Acta Dermatovenerol APA

Acta Dermatovenerologica
Alpina, Pannonica et Adriatica

2019;28:135-137
doi: 10.15570/actaapa.2019.33

Peutz–Jeghers syndrome in dermatology

Zofia Nevozinskaya[1,2], Iryna Korsunskaya[3], Luiza Sakaniya[3], Yury Perlamutrov[4], Vladimir Sobolev[3,5] ✉

Abstract

Peutz–Jeghers syndrome is a rare autosomal dominant disorder. Approximately 1:25,000 to 1:280,000 cases are registered annually. The pathogenesis of the disease is based on the mutation of the STK 11 gene on chromosome 19. Peutz–Jeghers syndrome is characterized by several symptoms: the formation of multiple hamartomatous polyps primarily in the gastrointestinal tract and hyperpigmentation of the mucous membranes and skin. Patients with Peutz–Jeghers syndrome often develop various malignant neoplasms, mainly localized in the pancreas and colon. We describe Peutz–Jeghers syndrome in a girl 4 years 7 months old. Initially, the child was diagnosed with vitiligo due to complaints of depigmentation of the skin of the face and hands. During re-examination after half a year, foci of hyperpigmentation on the lip and mucous membranes of the oral cavity were noted. Esophagogastroduodenoscopy showed the presence of a polypous lump in the stomach. Genetic consultation confirmed the diagnosis of Peutz–Jeghers syndrome. The absence of family history indicates a sporadic case characterized by diseases with an autosomal-dominant mode of inheritance. This clinical case demonstrates the need for gastroenterological and genetic examinations in the presence of lesions on the oral mucosa and the vermillion border of the lips to confirm or exclude Peutz–Jeghers syndrome.

Keywords: Peutz–Jeghers syndrome, pigmentation on the lips, polyposis of the intestine, vitiligo

Received: 1 March 2019 | **Returned for modification:** 19 May 2019 | **Accepted:** 25 July 2019

Introduction

Peutz–Jeghers syndrome is an autosomal dominant disease characterized by polyposis of the gastrointestinal tract, pigmentation of the skin and mucous membranes, and a predisposition to oncological diseases (1).

Peutz–Jeghers syndrome is characterized by the presence of multiple hamartomatous polyps throughout the gastrointestinal tract, which can cause abdominal pain, chronic bleeding, anemia, and bowel obstruction. Polyps in the gastrointestinal tract may develop at any age (2). In addition, hamartomatous polyps may have an extraintestinal localization, including in the renal pelvis, bladder, bronchi, nasal passages, or gall bladder. Hyperpigmentation on the skin and mucous membranes usually develops in early childhood. The foci are oval or round, ranging from 2 to 5 mm in size. The color of the spots varies from dark brown to black. The spots are localized on the skin (on the face around the eyes, mouth, fingers, and feet, and around orifices) and on the mucous membranes of the cheeks. With the passage of time, the rash on the lips may be resolved, but on the oral mucosa and fingers of the palms it remains for the rest of the patient's life. This is a very important diagnostic sign that persists even in the absence of polyps in the digestive tract.

This group of patients has a predisposition to develop malignant tumors with various locations: in the pancreas (3) and large intestine (4), and for women in the ovaries (5), cervix (6), and mammary glands (7).

Case report

The patient, a girl 4 years 7 months old, has been under our supervision since May 2016. She was admitted with complaints about the appearance of depigmentation sites on the skin of the face and hands that existed since the winter of 2016. The mother was unable to identify any specific cause of the rash on the child's skin.

The past history of disease includes frequent tonsillitis and acute respiratory viral infection. The girl's father suffers from psoriasis vulgaris, and the mother has had atopic dermatitis since childhood. Allergic anamnesis in the child is negative. The child is from her mother's first pregnancy, which involved no pathology. Her birth was physiological on the 40th week of pregnancy, without complications.

At the presentation, the girl was well. The weight of the child was 27.0 kg, and the child's height was 119.0 cm. No skeletal anomalies were noted. The peripheral lymph nodes were not enlarged. The nail plates and hair were unchanged, whereas the visible mucous membranes were free from rash. She had vesicular breathing without wheezing and the abdomen was soft and painless on palpation. At the time of admission, multiple depigmented foci on the face, trunk, upper, and lower extremities were noted without subjective complaints. The child was diagnosed with vitiligo (Fig. 1). The child was prescribed photoprotective topical medications, and consultation with related specialists was recommended. Complete blood count, urinalysis, and a basic metabolic profile were within normal ranges. In December 2016, during the second visit, the mother noted the presence of a rash on the vermillion border of the lips and the mucous membrane of the mouth (Figs. 2–3). She could not relate this rash to any cause. Moreover, the girl had no subjective complaints. Objectively, there were several oval and round spots ranging from 1 to 3 mm on the vermillion border of the left side of lower lip. On the mucosa of the oral cavity, multiple irregular spots measuring 3 to 5 mm were present. The color of the foci varied from light brown to dark brown. The tongue was clear and moist. The abdomen was soft upon palpation, and painful in the epigastric region, whereas symptoms of irritation of the

[1]N. I. Pirogov Russian National Research Medical University, Moscow, Russia. [2]Moscow Scientific and Practical Center for Dermatovenerology and Cosmetology, Healthcare Department, Moscow, Russia. [3]Center for Theoretical Problems of Physicochemical Pharmacology, Russian Academy of Sciences, Moscow, Russia. [4]A. I. Yevdokimov Moscow State University of Medicine and Dentistry, Moscow, Russia. [5]I. I. Mechnikov Research Institute for Vaccines and Sera, Russian Academy of Medical Sciences, Moscow, Russia. ✉**Corresponding author: vlsobolew@gmail.com**

peritoneum were absent. There was no hepatosplenomegaly. The presence of a rash on the lip and mucous membranes of the oral cavity led to suspicion of Peutz–Jeghers syndrome. The child was sent to a gastroenterologist for examination, where an abdominal ultrasound was performed which was within normal limits. Esophagogastroduodenoscopy revealed a tubular adenoma in the stomach.

Discussion

Peutz–Jeghers syndrome is a monogenic hereditary disease with an autosomal dominant type of inheritance. This case differs from the cases described by Matini et al. in the absence of a family history of the symptoms and signs of Peutz–Jegers syndrome (8). In connection with the absence of a hereditary history, a new mutation (sporadic case) is likely, which is typical for diseases with an autosomal dominant type of inheritance. In this case, the diagnosis of Peutz–Jeghers syndrome and benign neoplasm of the stomach was confirmed based on complaints, objective data, and laboratory and instrumental research methods.

This case is also interesting due to a rare combination of vitiligo and Peutz–Jeghers syndrome, which, to the best of our knowledge, was not previously reported.

In most cases, patients with hamartomatous polyps of the gastrointestinal tract present with complaints of abdominal pain, changes in bowel movement pattern, the presence of blood in stool, or symptoms of anemia. In our case, subjective complaints were absent. A similar case was described by Mozafarri et al., when a 57-year-old patient had primary complaints of pigmentation of the oral cavity for more than 15 years but did not experience altered functioning of the gastrointestinal system despite the presence of multiple hamartomatous polyps in the gastrointestinal tract (9).

Thus, in view of the rare occurrence of Peutz–Jeghers syndrome in the presence of a rash on the oral mucosa and vermillion border of the lips, it is necessary to perform a gastroenterological and genetic examination to confirm this diagnosis.

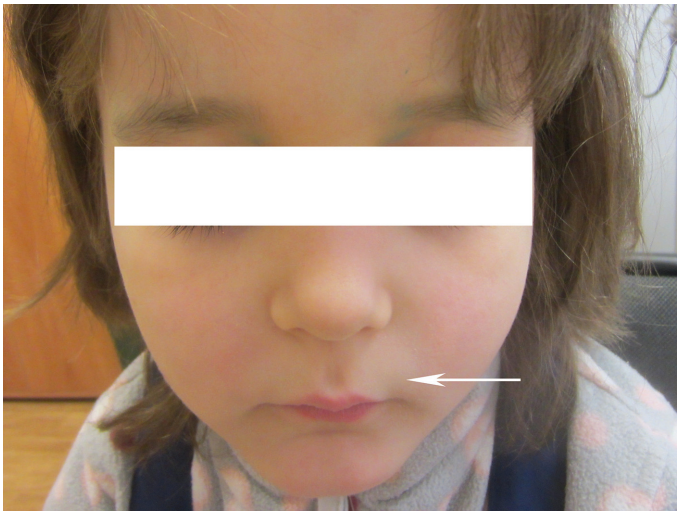

Figure 1 | Vitiligo on the face.

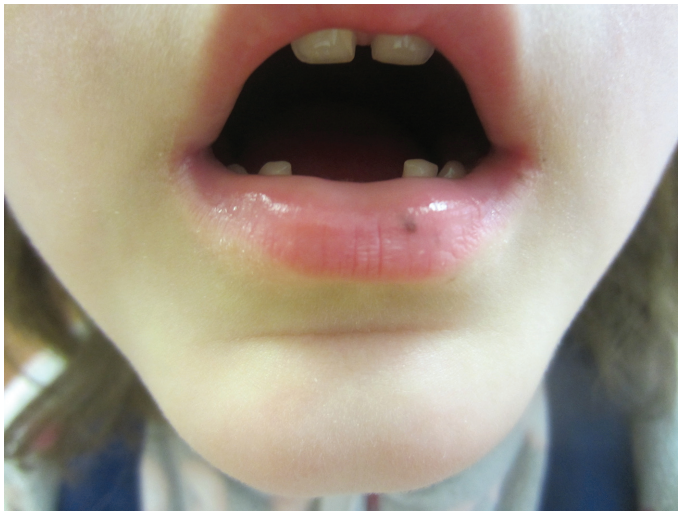

Figure 2 | Brown spot on the lower lip.

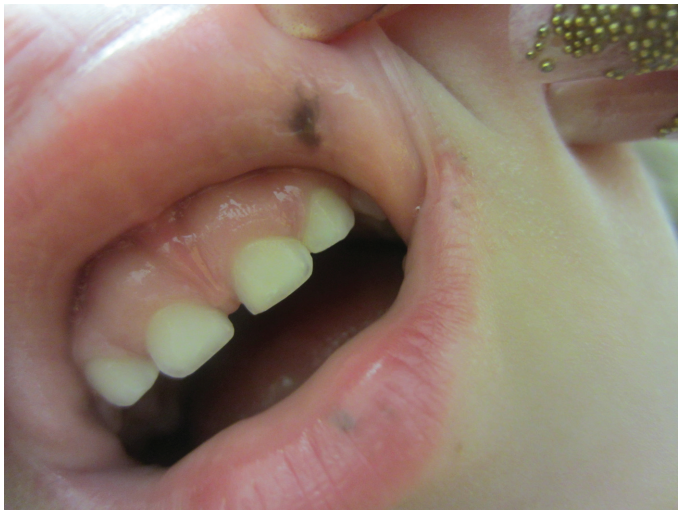

Figure 3 | Brown spot on the oral cavity mucosa.

References

1. McGarrity TJ, Amos CI, Baker MJ. Peutz–Jeghers syndrome [Internet]. Seattle (WA): GeneReviews®, University of Washington, Seattle; 1993–2019. 2001 Feb 23 [Updated 2016 Jul 14; cited 2018 Jan 21]. Available from: https://www.ncbi.nlm.nih.gov/books/NBK1266/.

2. Huang ZH, Song Z, Zhang P, Wu J, Huang Y. Clinical features, endoscopic polypectomy and STK11 gene mutation in a nine-month-old Peutz–Jeghers syndrome Chinese infant. World J Gastroenterol. 2016;22:3261–7.

3. Sohal DP, Willingham FF, Falconi M, Raphael KL, Crippa S. Pancreatic adenocarcinoma: improving prevention and survivorship. Am Soc Clin Oncol Educ Book. 2017;37:301–10.
4. Linhart H, Bormann F, Hutter B, Brors B, Lyko F. Genetic and epigenetic profiling of a solitary Peutz–Jeghers colon polyp. Cold Spring Harb Mol Case Stud. 2017; 3:a001610.
5. Bellfield EJ, Alemzadeh R. Recurrent ovarian Sertoli–Leydig cell tumor in a child with Peutz–Jeghers syndrome. Oxf Med Case Reports. 2016;8:186–9.
6. Ogura T, Ishiguro T, Muta Y, Fukuchi M, Nagai T, Kumagai Y, et. al. A family with Peutz–Jeghers syndrome having various malignant diseases. Gan To Kagaku Ryoho. 2016;43:2133–5.
7. Parkes A, Arun BK, Litton JK. Systemic treatment strategies for patients with hereditary breast cancer syndromes. Oncologist. 2017;22:655–6.
8. Matini E, Houshangi H, Jangholi E, Farjad Azad P, Najibpour R, Farshad A. Peutz–Jeghers syndrome with diffuse gastrointestinal polyposis: three cases in a family with different manifestations and no evidence of malignancy during 14 years follow up. Iran Red Crescent Med J. 2015;17:e19271.
9. Mozaffari HR, Rezaei F, Sharifi R, Mirbahari SG. Seven-year follow-up of Peutz–Jeghers syndrome. Case Rep Dent. 2016;2016:1–3.