A 57-year-old man presented with chronic fatigue, weakness, irritability, and lack of concentration. Physical examination revealed pale skin with grooved and brittle nails. In addition, hyperpigmented mucocutaneous lesions were noted in the mouth. Laboratory investigation was remarkable for iron deficiency anemia. Esophagogastroduodenoscopy (EGD) disclosed a large polypoid mass in the gastric cardia (Fig. 1 a). Chromoendoscopy with methylene blue (1% dilution) revealed a Kudo pit pattern 3L (large roundish and tubular pits; Fig. 1 b) which was confirmed by narrow-band imaging (Olympus, Tokyo, Japan; Fig. 1 c). Confocal laser endomicroscopy (EC-3870 CIFK, Pentax, Tokyo, Japan) showed disorganization of glands with suspicion of different cell types but no cellular atypia seen on confocal laser endomicroscopy. (e) No signs of atypia on endocytoscopy. (f) Enlarged mucosa seen on endoscopic ultrasound. (g) Multiple small polyps visualized in the jejunum on oral double-balloon endoscopy. (h) Ileocolonoscopy showing multiple polyps (2–25 mm in size).

Following genetic analysis, which disclosed mutation of the serine threonine kinase 11 (STK11)/LKB1 gene, a diagnosis of Peutz–Jeghers syndrome was made. Peutz–Jeghers syndrome, an autosomal dominant condition, is characterized by hamartomatous polyps in the gastrointestinal tract and mucocutaneous melanin pigmentation, and is caused by a germline mutation of the STK11 gene. Patients are at an increased risk of developing different types of gastrointestinal and non-gastrointestinal tumors. Most authorities recommend polypectomy for gastric/colonic polyps larger than 10 mm, and surgery is recommended for symptomatic or rapidly growing small intestinal polyps and asymptomatic polyps greater than 10–15 mm in diameter [1]. In our patient, all the polyps greater than 5 mm in diameter were removed in multiple sessions from the stomach, jejunum (using double-balloon endoscopy), and colon. Histopathological analysis revealed hamartomatous polyps. Follow-up EGD and colonoscopy were performed after 6 months. In keeping with international guidelines, surveillance endoscopy of the stomach, small bowel, and colon will now be performed every 2 years.

Panendoscopic characterization of Peutz–Jeghers syndrome

Fig. 1 (a) Large polypoid mass in the gastric cardia visualized on esophagogastroduodenoscopy. (b) Large, round, tubular pits demonstrating a Kudo pit pattern 3L on chromoendoscopy (1% methylene blue). (c) The pit pattern was confirmed on narrow-band imaging. (d) Disorganized glandular pattern with possibility of different cell types but no cellular atypia seen on confocal laser endomicroscopy. (e) No signs of atypia on endocytoscopy. (f) Enlarged mucosa seen on endoscopic ultrasound. (g) Multiple small polyps visualized in the jejunum on oral double-balloon endoscopy. (h) Ileocolonoscopy showing multiple polyps (2–25 mm in size).

Competing interests: None

Endoscopy_UCTN_Code_CCL_1AB_2AD_3AB

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Endoscopy 2010; 42: E235
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