A case of Cowden Syndrome Associated with Multiple Gastric Polyposis

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Cowden syndrome is a rare autosomal dominant disorder that is characterized by multiple hamartomas in a variety of tissues and this is associated with germline mutations in the PTEN (phosphatase and tensin homologue) gene, which is the tumor suppressor gene located on chromosome 10q23.3. A 29 year-old woman was presented with multiple gastric polyps. The laboratory results were normal except for mild anemia, with a hemoglobin level of 11.9 g/dl. EGD revealed multiple gastric, duodenal polyps and esophageal acanthosis. Colonoscopy revealed possible hamartomatous polyps in the rectum. Under the suspicion of Cowden syndrome, sonography of the thyroid and breasts was carried out, which revealed multiple thyroid masses. The patient underwent total thyroidectomy. The pathology was reported as invasive follicular carcinoma. A gene study by direct sequencing showed the presence of a PTEN mutation (c.633C>A /p.Cys211X).

Predictable factors of histologic discrepancy of gastric tumor between the endoscopic forceps biopsy and endoscopic treatment specimen

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Purpose: Recently, variable gastrointestinal tract tumors including early stage malignancies are treated by endoscopic procedure. But, sometimes the discrepancies of histologic diagnosis happened between the pretreatment results and post-treatment results, especially in the aspect of malignancy. So, we retrospectively reviewed the patients who were not diagnosed as malignancy by forcep biopsy and were treated by EMR or ESD. Then, we investigated the predictable factors of histologic discrepancy between forcep biopsied and endoscopically resected specimen in terms of malignancy by comparing the clinical and endoscopic features through the lesions which were diagnosed as adenoma in forceps biopsy but diagnosed as adenocarcinoma in endoscopically resected specimen. Methods: From March 2008 to April 2011, 129 gastric tumor lesions (119 patients) which were not diagnosed as malignancy and were treated with endoscopic procedure were enrolled retrospectively. We compared the pretreatment endoscopic forceps biopsy results and post-treatment specimen biopsy results, then, analyzed the tumor characteristics. Results: When we compared the biopsy results, histologic discrepancies happened in 21 cases (16.3%) in terms of malignancy. Especially, these situations occurred more frequently in depressed lesions than in the flat or elevated lesions (41.7% vs 13.7%, \(p=0.012\)), and in the lesions diagnosed as high grade adenomas than in those diagnosed as low or moderate grade adenomas (33.3% vs. 11.1%, \(p=0.004\)). Conclusions: Overall, 16.3% of meaningful histologic discrepancies happened in terms of malignancy between the forces biopsy and post endoscopic procedure specimen, and those occurred frequently in depressed type lesion and those diagnosed as high grade adenoma significantly.