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Peutz-Jeghers Syndrome, Importance of Appropriate Diagnosis and Follow-Up: A Case Report

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Introduction

Peutz-Jeghers syndrome (PJS) is an autosomal dominant inheritance disorder with a tendency to predominantly hamartomatous polyposis and cancer in the gastrointestinal tract, in the vermilion margin of the lips, buccal mucosa, around the mouth and nose, perianal area and hands and feet. Hyperpigmentation of the lips, which is one of the striking findings of this syndrome, can occur at any stage of life starting from infancy and tends to disappear after puberty. Due to polyps developing in the gastrointestinal tract, initial complaints are usually abdominal pain, invagination, and treatment-resistant iron deficiency anemia. Although the age of onset of these complaints is from childhood, the mean age at diagnosis is reported as 20s.

METHODOLOGY

When a 10-year-old male patient was referred by a dermatology doctor with complaints of bruising on his lips, it was learned that the patient had a long history of abdominal pain. It was reported that the patient had received iron deficiency treatment repeatedly but the anemia complaint still persisted. He had a history of rectal prolapse. Physical examination revealed height: 135 cm (25-50 p) weight: 40 kg (75-90 p), hyperepigmentation of the lips and hyperepigmentation of the buccal mucosa. System examination was normal. Physical examination revealed that length was: 135 cm (25-50 p) weight: 40 kg (75-90 p). Hyperepigmentation of the lips and buccal mucosa were observed, and systemic examination was normal. Laboratory tests Hemoglobin: 10.8 g / dL, White cell: 9700 / mm³, Platelet: 493,000 / μ L, MCV: 75 fl, RDW: 22%, Ferritin: 7.8 ng / mL (normal range: 30-400 ng / mL), Iron: 13 μ / dL (normal range: 33-193 μ / dl). Anisocytosis, polychromasia, and poikilocytosis were detected in peripheral smear. Stool hHb: negative Pt: 11.8 sec INR: 1.01 was found to be normal. In the upper endoscopy of the patient, several polyps narrowing to 1 cm, one 2 cm pylor narrowing in the stomach antrum and two polyps less than 1 cm in the duodenum were observed and polypectomy was performed to the small polyps, but the large polyp in the pyloric mouth was not removed endoscopically. Colonoscopy showed polyposis in terminal ileum and 3 polyps less than 1 cm in sigmoid colon. Polypectomy was performed on polyps in sigmoid colon. The polyps in the antrum and veduodenum were found to be compatible with hyperplastic polyps, whereas polyps in the sigmoid colon were compatible with hamartomatous polyps. Polyp was not detected in our patient's ear, nose and throat examination. genetic examination was sent. Family members were directed to screening for possible types of cancer.

CONCLUSION

In patients with hyperpigmented macular rash, most commonly located in the buccal mucosa and lips, PJS comes to mind with careful physical examination and family interrogation before the development of complaints such as treatment-resistant iron deficiency anemia, recurrent abdominal pain, and developmental retardation that we frequently encounter in childhood, these patients and other family members should be screened with further investigations and followed up for complications.

References

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