Rectal Carcinoma in a Young Female Patient with Peutz-Jeghers Syndrome: A Case Report

Hsiang-Lin Tsai\textsuperscript{a, c}, Chih-Hung Lin\textsuperscript{b}, Ya-Lin Cheng\textsuperscript{a}, Ching-Wen Huang\textsuperscript{a}, Jaw-Yuan Wang\textsuperscript{a, c, d}

Departments of \textsuperscript{a}Surgery and \textsuperscript{b}Pathology, and \textsuperscript{c}Cancer Center, Kaohsiung Medical University Hospital, and \textsuperscript{d}Institute of Clinical Medicine, Kaohsiung Medical University, Kaohsiung, Taiwan, ROC

Key Words
Peutz-Jeghers syndrome · Rectal carcinoma · Cancer risk · Hamartomatous polyp · Intussusception

Abstract
Objective: To report a case of rectal cancer in a patient with Peutz-Jeghers syndrome (PJS).

Clinical Presentation and Intervention: A 20-year-old woman with intermittent bloody stool of 4 months was admitted for examination. Gastroendoscopy revealed multiple polyps involving the stomach, small intestine, colon and a rectal adenocarcinoma. A diagnosis of PJS was made based on intestinal polyps with characteristic pathology and melanotic macules on the lips. After surgery and chemotherapy upon follow-up at 8 months, the patient did not have any signs of recurrence. Conclusion: This case showed that rectal carcinoma should be considered for young patients with PJS.

Case Report

A 20-year-old female patient presented with a 4-month history of intermittent bloody stool, poor appetite, abdominal pain and body weight loss of about 6 kg within 4 months. Physical examination revealed multiple café-au-lait pigmented macules on the lips and buccal mucosa (fig. 1a). Colonoscopy demonstrated 2 small...
polyps 20 and 40 cm from the anal verge, respectively, but a large mass 6 cm from the anal verge was also found simultaneously (fig. 1b). Gastroendoscopy showed multiple gastric polyps at the fundus. Biopsies of these colorectal tumors were performed and showed the colonic polyps were villous adenomas and the rectal mass was adenocarcinoma. Abdominal computed tomography revealed rectal adenocarcinoma with perirectal invasion and multiple enlarged perirectal lymph nodes (fig. 1c).

Finally, PJS with synchronous rectal cancer was diagnosed. Surgical intervention, i.e. lower anterior resection, of rectal cancer was performed. The size of this lesion was $6 \times 5.7$ cm, and histology showed arborizing muscular bundles characteristic of hamar-
tomatous polyps (fig. 2a), and invasive nests with pleomorphic cancerous glandular cells were detected microscopically (fig. 2b), as were lymph node metastases and vascular invasion (fig. 2c). The grade was 'well differentiated', and 8 of 22 lymph nodes had metastatic adenocarcinoma. The final pathological data revealed T2N2bM0 (UICC stage IIIB). One month after surgery, she suffered from an episode of recurrent jejunojejunal intussusception, and resection of the involved jejunum was performed. Two months after surgery, she was treated with adjuvant chemotherapy (FOLFIRI-4) without complications. At the last follow-up 8 months after diagnosis the patient is well and has not signs of recurrent disease assessed with abdominal computed tomography.

**Discussion**

In this case, the patient had no history of familial PJS and associated manifestations before the age of 20. Due to this, the diagnosis was delayed and development of malignancy occurred. Malignant degeneration of polyps in the rectum, small intestine, duodenum, and stomach at a very young age and subsequent advanced rectal carcinoma is a very rare occasion [4].

Clinical diagnosis is based on pigmentation and polyp pathology. The pigmentation is particularly in macular form, 1–5 mm in diameter, and found mostly in the buccal mucosa, on the lips and around the mouth [4]. Polyps associated with PJS are hamartomatous and have smooth muscle arborizing through the polyps, a feature almost unique to PJS.

PJS is caused by a germline mutation in the serine threonine kinase 11 (STK11) gene located on band 19 [4]. About 75% of patients have an STK11 mutation that can be identified with clinically available testing. Almost all patients with PJS appear to develop signs of the disease (high penetrance). However, there are wide ranges of expression with some patients presenting at a young age with severe symptoms while others only have mild symptoms late in life.

**Conclusion**

This case showed that rectal carcinoma should be considered for young patients with PJS.

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**References**