps is noticed.

On CT colonography (TRM (translucency-rendering-mode), overview, in addition to the colorectal polyps, a stenosing carcinoma is demonstrated at the splenic flexure of the colon (arrows).

CT colonography, detailed views of the stenosis at the splenic flexure of the colon (Fig. 3) shows on axial section (A) a localised wall thickening at the left hemicolon transversum with irregular narrowing of the lumen suggestive for carcinoma. On the endoluminal view (B), the carcinoma presents as a broad-based attachment to the colon wall.

CT views at the level of coincidental CAD–detected lesion at the preterminal ileum (Fig. 4) shows on axial section (A) a polyp (diameter: 5 mm) with the same HU-density value as the colonic polyps and TRM aspect of soft tissue, suggestive for small intestine polyp. Images obtained during pillcam-procedure (B, C) confirm the intestinal polyp.

Radiological diagnosis

The endoscopic and CTC findings, combined with the familial history of coloncarcinoma made the diagnosis of familial adenomatous polyposis coli with invasive adenocarcinoma at the splenic flexure. As a coincidental finding a small intestine polyp was visualised on CTC.

Discussion

Familial adenomatous polyposis (FAP) coli is an autosomal-dominant syndrome, most commonly caused by mutations in the APC-gene at chromosome 5q21. Patients with FAP develop numerous polyps from early teenage years and onwards, mostly in the large bowel but also in the small intestine.

While these polyps start out benign, malignant transformation into colon cancer can occur when not treated. Therefore, strict surveillance is recommended for all FAP patients and at risk family members, but a prophylactic colectomy could be recommended to reduce the risk of colorectal cancer.

Although endoscopic colonoscopy has the advantage of offering the ability to take biopsies, CTC can be a very helpful tool when endoscopic colonoscopy is incomplete.

Furthermore with the aid of dedicated software tools, such as CAD and TRM, in our opinion, less obvious lesions can be picked up. Furthermore, CAD and TRM enable the radiological view to be extended.

In the presented case a small intestine polyp was detected, as coincidental finding in a patient with FAP.

Bibliography