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Case Report

Peutz-Jeghers syndrome with disappeared pigmentation on lips after operation: A case report

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Peutz-Jeghers polyps are hamartomas of the small bowel which occur as part of the Peutz-Jeghers syndrome, a rare autosomal dominant condition characterized by gastrointestinal polyps and mucocutaneous melanotic pigmentation. We, hereby, report a case of a young female suffering from this syndrome who presented with hyperpigmentation of the lips and acute intestinal obstruction due to intussusception. An exploratory laparotomy was performed which revealed jejuno-jejunal and jejuno-ileal intussusceptions for which resection and anastomosis was done. There were multiple intraluminal polyps in the jejunum which upon histopathological examination, showed features of Peutz-Jeghers polyp. But, on postoperative day 11, we found the pigmentation on her lips disappeared, the patient behaved well in the postoperative period.

Keywords: Hamartoma, intussusception, intestinal obstruction, lips, polyps, Peutz-Jeghers syndrome.

Peutz-Jeghers syndrome (PJS) is an autosomal dominant condition characterized by the combination of hamartomatous polyps of the gastrointestinal tract and hyperpigmentation of the buccal mucosa, lips and digits.

The polyps may cause bleeding or intestinal obstruction (from intussusception). While the hamartomatous polyps themselves only have a small malignant potential, patients with this syndrome are at greater risk of developing carcinomas of the pancreas, liver, lungs, breast, ovaries, uterus, testicles and other organs. [(1)] Herein, we report a case of multiple Peutz-Jeghers polyps leading to intussusception and presenting with small bowel obstruction as an emergency, but disappeared pigmentation on the lips after operation. Such a presentation is rare in the Democratic People's Republic of Korea (DPRK) and due to scarcity of literature on Peutz-Jeghers polyps/ syndrome in DPRK we felt reporting this case may be a noteworthy contribution.

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Case Report

In December of last year, a 7-year-old girl presented to the emergency department with complaints of colicky abdominal pain and bilious vomiting along with history of abdominal distention and non-passage of flatus and stools for two days. There was a past history of intussusception for similar complaints at the age of 6 years.

The parents were well aware that the child had intussusception which was non-surgically corrected. Since then the child had been asymptomatic, but there were pigmented lesions on the lips and tongue. The abdomen was distended with tenderness in the lower abdomen. Ultrasonography of the abdomen showed the "target sign''of intussusceptions. The patient underwent an emergency exploratory laparotomy after resuscitation with intravenous fluids and antibiotics. Intraoperative findings revealed two small bowel intussusceptions (jejuno-jejunal and jejuno-ileal) with gangrene of the gut loops in the jejuno-ileal intussusception. We reduced the jejuno-jejunal intussusception and resected the gangrenous distal segment with end to end jejuno-ileal anastamosis. On careful examination of the proximal bowel, there were multiple polyps present intraluminally in a segment of the jejunum which was also resected and after enterotomy of this segment, multiple polyps were

noticed. The patient recovered well with disappeared pigmentation on lips on postoperative day 2 but not on the tounge and was discharged on postoperative day 11.

Discussion

Peutz-Jeghers syndrome (PJS) is rare a autosomal dominant disease characterized by hamartomatous polyps in the gastrointestinal tract and mucocutaneous pigmentation. It was first reported by Peutz in 1921 and the definitive clinical description of the syndrome was written by Jeghers in 1949. The prevalence of PJS varies from one study to another, its estimated incidence has been noted from 1 in 8,300 to 1 in 120,000 births, and both sexes are almost equally affected. [2] The cause of PJS in most cases (66 - 94%) appears to be a germline mutation of the *STK11/LKB1* (Serine/Threonine Kinase 11) tumor suppressor gene, located on band 19p13.33. [1] The clinical picture of the syndrome includes two major components: mucocutaneous pigmentation and hamartomatous polyposis of the gastrointestinal tract. Fewer than 5% of patients with PJS lack the abnormal mucocutaneous pigmentation, and fewer than 5% of patients with the pigmentation have no Peutz-Jeghers polyps. [3]

Mucocutaneous pigmentation caused by melanin aggregation is the characteristic finding of PJS and is presented in 93% of PJS patients, but not all. Pigmented lesions are commonly seen on the lips, perioral region, conjunctiva, nostrils and buccal mucosa; and sparsely on the fingers, soles of the feet, palms, anal area and intestinal mucosa.[4] The histological features of PJS polyps are well documented. These polyps are hamartomatous and have a specific configuration of smooth muscle cell branches covered by either normal or hyperplastic mucosa native to the involved site.[5] The diagnostic criteria for PJS proposed by the Johns Hopkins Registry include histopathologically verified hamartomatous polyps with at least two of the followings: small-bowel location for polyposis, mucocutaneous melanotic pigmentation, and a family history of Peutz-Jeghers syndrome. [6] Our patient had histopathologically verified hamartomatous polyps with two of the additional criteria, i.e., small bowel polyposis and mucocutaneous pigmentation. As the polyps can develop at any region in the entire gastrointestinal tract, their recurrence is quite common, making it a challenge for the surgeon to decide the amount of resections that should be done.[7, 8] There are no reports in the literature regarding the disappearance of pigmentation on the lips post-resection. The patient described above has been lived in geologically special region of the county far away from the capital. Perhaps there seems to be a relation between the germline mutation of the *STK11/LKB1* tumor suppressor gene and a personal life environment and operation.

Conclusion

Even though fewer than 5% of patients with PJS lack the abnormal mucocutaneous pigmentation, disappeared pigmentation on the lips cannot be found. We would like to emphasize that in cases of Peutz-Jeghers syndrome, further research is needed to determine the relationship between disappeared pigmentation on the lips and personal conditions including environment and resection of gastrointestinal tumors.

Conflict of interest

None of the authors has any potential conflict of interest to disclose.

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