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EC GASTROENTEROLOGY AND DIGESTIVE SYSTEM

Case Series

Peutz Jeghers syndrome

Bertha Idrogo Regalado[1]* and Oscar Frisancho Velarde[2]

[1]*Resident Doctor, Department of Digestive Diseases, Edgardo Rebagliati Martins National Hospital, SALUD, Lima, Perú*

[2]*Assistant Doctor, Department of Digestive Diseases; HNERM Lima, Perú*

***Corresponding Author**: Bertha Idrogo Regalado, Resident Doctor, Department of Digestive Diseases, Edgardo Rebagliati Martins National Hospital, SALUD, Lima, Perú.

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Abstract

Peutz Jeghers syndrome is characterized by gastrointestinal hamartomatous polyps and mucocutaneous pigmentation, has a high risk of intestinal resections due to ischemia secondary to intussusception. The risk of digestive cancer is nine more than the general population. We reported five patients diagnosed with PMS, three presented intestinal intussusception, one had upper gastrointestinal bleeding and one died with pancreatic neoplasia.

Keywords: *Peutz-Jeghers Syndrome; Colonic Polyposis; Multiple Polyposis*

Introduction

SPG is characterized by gastrointestinal hamartomatous polyps and mucocutaneous pigmentation, has a high risk of intestinal resections due to ischemia secondary to intussusception. The risk of digestive cancer is nine more than the general population.

Hamartomatous polyps are located throughout the gastrointestinal tract being more frequent in the colon (60%), stomach (25 - 50%), small intestine (20 - 40%); They are variable in size, larger in the small intestine, and can be pediculated or sessile.

The mucocutaneous pigmentation is melanic, in the form of small macules of 1 to 5 mm around the mouth, eyes, perianal, fingers and toes; Mucosal lesions persist for long years, the skin may subside over time.

We reported five patients diagnosed with PMS, three presented intestinal intussusception, one had upper gastrointestinal bleeding and one died with pancreatic neoplasia.

Presentation of Clinical Cases

Patient 1: Woman, 29a with hyperchromic spots on lips and oral mucosa, endoscopic diagnosis, multiple gastric and colonic polyps, studied with jejunal intussusception due to a large polyp.

Patient 2: Male of 41st, antecedent brother with SPG (Pac 3), history of melanosis in lips, hands and feet. Also multiple colonic polyps, died by pancreas NM.

Patient 3: Male of 28a, brother of Pac 2. With melanosis on lips, hands and feet, multiple gastroduodenal and colonic polyps. He attended with obstruction of the small intestine by intussusception.

Patient 4: Male of 21a; melanosis on lips, hands and feet; also multiple colonic hamartomas. He required thin bowel resection due to intussusception caused by a polyp.

Patient 5: A 51-year-old male, melanosis in the palm of the hands, presented upper gastrointestinal bleeding, endoscopy: a 30x40 mm pedicle polyp on the anterior side of the duodenal bulb. Pathological anatomy: Hamartomatous polyp.

Clinical Cases

Patient 1: Woman, 29a with hyperchromic spots on lips and oral mucosa, endoscopic diagnosis, multiple gastric and colonic polyps, studied with jejunal intussusception due to a large polyp.

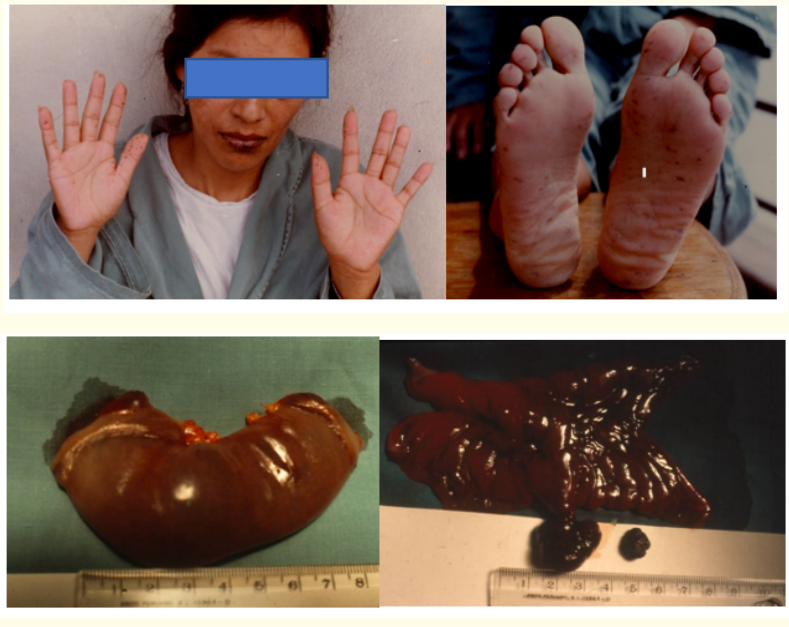

Figure 1

Patient 2: Male of 41st, antecedent brother with SPG (Pac 3), history of melanosis in lips, hands and feet. Also multiple colonic polyps, died by pancreas NM.

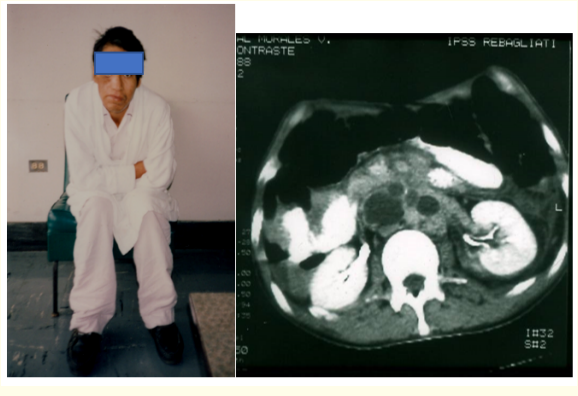

Figure 2

Patient 3: Male of 28a, brother of Pac 2. With melanosis on lips, hands and feet, multiple gastroduodenal and colonic polyps. He attended with obstruction of the small intestine by intussusception.

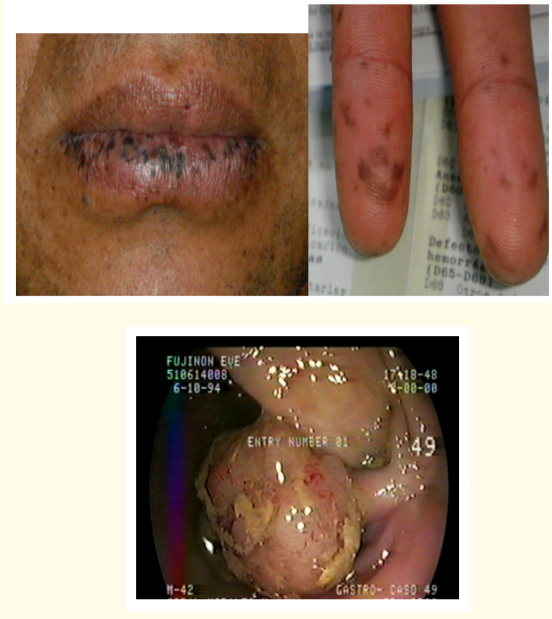

Figure 3

Patient 4: Male of 21a; melanosis on lips, hands and feet; also multiple colonic hamartomas. He required thin bowel resection due to intussusception caused by a polyp.

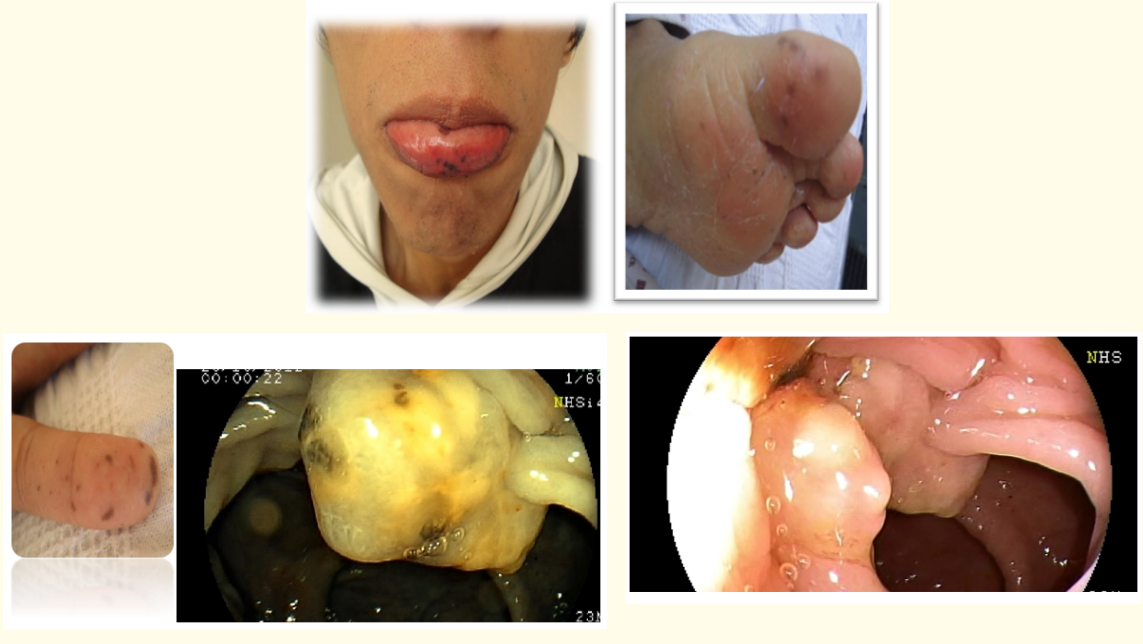

Figure 4

Patient 5: A 53-year-old male has hematemesis and mane (first episode 2 months ago), without endoscopic study. Hemoglobin 5.4 gr/dl.

Endoscopy: When transposing pylorus, a large pediculated lesion on the posterior side of a 40x50 mm multilobed bulb, eroded, oozing bleeding, is removed by polypectomy.

Diagnosis: Giant Polyp O-IP in the duodenum.

Pathological Anatomy of Duodenal Polyp: Hamartomatous Polyp of the Duodenum, negative for neoplastic cells.

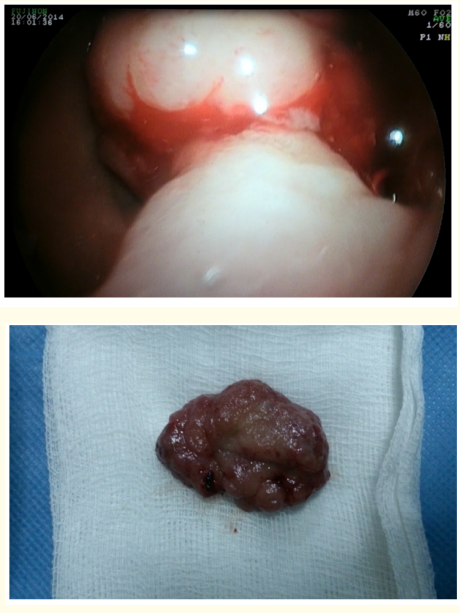

Figure 5

Discussion

Peutz-Jeghers syndrome is an autosomal dominant syndrome characterized by multiple hamartomatous polyps in the gastrointestinal tract, mucocutaneous pigmentation and increased risk of gastrointestinal cancer. It is a rare entity with an estimated prevalence of 1: 8000 to 1: 200,000, with equal involvement in men and women.

It is associated with the germ mutation in the STK11 (LKB1) gene that encodes a threonine kinase serine mapped on chromosome 19p.

There are two main types of clinical manifestations of SPJ:

1) Mucocutaneous pigmentation in the form of macules and multiple gastrointestinal polyps. The characteristic mucocutaneous pigmentation (melanin spots) are present in more than 95% of patients, present in lips, perioral region, palm of hands, oral mucosa and soles.
2) Hamartomatous polyps are present in most patients with JPS and can occur anywhere in the gastrointestinal tract. Polyps develop in the first decade of life and many patients are symptomatic between 10 and 30 years of age. The range of polyps is 1 to more than 20 per segment of intestine.

The risk of gastrointestinal cancer ranges between 37 and 93% with an average age of 42 years when making the diagnosis. The most common sites of malignant tumors of the gastrointestinal tract are the colon and pancreas and the most common sites of extraintestinal

tumors are the breasts. Women also have an increased risk of cervical tumors including malignant adenoma of the cervix and benign ovarian tumors known as „sexcord" tumors with annular tubules. Men with SPJ have a higher lifetime risk of testicular tumors of Sertoli cells.

The clinical diagnosis of SPJ requires the presence of any of the following:

- Two or more polyps with confirmed histology of Peutz-Jeghers.
- Any number of Peutz-Jeghers polyps detected in an individual who has a family history of Peutz-Jeghers or close relatives.
- Any number of Peutz-Jeghers polyps in an individual who has mucocutaneous pigmentation.

About 50% of patients are asymptomatic until before diagnosis, some patients present with intestinal obstruction caused by intussusception or occlusion of the gastrointestinal lumen caused by the polyp, as well as abdominal pain due to infarction, acute or chronic rectal bleeding caused by ulceration of the polyp. About 69% of patients experience intussusception during their lifetime, being more frequent in the small intestine.

Patients with Peutz-Jhegers clinical criteria should undergo a genetic mutation test in the STK11 gene. Genetic testing of an individual who meets the clinical criteria for SPJ serves to confirm the diagnosis; however, the presence of a pathogenic mutation of STK11 in a person who meets the clinical criteria for SPJ does not exclude the diagnosis of SPJ.

Of the five cases presented, 03 of them presented with intestinal intussusception caused by one of the polyps in the intestine, another one was diagnosed by upper gastrointestinal bleeding caused by laceration of duodenal polyp observed during endoscopy and one of them presented with pancreatic cancer, one of the most frequent malignancies as mentioned in the literature [1-9].

Conclusion

Peutz-Jeghers syndrome is an autosomal dominant disease characterized by the presence of multiple hamartomatous polyps in the stomach, small intestine and colon, accompanied by pigmented mucous skin lesions.

Patients with perioral or oral pigmentation and/or greater than or equal to 2 gastrointestinal hamartomatous polyps or a family history of Peutz-Jeghers syndrome should be evaluated for this syndrome.

Polyps may present with complications such as digestive bleeding, obstruction or invagination.

The search for GI cancer in patients with Peutz-Jeghers syndrome includes colonoscopy, upper digestive endoscopy and endoscopic capsule endoscopy.

In general, colonic polyps > 1 cm are resected.

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