Panendoscopic characterization of Peutz–Jeghers syndrome

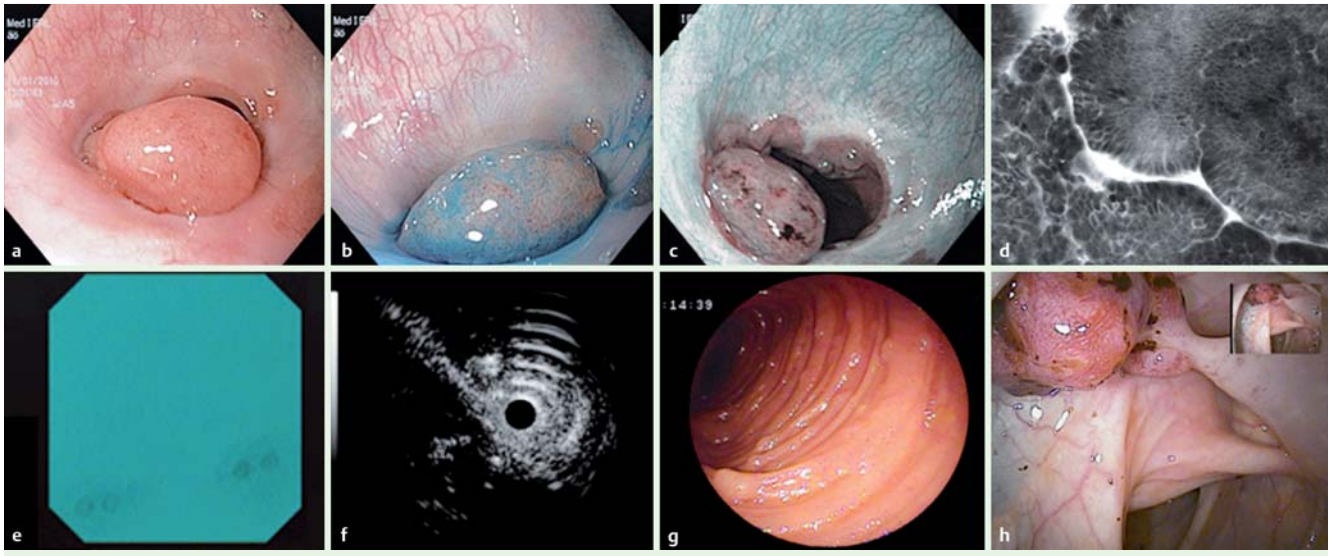

Fig. 1 (**a**) Large polypoid mass in the gastric cardia visualized on esophagogastroduodenoscopy. (**b**) Large, round, tubular pits demonstrating a Kudo pit pattern 3L on chromoendoscopy (1 % methylene blue). (**c**) The pit pattern was confirmed on narrow-band imaging. (**d**) Disorganized glandular 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).

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 3 L (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 there was a lack of atypia (**Fig. 1 d**). Endocytoscopy (XEC-120-U, Olympus, Tokyo, Japan) of the polyp revealed normal cells without nuclear or cytoplasmic changes (**Fig. 1 e**), and endoscopic ultrasound showed enlargement of the mucosa but no muscularis infiltration (**Fig. 1 f**). Subsequently, an oral double-balloon endoscopy (Fujinon, Omiya, Japan) disclosed multiple polyps in the jejunum, most of them smaller than 10 mm in diameter (**Fig. 1 g**). Endoscopic examination was completed with ileocolonoscopy, which revealed multiple polyps ranging in size from 2 mm to 25 mm (**Fig. 1 h**). 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.

Competing interests: None

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Reference

1 *Giardiello FM, Trimbath JD.* Peutz–Jeghers syndrome and management recommendations. Clin Gastroenterol Hepatol 2006; 4: 408 – 415

Bibliography

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