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

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).

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 Cdx2 mutant mice. Nature 1997; 386: 84–87


References

Bibliography
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