Gastrointestinal manifestations in Cowden syndrome. Screening recommendations

Dear Editor,

Cowden syndrome (CS) or multiple hamartoma syndrome is a rare entity related to the PTEN gene. Its classic phenotype includes hamartomatous lesions in any localization — these patients have mainly cutaneous lesions but also gastrointestinal lesions are frequent. Other tumors related to CS are breast, thyroid and endometrial malignant neoplasms for which specific screening recommendations have been established (1,2). Recently, series of CS patients with gastrointestinal polyps of other nature as well as colonic adenocarcinomas have been published (3,4). Thus, colorectal cancer (CRC) is currently considered one of the diagnostic criteria of the PTEN hamartoma tumor syndrome spectrum (2) of which CS is a part (5). This has implied the novel inclusion of colorectal cancer (CRC) screening as part of the recommendations of some of the clinical guidelines.

Case report

66-year-old woman. Personal history of right breast carcinoma (52 years), endometrial carcinoma (53 years), follicular thyroid carcinoma (56 years), sphenoidal meningioma (63 years), renal angiomyolipoma (65 years) and left breast carcinoma (66 years). Father diagnosed with a gastric cancer (72 years). With the previous findings in addition to a characteristic phenotype (macrocephaly, benign cutaneous lesions and oral mucosa papillomas) a high clinical suspicion of CS was established, later confirmed by genetic testing. The patient was referred to our department for endoscopic assessment.

Esophageal acanthosis and three hamartomatous gastric polyps were found in the upper endoscopy. Fifteen polyps were resected in the colonoscopy (7 adenomas, 2 serrated adenomas, 4 hyperplastic polyps, 1 perineuroma and 1 submucosal leiomyoma). The study was completed by small intestine (SI) video capsule endoscopy, where multiple polyps from 5 to 10 mm in diameter were found (Fig. 1).

Discussion

CS has been associated to gastrointestinal polyps, mainly of the colon, and of different nature (hamartomatous, inflammatory, hyperplastic and adenomatous, among others), although the International Cowden Syndrome only considers hamartomatous lesions in its diagnostic criteria (2,3,6). CRC risk for CS patients was not defined until 2010, albeit several studies had described gastrointestinal neoplasms in cases with CS (7).

CRC screening for CS cases was included initially as a suggestion in the NCCN guideline in 2013. The 1.2014 NCCN guideline version established the recommendation of performing an initial endoscopy at age 35 with a 5-yearly endoscopic surveillance in CS patients (6). In our case, an upper endoscopy was performed in analogy to the recommendations for other hamartomatous polyposis. Furthermore, a SI video capsule endoscopy was carried out because of the high number of colonic and gastric hamartomas found in our patient. Nevertheless, an increased risk for gastrointestinal extracolonic neoplasms has not been established to date in CS (2,8), and additional examinations should only be performed in symptomatic cases (8). With respect to the SI study there are only two papers that describe the findings of capsule endoscopy in patients with CS (9,10). Thus, the recommendation for the screening of extracolonic gastrointestinal neoplasms is, at this moment, excluded from the clinical guidelines due to the lacking available evidence.
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References


Fig. 1. Gastrointestinal endoscopy. A. Colonoscopy: 20 mm subpedunculated polyp in distal transverse colon (asterisk), diverticuli (arrow). B. Gastroscopy: 20 mm congestive pedunculated polyp in prepyloric region (asterix). C. Video capsule endoscopy: Variable sized polyps in small intestine (asterix).