Diagnosis and management of the first reported case of esophageal, gastric, and small-bowel heterotopia in the colon, using confocal laser endomicroscopy

Fig. 1 Endoscopic, confocal imaging, and histology of the colon. **a** Large areas of pale abnormal-looking mucosa adjoining normal looking mucosa at endoscopy. **b** On confocal imaging villi are seen. **c** Histology depicting short villi, presence of Paneth cells, and a well-developed brush border.

Fig. 2 Cdx2 staining. **a** Cdx2-positive (arrow) in the normal colonic mucosa (A) but negative in the adjacent esophageal mucosa (B). **b** Cdx2 also positive in the aberrant small-intestinal mucosa.

A 12-year-old boy was referred to our department with a history of diarrhea persisting since the age of 2 months. The stool frequency was 5–20 times a day without any associated blood or mucus. Extensive investigations failed to reveal a cause. However, at 7 years of age and following endoscopy and further histology, a diagnosis of esophageal, gastric, and small-bowel heterotopia in the colon was made (● Fig. 1a and 1c). Immunostaining for Cdx2 was positive in the normal colonic mucosa (● Fig. 2a) and the heterotopic small-bowel mucosa (● Fig. 2b) and negative in the heterotopic gastric and esophageal mucosa (● Fig. 2a). A repeat ileocolonoscopy was performed using the Pentax EC3870CCLK (Pentax Europe, Hamburg, Germany) confocal endomicroscope to characterize the extent of the heterotopic tissue and perform targeted argon plasma coagulation (APC). Confocal imaging showed the presence of villi, gastric pits, and squamous epithelium adjacent to normal colonic mucosa (● Fig. 1b). A polyp was also detected in the sigmoid colon. Following APC the patient showed dramatic clinical improvement with only two or three normal stools per day.

Heterotopia is the presence of normal tissue at an abnormal site. This is the first reported case of heterotopic gastric, small-intestinal mucosa, and squamous mucosa in the human colon [1,2]. The Cdx2 gene expresses in the colon and regulates intestinal cell differentiation [3]. Cdx2 ± mutant mice develop colonic heterotopia [4]. Our patient had similar features, with the absence of Cdx2 in heterotopic gastric and esophageal tissue suggesting a possible link. Confocal endomicroscopy was useful in targeting biopsies to abnormal mucosa and performing endotherapeutic procedures. All of the three cases reported so far with squamous metaplasia in the colon had long-standing inflammatory bowel disease [5]. In our case the presence of small-bowel and gastric mucosa in addition to squamous mucosa suggested that metaplasia was unlikely. Moreover the child improved considerably following APC and without any active treatment for inflammatory bowel disease.

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