Peutz–Jeghers syndrome (PJS) is a rare, autosomal dominant condition that is characterized by hamartomatous polyps in the gastrointestinal tract and mucocutaneous melanin pigmentation [1, 2]. Confocal laser endomicroscopy (CLE) is rapidly emerging as a valuable tool for gastrointestinal endoscopic imaging, enabling the endoscopist to obtain an “optical biopsy” of the gastrointestinal mucosa during the endoscopic procedure [3–5].

Here, we present the case of a patient with genetically confirmed PJS who was referred for surveillance endoscopy. Esophagogastroduodenoscopy revealed a large, polypoid mass of about 20 mm in diameter located in the gastric fundus (Fig. 1a). Pit pattern analysis using narrow-band imaging (Olympus, Tokyo, Japan) revealed large roundish and tubular pits (Kudo 3 L). After administration of 3 mL intravenous fluorescein 10 % (Alcon Laboratories, Texas, USA), CLE (EG-3870 CIK, Pentax, Tokyo, Japan) was performed. Endoscopic histology revealed large, irregular cells with increased optical density and fluorescein leakage (Fig. 1b). According to these changes the diagnosis of high-grade intraepithelial neoplasia was made. Corresponding histopathological analysis confirmed the presence of low-grade intraepithelial neoplasia and a focal high-grade intraepithelial neoplasia as well (Fig. 1c).

Colonoscopy (Pentax EC-3870 CIFK, Tokyo, Japan) showed seven polyps ranging from 2 mm to 15 mm in diameter (Fig. 2a). CLE of the polyps showed disorganization of glands, occurrence of different cell types, and the presence of muscle fibers. Histopathological examination showed an overall villous architecture and markedly interdigitating smooth muscle bundles of varying thickness, which arose from the muscularis mucosae (Fig. 2c).

This is the first report of CLE in diagnosing polyps in PJS. Our case highlights the utility of CLE for the in vivo analysis of intestinal polyps. Thus, CLE may significantly improve the clinicohistopathologic evaluation and may prevent diagnostic and therapeutic delays.

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References


Bibliography

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