

ERCP; (5) fibrotic change with histopathologic lymphocytic infiltration; (6) absence of symptoms or only mild symptoms, usually with an absence of acute attacks of pancreatitis; (7) constriction of the common bile duct in the pancreas with proximal dilatation, and frequent cholestasis and hyperbilirubinaemia; (8) no pancreatic calcification; (9) no pancreatic cysts; (10) occasional association with other autoimmune diseases; and (11) effectiveness of steroid therapy [2]. This condition has also been named sclerosing pancreatitis and idiopathic fibrosing pancreatitis [3,4]. Autoimmune pancreatitis has been described in association with Sjögren syndrome, Crohn's disease and ulcerative colitis [5]. Its incidence in Japan is 5.93 patients per 100 000 inhabitants per year, with a prevalence of 4.78 patients per 100 000 inhabitants [6]. Sjögren syndrome was associated with 25% of the cases of autoimmune pancreatitis in a review of 40 institutions in Japan [7]. The second most common associated disease was primary sclerosing cholangitis (13%), followed by systemic lupus erythematoses.

In line with the Japanese criteria for the diagnosis of autoimmune pancreatitis, our patient had obstructive jaundice secondary to fibrosing pancreatitis, with a probable autoimmune aetiology associated with ulcerative colitis. In some patients with Crohn's disease and ulcerative colitis, this condition has been associated with antibodies against pancreatic juice [8]. Other authors have found high serum IgG4 concentrations, suggesting an autoimmune pathogenesis [9]. Glucocorticoid therapy induces clinical remission and would be the first-line treatment [6,9], although many patients require subsequent operation [3].

Autoimmune pancreatitis should be considered in young patients with obstructive jaundice [10],

especially those affected with chronic inflammatory or autoimmune diseases [11].

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Variation of a variation: Case report of attenuated familial adenomatous polyposis

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Abstract

Background. First described in 1988, attenuated familial adenomatous polyposis (AFAP) is a rare autosomal dominant precancerous condition of the gastrointestinal tract. Few reports have described adenocarcinomatous change in the

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gastroduodenal region thus far. *Case outline.* We report a case of AFAP presenting with extensive gastric polyposis and ampullary adenocarcinoma in absence of a positive family history of gastrointestinal cancer and a novel mutation.

Key Words: *Attenuated familial adenomatous polyposis, ampullary carcinoma, CHRPE*

Case report

A 35-year-old man was referred to us for evaluation of his abnormal liver enzymes. Gamma-glutamyl transpeptidase (γ -GT) level had risen to over 2000 IU/L and his CA 19-9 level was >1000 kU/L. Magnetic resonance (MR)-cholangiography showed the common bile duct to be slightly dilated but no stone or tumour was identified. Upper gastrointestinal endoscopy showed multiple gastric polyps within the proximal two-thirds of the stomach and a friable 1–2-cm villous polyp at the ampulla of Vater. Gastric biopsy revealed foveolar hyperplasia with atypia suggestive of adenomatous change and biopsy of the ampullary lesion showed severe dysplasia. At colonoscopy seven small non-malignant adenomatous polyps were removed from the left colon. Fundoscopy revealed multiple, bilateral congenital hypertrophy of retinal pigment epithelium (CHRPE) lesions.

The presence of widespread adenomatous lesions within the body and fundus and their potential for malignant transformation in view of his age, led us to perform a total gastrectomy and pancreaticoduodenectomy with a Roux-en-Y reconstruction. Histology of the resected ampullary lesion revealed foci of well-differentiated adenocarcinoma with microinvasion of the stroma.

The patient was referred for genetic counselling. There was no family history of bowel cancer or polyps.

The limited extent of involvement of the colon was consistent with AFAP. He was found to have the G126X mutation in exon 3 of the APC gene. This particular truncating mutation has not been described previously [1].

A CT scan at 9 months showed no signs of recurrence of his ampullary tumour. Follow-up colonoscopy at 1 year post surgery revealed normal mucosa and a biopsy taken from terminal ileum was identified as normal.

Discussion

AFAP is a rare precancerous condition of the gastrointestinal tract. Few reports have described adenocarcinomatous change in the gastroduodenal region thus far [2,3]. AFAP is a milder variant of familial adenomatous polyposis (FAP) transmitted in an autosomal dominant fashion [4]. It is caused by

mutations clustered either at the far 5' end (before codon 536) or at the 3' end (after codon 1596) of the APC gene [5]. Individuals with AFAP develop fewer polyps (average 40–50) and have a later onset of diagnosis of bowel cancer, at a mean age of 56 years [4,6]. AFAP usually affects the proximal colon with sparing of the rectum [4]. CHRPE is the most common extraintestinal finding in FAP. It is an oval pigmented lesion surrounded by a pale halo. They are rarely reported in AFAP patients, particularly AFAP patients with mutations in the 5' part of the APC gene [7]; 25% of AFAP cases arise sporadically while others may have affected siblings with unaffected parents. Such cases might result from a new APC gene mutation or gonadal mosaicism in a clinically unaffected parent [8]. While the correlation between the location of APC (adenomatosis polyposis coli) germline mutation and the development of AFAP has been well documented, the mechanism for AFAP remains to be elucidated [9,10].

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