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A Giant Gastric Hyperplastic Polyp in Non-Familial Juvenile Polyposis of the Stomach: A Case Report

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1. Abstract

Juvenile Polyposis Syndrome (JPS) is a rare genetic disorder characterized by juvenile polyps of the gastrointestinal tract, which may also occur in people with no family history of the disorder. It usually involves the colorectum and more rarely the stomach. Juvenile polyps can vary in size from a few millimeters to over 5cm, and, although they have microscopically specific characteristics, the distinction between them and inflammatory polyps is often difficult. Here we present a case of 41 year old woman with no family history of polyposis who underwent esophagogastroduodenoscopy (EGDS) because of anemia that showed a huge and extensive gastric lesion (10 cm in diameter) and other small polyps scattered through the stomach lumen and duodenum. The histology report revealed the big lesion and the polyps were hyperplastic polyps with infiltration of inflammatory cells. Due to clinical history and EGDS findings, the patient underwent a pylorus-preserving gastrectomy, and to define a final diagnosis genetic test was performed and showed mutation in the SMAD4 gene. Thus non-familial juvenile polyposis of the stomach was diagnosed. An original feature of the current case was the rare finding of huge hyperplastic gastric polyp in a woman with rare non-familial JPS of the stomach. In fact, to our best knowledge this is the first report describing a so giant gastric polyp in JPS of the stomach in an adult woman. Furthermore, our report suggests that it is noteworthy to consider JP as differential diagnosis in patients with non-familial polyposis, anemia or hypoproteinemia and polyps histologically diagnosed as hyperplastic and performing genetic testing for confirming diagnosis and helping in the timely and appropriate management.

2. Introduction

Juvenile polyposis (JP) is an uncommon genetic syndrome characterized by the development of numerous hamartomatous and non-neoplastic polyps. Polyps predominantly occur in the colorectum, and more rarely in the stomach, duodenum, jejunum, and ileum. In approximately 25 percent of cases, it results from new mutations in the gene SMAD4 or BMPR1A and occurs in people with no family history of the disorder [1]. Juvenile polyposis of the stomach is not easy to diagnose because of rare phenotype and difficulty of differential diagnosis between juvenile polyp and hyper-

plastic polyp histologically by biopsy specimens. We herein report a case of huge hyperplastic gastric polyp in non-familial polyposis of the stomach who was treated by total gastrectomy and finally diagnosed as juvenile polyposis syndrome by genetic examination.

3. Case Report

A 41 year old female was admitted to our Internal Medicine Unit on November 2018 because of microcytic hypochromic anemia since three years for which she had been taking iron supplementation. Her clinical history was unremarkable except for an hypothyroidism treated with daily dose of levothyroxine. She referred metrorrhagia in the last 2 years why she underwent several gynecological examinations and was under hormonal treatment. She didn't have family history of cancer or polyposis. Clinical evaluation showed only skin pallor. Laboratory analysis confirmed microcytic hypochromic anemia (hemoglobin 8 g/dL) with iron deficiency and revealed hypoproteinemia (5.1 g/dl). Thyroid hormones and neoplastic markers were all within the normal range. Antinuclear antibody, anti-ds-DNA, and anti-transglutaminase antibodies were negative. During the work-up of anemia, esophagogastroduodenoscopy (EGDS) was performed that showed a very giant and extensive bulging gastric lesion (10 cm in diameter) located on the lesser curvature, involving three-quarters of the gastric wall (Figure 1), and other small (less than 1 cm in the largest diameter) pedunculated or sessile polyps scattered through the stomach lumen and very few in the duoedenum. The mucosal surface of the great lesion was erythematous and edematous with some erosions; biopsy specimens were collected and the pathological examination revealed that it was hyperplastic polyp of the stomach with infiltration of inflammatory cells. Helicobacter pylori test was negative. A total-body computed tomography (CT) scan confirmed the large gastric lesion that stretched over a width of 10 cm with mild enhancement after i.v. contrast administration (Figure 2). Due to the clinical history, the radiology and EGDS findings, by prior arrangement with the surgeons, the patient was discharged from our unit and admitted to the surgery unit, where she underwent firstly a complete colonoscopy showing two sigmoid colon polyps that were both excised, and, after that, a pylorus-preserving gastrectomy. The histopathology report identified the sigmoid colon specimens as a tubulovillous adenoma with focal high grade dysplasia and a sessile serrated polyp, and confirmed the stomach lesion as a giant hyperplastic polyp with multiple erosions. Microscopically, the stomach lesion was characterized by widely separated irregular glands lined by tall mucin-secreting foveolar cells within an edematous stroma infiltrated by lymphocytes, macrophages and eosinophils (Figure 3) and was associated with intestinal metaplasia of the adjacent gastric mucosa. In addition, a few and small duodenum polyps were also found and examined resulting as hyperplastic polyps as well. Because of morphologic similarities among gastric hyperplastic polyps, juvenile polyps and other hamartomatous polyps observed in different polyposis syndrome, and to define a final diagnosis, genetic test was performed and showed mutation in the SMAD4 gene. Postoperative course was uneventful and the patient was finally discharged after 9 days from surgery with a diagnosis of JPS.

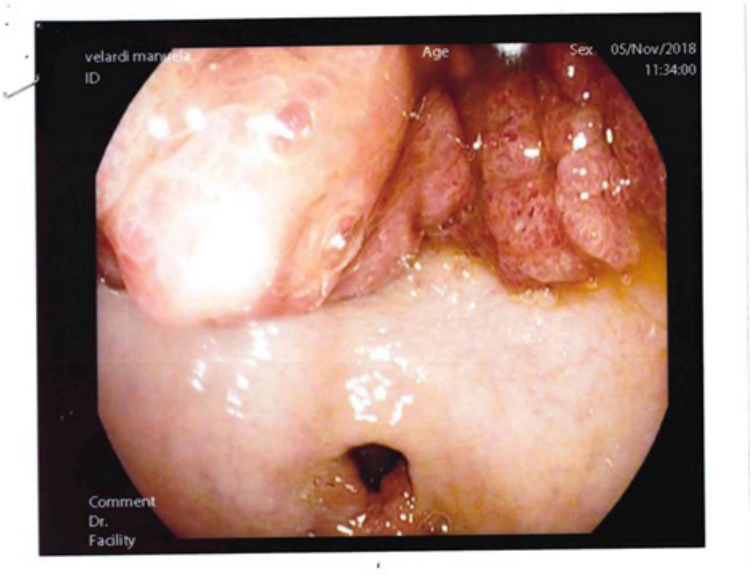

Figure 1: EGDS image showing a giant and extensive gastric lesion located on the lesser curvature

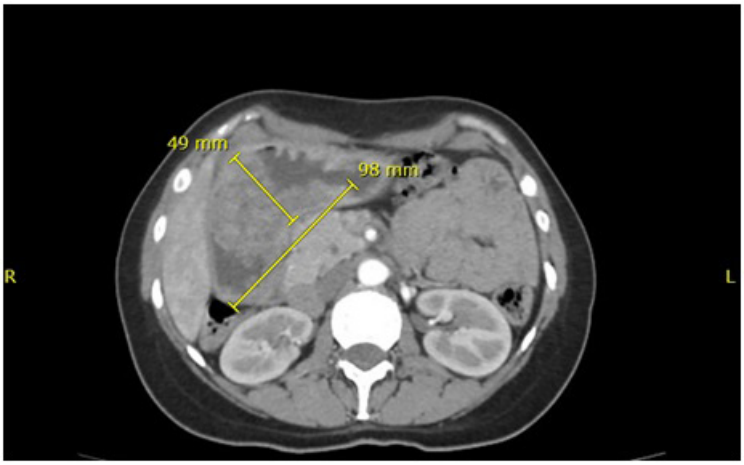

Figure 2: Contrast enhanced abdominal computed tomography (CT): arterial phase

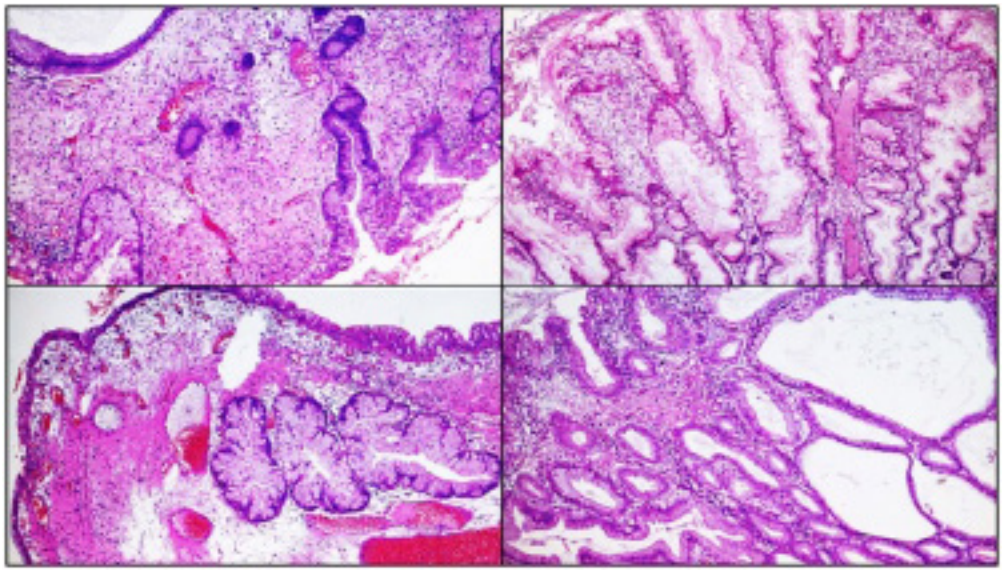

Figure 3: Hematoxylin and Eosin (H&E) staining, original magnification x 40.
Hyperplastic polyp of the stomach containing an edematous stroma infiltrated by lymphocytes, macrophages and eosinophils, with widely irregular and cystic glands lined by tall mucin-secreting foveolar.

4. Discussion

Juvenile polyposis syndrome is an autosomal dominant condition reported to be associated with germline mutation in the SMAD4 or BMPR1A genes. Mutations in these genes are observed in about 60% of patients with JPS, and in approximately 25 % of cases they occur in people with no family history of the disorder [2].

The pathology can generally presents as juvenile polyposis of infancy, generalized juvenile polyposis and juvenile polyposis coli. These last two forms usually occur later in childhood or in adult life.

Polyps in JPS predominantly occur in the colorectum, and rarely they can be found in the stomach in the absence of colorectal polyps. Watanabe et al. first reported a case of juvenile polyposis of the stomach in 1979 [3]. Macroscopically, the polyps can vary in size from a few millimeters to over 5 cm, while microscopically, a juvenile polyp is characterized by an abundance of edematous lamina propria with inflammatory cells and cystically dilated glands, however the distinction between an inflammatory and a juvenile polyp is often difficult [1].

To the best of our knowledge, this is the first report describing a giant gastric polyp in rare non familial juvenile polyposis of the stomach in an adult woman.

In our case, hyperplastic polyposis of the stomach was observed, with a huge hyperplastic polyp of the lesser curvature that could mimic a malignant tumor lesion. After gastrectomy, histology confirmed the big lesion as a hyperplastic polyp with multiple erosions and intestinal metaplasia of the adjacent gastric mucosa. The other small polyps scattered in the stomach and duodenum were also reported as hyperplastic polyps.

In the present case it was difficult to make a diagnosis of JPS prior to the genetic test because all biopsy specimens indicated hyperplastic changes and the patient had no family history of juvenile polyps.

Moreover, although the patient showed anemia and hypoproteinemia which are the most frequently clinical features observed in JPS (89% and 67%, respectively), these laboratory signs are not specific, making necessary different differential diagnoses.

Generally, hyperplastic polyps develop in chronic gastritis patients and an important predisposing factor is Helicobacter pylori gastritis [4]. However, duodenal and gastric polyps in JPS may occur that histologically resemble hyperplastic polyps or inflammatory polyps in many cases [5].

Previous studies showed that there is histological overlap among hyperplastic polyp, juvenile polyp, and inflammatory polyp of the colorectum [6], as well as between juvenile polyp and hyperplastic polyp of the stomach [5]. Indeed, most gastric polyps in JPS patients are indistinguishable from gastric hyperplastic polyps and have been diagnosed as hyperplasic polyps.

Due to these morphologic similarities among gastric hyperplastic polyps, juvenile polyps, and other hamartomatous polyps, and the frequent lack of family history in these patients, the genetic analysis is a helpful diagnostic test in cases of polyposis involving the stomach to distinguish among sporadic and isolated hyperplastic polyposis and the different syndromic gastric polyposis (i.e. JPS, Peutz-Jeghers syndrome, Cronkhite-Canada syndrome or Menetrier disease).

In this patient, only genetic analysis allowed to make a diagnosis of JPS. It is known that patients with JP due to SMAD4 mutation may have a juvenile polyposis-Hereditary Hemorrhagic Teleangiectasia (HHT) overlap syndrome [7]; however, in our case, there were not the clinical criteria for HHT diagnosis.

Moreover, upper gastrointestinal polyposis and gastric cancer have been associated with SMAD4 germline mutation [1], and a study revealed that 8.3% of polyps in JPS contain mild or moderate dysplasia and 14% patients developed cancers [8]. Thus, although rare, recognition of juvenile polyposis of the stomach is important in view of the consequences for patients and their family. Since there is a high occurrence of gastric cancer in these patients, especially when SMAD4 mutation is present, it is crucial not delaying the diagnosis and the surgical intervention.

In conclusion, an original feature of the current case report was the rare finding of a huge hyperplastic gastric polyp in a woman with non-familial polyposis of the stomach finally diagnosed as JPS. Furthermore, our report suggests that it is noteworthy to consider JP as differential diagnosis in patients with non-familial polyposis, anemia or hypoproteinemia and polyps histologically diagnosed as hyperplastic and performing genetic testing for confirming diagnosis and helping in the timely and appropriate management.

5. Conflict of Interest Statement

The authors declare no conflict of interest.

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