A Rare Case of a Juvenile Polyp of Patient with Peutz-Jeghers Syndrome, Complicated with Intussusception of the Small Intestine

Anna Rycyk1, Beata Kasztelan-Szczerbinska1, Halina Cichoz-Lach1

1 Department and Clinic of Gastroenterology with Endoscopy Unit, Medical University of Lublin, Lublin, Poland

Corresponding author: Anna Rycyk, Department and Clinic of Gastroenterology with Endoscopy Unit, Medical University of Lublin, Lublin, Poland; Email: aniarycyk@op.pl

Received: 6 Apr 2021 ♦ Accepted: 2 June 2021 ♦ Published: 31 Aug 2022

Abstract

We report a rare case of Peutz-Jeghers syndrome (PJS) in a 35-year-old female. The patient was diagnosed with PJS when she was 11 years old. She has remained under observation since then. We strongly believe that PJS is a very rare diagnosis. However, it can have serious complications such as the intussusception we observed in our patient. Her condition (recurrent abdominal pain and vomiting) in childhood required further diagnostic procedures. Although the diagnosis of PJS was made, among many resected polyps, one of them appeared to be a juvenile polyp. The diagnosis was confirmed in the histopathology report, which was incredibly unique. Genetic testing revealed LKB1/STK11 gene mutation. Clinicians should be aware of the malignant potential in the course of PJS. Hence, these patients require tailor-made management, long-term follow-up, and our particular attention.

Keywords
cancer, colonic polyp, gastrointestinal polyps, hamartoma polyp, Peutz-Jeghers syndrome

INTRODUCTION

Peutz-Jeghers syndrome (PJS) is a rare autosomal dominant inherited disorder that always requires early diagnosis. It is characterized by the presence of hamartomatous polyps in the gastrointestinal tract, typically found in the small intestine and colon.[1] The PJS patients have an increased risk of malignancy compared with the general population.[2] Thus, they require a long-term follow-up to assess the diagnosis, treatment of malignant changes and other severe complications secondary to Peutz-Jeghers syndrome. Considering the recent advances in treatment, early management of PJS individuals is essential to avoid an unnecessary surgical emergency.

CASE REPORT

Herein, we present a case of a 35-year-old female who was admitted to the Department of Gastroenterology in July 2020, with Peutz-Jeghers syndrome recognized in 1996. The physical examination revealed no abnormalities, and the patient denied any ailments. Her medical history started when she was 10 years old. She had exhibited recurrent abdominal pain and vomiting during her childhood. However, no mucocutaneous pigmentation was observed. Nevertheless, she did not have a history of PJS in a close relative, her condition required further diagnostic procedures.

Intussusception of small intestine

In December 1995, our patient underwent a resection of her small bowels due to an intussusception of the small intestine.
and its necrosis. During the surgery, two meters of the small intestine were found to be full of multiple polyps. Upper GI endoscopy showed many polyps of the antrum and the duodenum. Two months later, our patient underwent her first colonoscopy. Eleven polyps from the sigmoid colon and the rectum were removed. Five months later, 8 large intestinal polyps were resected endoscopically. Next, after 7 months, 30 polyps were found in the stomach, one of which was removed. During the next years, 23, 18, 12, 11, 14, 4, and 4 gastrointestinal polyps were removed in iterative colonoscopy procedures, consecutively.

Another surgery

It is a well-documented fact that patients with Peutz-Jeghers syndrome are at a high risk of developing both gastrointestinal and extraintestinal malignancies. Hence, our patient had a computed tomography (CT) scan of the abdomen. The CT scan indicated the presence of 60-mm focal changes in the left lumbar abdominal region. When she was 19 years old, in January 2005, on account of suspicion of a small intestine tumor, she underwent another resection of the small intestine localized 100 cm distally to the ligament of Treitz with intraoperative polypectomy of 51 polyps. The postoperative histopathology report excluded malignancy. Thus, the focal changes appeared to be a Peutz-Jeghers-type polyp that is defined as a hamartomatous polyp with the pathological features of Peutz-Jeghers syndrome but lacking the pigmentation and heritability.[3]

In March 2006, our patient underwent a uterine curettage due to a benign hydatidiform mole. In August, 2007, seven Peutz-Jeghers polyps were removed. One year later, enteroscopy was performed during which another 7 hamartoma polyps were found 150 cm distally to the stomach and were excised.

Additionally, two years later in March, July, and October, while undergoing enteroscopy, 20 polyps, with diameters ranging from 1 cm to 5 cm, were removed; 10 polyps with 1-3 cm in diameter; and several dozens more, respectively. Interestingly, histopathology report showed a fragment of a juvenile polyp of the small intestine, and the rest of the detected polyps were described as Peutz-Jeghers polyps. This discovery of the juvenile polyp in our patient’s small intestine did not affect her model of screening. Moreover, one juvenile polyp does not fulfill the criteria of juvenile polyposis syndrome (JPS). However, it is a unique finding.

When she was 29 years old, due to her plans of getting pregnant in the nearest future, she underwent enterolysis. The patient underwent polyectomy 14 times from 2010 to 2020. During the last hospitalization, our patient presented no oral or anal mucocutaneous pigmentation. The laboratory test results showed no abnormal values: haemoglobin level was 12.0 g/dL, red blood cell (RBC) count 4.32×10⁶/µl, mean corpuscular volume (MCV) count 84.1 fL, platelets 284×10³/µl, the international normalized ratio (INR) was 1.1, the sodium level was 139 mmol/L, the potassium level - 4.2 mol/L, the renal function test was normal, the inflammatory markers, such as serum C-reactive protein (CRP) was 2.611 mg/L. Ultrasonography of the abdomen revealed no pathology. Twenty-five polyps were resected endoscopically from the sigmoid colon and rectum (Fig. 1). The histopathology report was suggestive of Peutz-Jeghers polyps. The patient underwent genetic testing, which was positive for a germline mutation (frameshift deletion in exon 1) in the STK11/LKB1 gene, confirming the diagnosis of Peutz-Jeghers syndrome. Genomic DNA was extracted from peripheral blood. She remains under observation and she is scheduled for a follow-up with an upper GI endoscopy in October this year. Additionally, our patient’s sister was recommended to perform a colonoscopy.

DISCUSSION

Peutz-Jeghers syndrome was first reported by Peutz in 1921 and described in 1949 by Jeghers.[4] The etiology of Peutz-Jeghers syndrome remains unknown. The clinical symptoms are non-specific such as anemia, nausea, abdominal pain, and intestinal intussusception. Typical melanin spots on lips, buccal mucosa, and digits might be observed in PJS patients. According to the World Health Organization (WHO), diagnosis of PJS requires some specific criteria to be considered: three or more histologically confirmed Peutz-Jeghers polyps or any number of Peutz-Jeghers polyps with family history of Peutz-Jeghers syndrome or characteristic mucocutaneous pigmentation with a family history...
of Peutz-Jeghers syndrome or any number of Peutz-Jeghers polyps and characteristic mucocutaneous pigmentation.[5]

The diagnosis of PJS is characteristic for childhood or early adulthood. The male-to-female ratio for PJS is almost 1:1. Pigmentations are typically observed in childhood while they may even disappear in adulthood.[6] Our patient has not presented any pigmentation since she remains under our observation. Therefore, this case falls under the first category of WHO criteria for PJS diagnosis. Polyps in the outcome of Peutz-Jeghers syndrome can occur anywhere in the gastrointestinal tract, as well as in the nostrils, urinary bladder, or lungs.[7] Typically, the Peutz-Jeghers polyps measure from 1 to 50 mm. Other than intussusception, complications that can be observed in patients with PJS are hemorrhage, occlusion, gastrointestinal necrosis, perforation, and more in PJS individuals.[1] The majority of individuals with Peutz-Jeghers syndrome have been found to have germline mutation of the serine/threonine kinase-11 (LKB1/STK11) gene, located on chromosome 19p13.3.[2]

Even though LKB1 mutation is claimed to be a tumor suppressor gene, nowadays, it is also considered as a tumorigenesis promoting gene. LKB1 is responsible for inducing pathways that reduce oxidative stress. Since LKB1 gene signaling is believed to be necessary to reduce cancer cells invasion, therefore, any mutation in this gene is possible to cause proliferation of cancer cells.[8] The possible risk for developing any cancer in the course of Peutz-Jeghers syndrome at the age of 30 is up to 5%, at the age of 40 the risk is almost 21%, and in the sixties it is almost 60%.[2]

What is peculiar about our patient is that one of the histopathology reports (in 2009) detected one juvenile polyp. However, a juvenile polyposis syndrome diagnosis requires the presence of either juvenile polyps or family history of JPS.[9] Furthermore, JPS can be diagnosed only in the presence of either juvenile polyps or family history.

Our patient underwent two surgical procedures in the course of the disease as we mentioned above. Furthermore, in 2009, the histopathology report confirmed the coexistence of Peutz-Jeghers polyps and a juvenile polyp. Since then, there was no histopathology report that would show the presence of juvenile polyps. However, detecting one juvenile polyp does not have any influence on the screening pathway of our patient. Furthermore, it is unique because to our knowledge, it is very rare to recognize both types of polyps in one patient. Our patient remains under observation and she is expected for the next hospitalization in the appointed term.

REFERENCES

1. Perrod G, Samaha E, Perez-Cuadrado-Robles E, et al. Small bowel polyp resection using device-assisted enteroscopy in Peutz-Jeghers Syndrome: Results of a specialised tertiary care centre. United Eur Gastroenterol J 2020; 8(2):204–10.

2. Patel R, Hyer W. Practical management of polyposis syndromes. Frontline Gastroenterol 2019; 10(4):379–387.

3. Goto A, Nishikawa J, Nagao M, et al. Solitary Peutz-Jeghers-type polyp of the stomach. Intern Med 2020;59(16):2083–4.

4. Cai HJ, Wang H, Cao N, et al. Peutz-Jeghers syndrome with mesenteric fibromatosis: A case report and review of literature. World J Clin Cases 2020; 8(3):577.

5. Nasri S, Kellil T, Chauquech MA, et al. Intestinal intussusception in Peutz Jeghers syndrome: A case report. Ann Med Surg 2020; 54:106–8.

6. Hammouda SB, Njima M, Abdeljelil NB, et al. An unusual presentation revealing Peutz-Jeghers syndrome in adult. Ann Med Surg (Lond) 2020; 58:87–90.

7. Tomas C, Soyer P, Dohan A, et al. Update on imaging of Peutz-Jeghers syndrome. World J Gastroenterol 2014; 20(31):10864.

8. Li TT, Zhu HB. LKB1 and cancer: The dual role of metabolic regulation. Biomed Pharmacother 2020; 123:110872.

9. Kozacek K, Santos RL, Abdo M, et al. Cancer within the family tree: risks, diagnosis and treatment of juvenile polyposis syndrome. BMJ Case Rep 2020; 13(8):e236845.

10. Gao XH, Li J, Zhao ZY, et al. Juvenile polyposis syndrome might be misdiagnosed as familial adenomatous polyposis: a case report and literature review. BMC Gastroenterol 2020; 20:1–9.

11. Chen YW, Tu JF, Shen WJ, et al. Diagnosis and management of a solitary colorectal juvenile polyp in an adult during follow-up for ulcerative colitis: A case report. World J Gastroenterol 2020; 26(8):877–82.

12. Blatter R, Tschupp B, Aretz S, et al. Disease expression in juvenile polyposis syndrome: a retrospective survey on a cohort of 221 European patients and comparison with a literature-derived cohort of 473 SMAD4/BMPR1A pathogenic variant carriers. Genet Med 2020; 22(9):1524–32.

13. Miyahara Y, Ishida H, Kawabe K, et al. A novel germline BMPR1A variant (c. 72_73delGA) in a Japanese family with hereditary mixed polyposis syndrome. Jpn J Clin Oncol 2020; 50(7):826–9.
Редкий случай ювенильного полипа у больного с синдромом Пейтца-Егерса, осложнившегося инвагинацией тонкой кишки

Анна Рицик1, Беата Каштелан-Шчербинска1, Халина Чихоз-Лах1

1 Кафедра и клиника гастроэнтерологии с отделением эндоскопии, Медицинский университет Люблина, Люблин, Польша

Адрес для корреспонденции: Анна Рицик, Кафедра и клиника гастроэнтерологии с отделением эндоскопии, Медицинский университет Люблина, Люблин, Польша; Email: aniarycyk@op.pl

Дата получения: 6 апреля 2021 ♦ Дата приемки: 2 июня 2021 ♦ Дата публикации: 31 августа 2022

Образец цитирования: Rycyk A, Kasztelan-Szcerbinska B, Cichoz-Lach H. A rare case of a juvenile polyp of patient with Peutz-Jeghers syndrome, complicated with intussusception of the small intestine. Folia Med (Plovdiv) 2022;64(4):693-696. doi: 10.3897/ folmed.64.e67044.

Резюме

Мы сообщаем о редком случае синдрома Пейтца-Егерса (СПЕ) у 35-летней женщины. Пациентке был поставлен диагноз СПЕ, когда ей было 11 лет. С тех пор она находится под наблюдением. Мы твёрдо убеждены, что СПЕ— очень редкий диагноз. Однако это может привести к серьёзнм осложнениям, таким как инвагинация, которую мы наблюдали у нашего пациента. Её состояние (периодические боли в животе и рвота) в детском возрасте требовало дополнительных диагностических мероприятий. Хотя был поставлен диагноз СПЕ, среди многих резецированных полипов один из них оказался ювенильным полипом. Диагноз был подтверждён гистопатологическим отчётом, который был невероятно уникальным. Генетическое тестирование выявило мутацию гена LKB1/STK11. Клиницисты должны быть осведомлены о злокачественном потенциале при СПЕ. Следовательно, эти пациенты требуют индивидуального лечения, длительного наблюдения и особого внимания.

Ключевые слова
рак, полип толстой кишки, полипы желудочно-кишечного тракта, полип гамартомы, синдром Пейтца-Егерса