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LETTER TO THE EDITOR

The role of prophylactic gastrectomy in patients with juvenile polyposis syndrome

Dear Editor,

Juvenile polyposis syndrome (JPS) is a rare disorder of autosomal dominant inheritance characterized by multiple distinct juvenile polyps in the gastrointestinal tract [1]. The diagnostic criteria of JPS require the presence of at least five juvenile polyps in the colo-rectum, juvenile polyps throughout the gastrointestinal tract, and any number of juvenile polyps in conjunction with a family history of juvenile polyposis [2]. JPS has been linked genetically to mutations in either SMAD4 or BMPR1A present in 40% of patients [3]. The physiologic role of SMAD4 has been confirmed as a tumor suppressor gene and as such, JPS secondary to SMAD4 mutations has been considered a preneoplastic condition [4].

Patients with JPS most commonly present with colorectal polyps (98%) followed by gastric polyps (14%) and the small intestine (8.8%) [5]. Unlike BMPR1A mutations, SMAD4 has been involved in extra-colonic involvement of JPS and increased the risk of massive gastric polyposis [6,7]. Gastric polyposis in SMAD4 mutation carriers is discovered in patients with a median age of 41 years at diagnosis during routine investigations for iron deficiency anemia, abdominal pain, vomiting, early satiety, weight loss, hypoproteinemia or hypergastrinemia [8,9]. They can occasionally be asymptomatic and discovered incidentally. Patients often have numerous polyps involving most of the gastric mucosa, but some may have a lower number of polyps still occupying the majority of the gastric volume [8,9]. These polyps are at risk of intrinsic dysplastic changes leading to cancers mainly among patients with at least three juvenile polyps or a family history of juvenile polyps.

Overall, the risk of gastric cancer in JPS patients is approximately 21% with a probably higher risk in JPS secondary to SMAD4 mutations [10]. In the absence of clear guidelines, the optimal management of patients with JPS is debated between prophylactic gastrectomy and endoscopic surveillance with iterative polypectomies. Endoscopic surveillance may be associated with less morbidity but, unfortunately, its role in confirming a polyps' benignity is questionable. On the other hand, total gastrectomy provides a complete oncologic staging in addition to cure but is not free of complications with a 30-day mortality ranging between 1—6% [11]. Laparoscopic-assisted total gastrectomy was not associated with perioperative mortality but early postoperative morbidity rate reached 19% including wound complications (5.3%), ileus (2.3%), and anastomotic leak requiring reoperation (0.8%) [12]. Long-term complications were independent of the surgical method and include alterations in the eating habits, dumping syndrome, diarrhea and weight loss [13].

The ability of routine endoscopic surveillance to differentiate early cancer polyps limits its ability to recommended prophylactic or curative surgery. Multidisciplinary meetings including surgeons, endoscopists, genetists and dietitians are recommended for patient counseling to highlight the realities inherent in the data. There is evidence supporting genetic testing starting at age 4 in at risk children and upper and lower endoscopy surveillance starting at age 12 if symptomatic, at a frequency of 1 to 3 years on the basis of severity [14]. Later data suggested that patients at low genetic risk may no longer require frequent screening endoscopy, whereas gene carriers can be targeted for close endoscopic surveillance and early intervention to prevent the development of gastrointestinal cancers [15]. As such, we believe that prophylactic laparoscopic gastrectomy by an expert surgeon is well suited for symptomatic JPS patients and asymptomatic JPS patients with SMAD4 mutations or unmanageable gastric polyposis by endoscopy (> 50—100 polyps) [16,17]. Total gastrectomy should be privileged over partial gastrectomy to prevent the occurrence of gastric polyps in other gastric locations. Patients should be advised to the risk of potential short- and long-term complications. They should also be familiarized with post-gastrectomy diet and informed that it may take up to one year for weight stabilization and accommodation to dietary modifications to take place.

A thorough clinical and genetic workup should be considered in all patients presenting with numerous juvenile gastric polyps. JPS patients commonly have diffuse involvement of the stomach and may also be at high risk of cancer degeneration whenever associated with high-risk

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genetic features. Unfortunately, trials regarding the management of these patients are sparse. As such, the daily clinical practice is mainly based on expert opinions instead of universal standardized protocols. Our approach to JPS recommends endoscopic surveillance with polypectomy in asymptomatic patients with few gastric juvenile polyps (< 50—100 polyps). Prophylactic laparoscopic total gastrectomy is reserved to asymptomatic patients with high-risk genetic features or massive gastric involvement mutations or symptomatic patients independent of any genetic predisposition.

Disclosure of interest

The authors declare that they have no competing interest.

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