ejpmr, 2020,7(2), 09-11

SJIF Impact Factor 6.222

EUROPEAN JOURNAL OF PHARMACEUTICAL AND MEDICAL RESEARCH

www.ejpmr.com

Case Study

ISSN 2394-3211

EJPMR

PEUTZ-JEGHERS SYNDROME: ABOUT A NEW CASE REPORT

M. A. Lkousse*, K. Gharbi, A. Elfarouki, S. Roudi, A. Aiterrami, S. Oubaha and Z. Samlani and K. Krati

Hepato-Gastro-Enterology Department, University Hospital Mohammed VI, Marrakech, 4000, Morocco.

***Corresponding Author: Dr. M. A. Lkousse**

Hepato-Gastro-Enterology department, University hospital Mohammed VI, Marrakech, 4000, Morocco.

Article Received on 23/11/2019 **Article Revised on 14/12/2019** **Article Accepted on 04/01/2020**

SUMMARY

Peutz-Jeghers syndrome (PJS) is an unusual gastrointestinal hamartomatous polyposis, inherited in an autosomal dominant pattern, defined by the association of periorificial lentiginosis with gastrointestinal, pulmonary and reproductive organs involvement. The case of a 18 years old man with an intussusception is reported. A polyp was proved to be the cause of the intussusception. Histologically, it was a hamartoma. Most of these patients have recurrent episodes of intussusception caused by polyps, with an increased risk of malignant disease, and require endoscopic screening and regular periodic monitoring.

KEYWORDS: Peutz-Jeghers, polyposis, hamartoma, lentiginosis, periodic monitoring.

INTRODUCTION

Peutz-Jeghers Syndrome inherited in an autosomal dominant pattern[1], is a rare disease, characterized by typical pigmented perioral macules, pigmented spots of the oral mucosa, and multiple hamartomatous polyps in the gastrointestinal tract.[2,3]

The skin signs are prominent, lentiginosis type is most commonly found on the lips and oral mucosa. Gastro intestinal polyps are the second cardinal sign of this syndrome and can be an early indicator of the disease when they manifest as complications such as gastrointestinal bleeding and occlusions.

People with (PJS) have an increased risk of cancer[4], with a risk estimated at 93%. The gastrointestinal tract (esophagus, stomach, small intestine, colon, rectum and pancreas), lung, prostate, breast and reproductive organs are the most affected.[5,6]

A few years ago, two groups of independent investigators defined the mutated gene responsible for PJS.[7,8,9] The gene was located on chromosome 19p34-p36 and is known as STK 11, a serine-threonine kinase that is involved in the regulation of growth control.[10,11,12]

CASE REPORT

An 18-year-old student, who appeared normal and had no familial history of PJS or any other polyposis syndrome. He is the eldest of two apparently healthy daughters. His mother first noticed pigmented spots around his lips at the age of five. The evolution was marked by the extension of these brownish lesions. When examined, additional pigmentation of the oral mucosa, eyes, toes and fingers was observed (Figure 1).

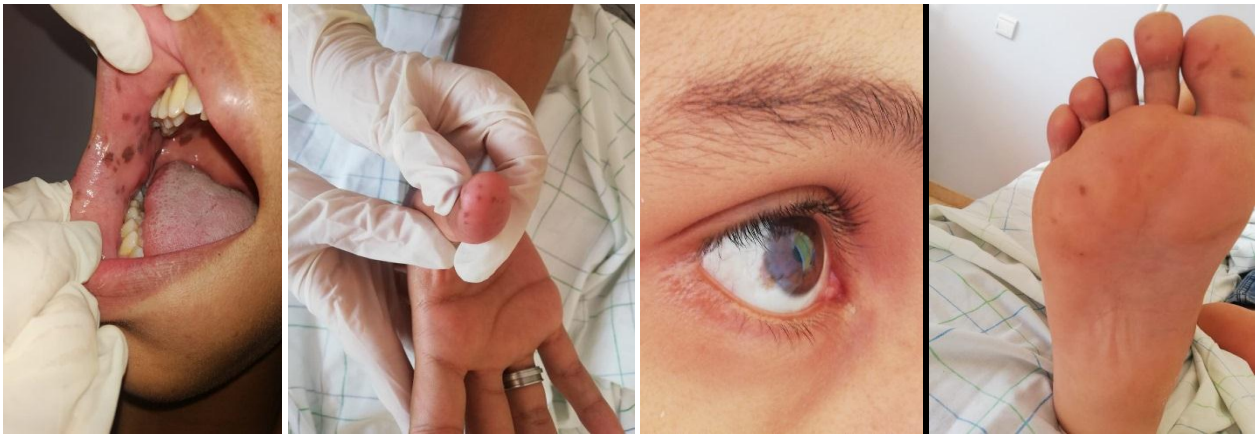

Figure 1: Brown Macules on The Oral Mucosa, Ocular Toes and Fingers.

He came to the emergency room for an acute bowel obstruction. Examination revealed an ileum intussusception secondary to a light-shedding polyp with intestinal necrosis (about 1 m in length), an intestinal resection carrying the polyp was made.

Macroscopically, at the opening of the resected part, we note the presence of five polypoid lesions, the largest was 3.5 x 4 x 2 cm. Histologically, it is a hamartomatous proliferation made of smooth muscle fiber within a hyperplastic mucosa confirming the clinical diagnosis of PJS (Figure 2). After the operation, the young boy was in good condition with close and regular supervision.

The diagnosis of (PJS) was made in the presence of the association of this hamartomatous polyps with peri orificial predominantly lentiginosis. The patient underwent a careful clinical examination, an abdominal and prostate ultrasound, looking for cancer, which were normal. The patient was well explained about the PJS, the risk of recurrence of complications, the risk of cancer at a very young age, and the periodic monitoring that he must do.

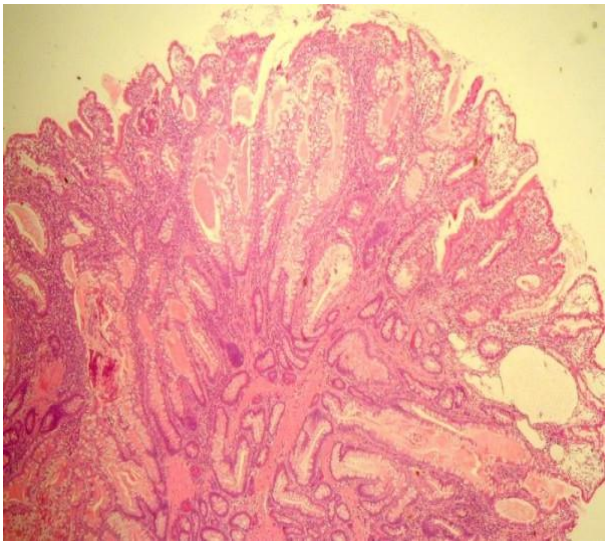

Figure 2: Histological Image Showing A Peutz-Jeghers Polyp (H and E Staining), Demonstrating Glandular Disorganization, Hamartomatous Appearance, and Branched Bundles of Smooth Muscle.

DISCUSSION

Peutz-Jeghers Syndrome is a rare autosomal dominant inherited disease, defined by the combination of periorificial lentiginosis (nose, lips, anal and genital areas), digestive, pulmonary and reproductive organ involvement. In 1921, Jan Peutz, a Dutch doctor reported a family case with gastrointestinal polyposis and mucocutaneous lentiginosis.[13] In 1949, an American physician named Harold Joseph Jeghers published a detailed description of patients with intestinal polyposis and skin pigmentation, the syndrome is named after the two physicians.[13] The prevalence is estimated at 1 / 200 000, without predominance of sex or race, the mean age of diagnosis is 22 years on a review of 75 cases.[13]

Diagnostic criteria proposed by Giardello and al for the PJS[14] requires histopathologic confirmation of gastrointestinal hamartomatous polyps and two of the following: small intestine polyposis, positive family history, spotted skin or mucous brown macules. In our case study, polyps in the small intestine were found during the operation, histologically confirmed as hamartomas, and the boy had typical mucocutaneous hyperpigmentation. A molecular genetic test for the STK11 gene confirms the diagnosis, but was not performed in our case.

Skin signs are a telltale of the disease, but they are not the first to appear. In all cases, they are lentiginous type which result from cutaneous and/or mucosal deposits of melanin producing hyper pigmented dark brown or black irregularly oval macules and are generally less than 5mm in diameter. They are almost constant but not pathognomonic, since a similar appearance can be observed in some normal subjects; they mainly concern the perimeters of the oral orifice (94%), eyes and nostrils (66%), hands and feet (62%); the perianal mucosa is much less frequently affected.[15]

Multiple hamartomatous polyps in the gastrointestinal tract are the hallmark of PJS. Gastrointestinal polyps are mainly found in the small intestine. They can also be found in the stomach and the large intestine.[16]

Histologically, the polyps have hamartomatous of tubulovillous architecture without dysplasia. The axis of the villi is characterized by bundles of smooth muscle fibers, which differentiates them from adenomas. The tubes are lined with tall cylindrical cells, sometimes enterocytic, sometimes caliciform, with increased mucosecretion. Generally, these polyps have no malignant potential.[17]

The (PJS) is characterized by a high risk of digestive and extra-digestive cancer. According to Lim et al, in a study of 240 patients with PJS, the risk of cancer is 1% before the age of 20; 19% before the age of 40; 63% before the age of 60 and 81% before the age of 70.[18] In their studies of 419 people with PJS, Hearle et al found the same results as Lim. The most common cancers in this analysis were digestive cancers. Breast cancer was the most common for extra-digestive cancers.[19] In our case, we have not yet detected the existence of cancer in our patient.

In PJS, polypectomy is recommended for any polyp larger than 1 cm in diameter because of the risk of complications. Bleeding, invagination or intestinal occlusion is an indisputable surgical indication. It should be kept in mind that complications may recur. Therefore, excessively extensive bowel resections should be avoided as far as possible.[20]

The management of (PJS) is based primarily on the monitoring and treatment of hamartomatous polyps. There is no consensus on the frequency of surveillance of these patients, but it is desirable that asymptomatic patients should have endoscopy every 2 years for monitoring and ablation of polyps. Magnetic resonance imaging has shown success as a monitoring modality for small intestine and testicular, abdominal ultrasound is performed for pancreatic cancer screening. In women, mammography and transvaginal ultrasound are performed every 1 to 2 years.

A blood count should be performed to detect anemia caused by blood loss.[21] There is no standard treatment for mucocutaneous pigmentation, which is present in the majority of patients. Treatment involves cryosurgery, dermabrasion and Q-switched laser.[21]

CONCLUSION

A monitoring protocol must be established for each patient with PJS to avoid complications. Although the frequency of PJS is low, it is important to be aware of it.

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