Upper Endoscopic Biopsy Findings in Children with Trisomy 21

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Introduction

Trisomy 21 (T21) is associated with increased risk of celiac disease (CD) and reflux esophagitis. Other gastrointestinal (GI) pathologies including gastric heterotopia (GH) have not been previously reported in T21.

Objective

Our aim was to study upper endoscopic biopsies in T21 to assess for associated GI pathology.

Methods

After approval from the Research Ethics Board, T21 patients' records from 2000-2015 were reviewed. We included patients under 18 years of age who had upper endoscopies with biopsies. Upper endoscopic reports and biopsies were reviewed, including the duodenum, duodenal cap, gastric antrum and body, and esophagus.

Results

Study Population

Fifty patients were identified: 27 male, 23 female. Mean age was 7.8 years old (range: 12 months to 17 years).

Histopathologic Findings

Duodenum

	Duodenum (n=49)	Duodenal Cap (n=33)
Normal	27 (55%)	16 (48%)
Celiac disease	17 (35%)	13(39%)
Duodenitis	1 (2%)	2 (6%)
Gastric heterotopia	1 (2%)	3 (9%)

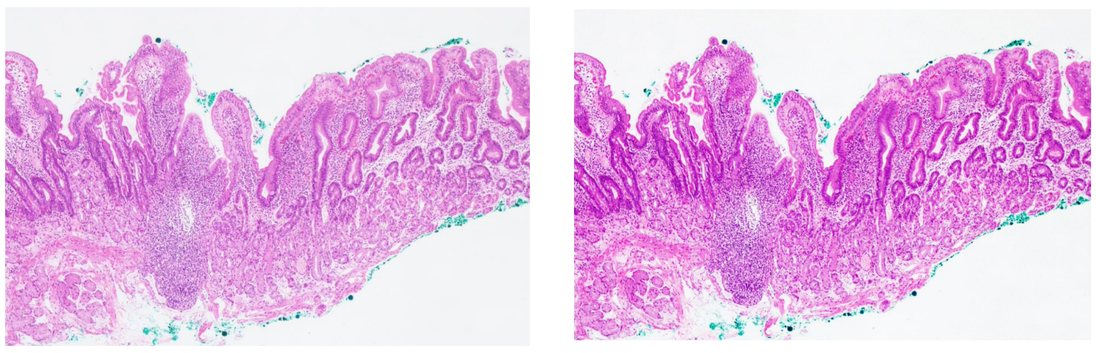

Figure 1. Duodenal mucosa with gastric heterotopia. Note absence of villi and goblet cells in the gastric heterotopia compared to native duodenal mucosa. HE X100

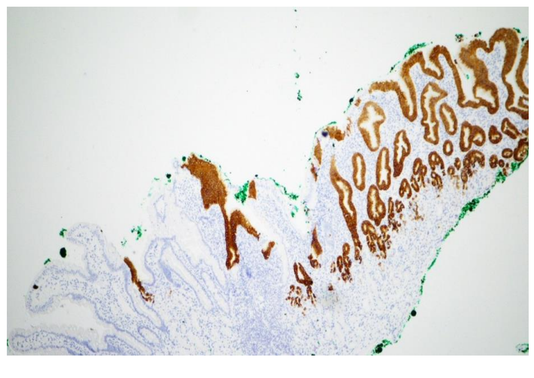

Figure 2. Same biopsy as Fig. 1 showing duodenal mucosa with gastric heterotopia. Note strong expression highlighting gastric heterotopic epithelium compared to absent staining in the native duodenal epithelium. MUC 5 X100

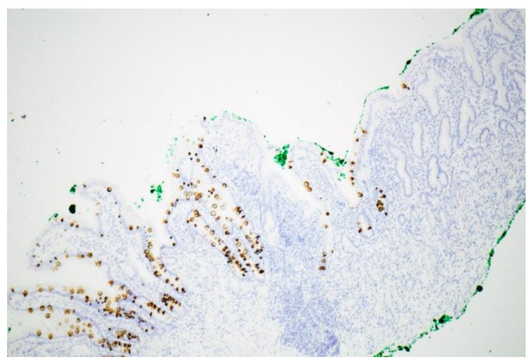

Figure 3. Same duodenal biopsy with MUC 2 expression highlighting goblet cells in the native duodenal mucosa. Note absent staining in gastric heterotopia. MUC 2 X100

Stomach

	Gastric Antrum (n=35)	Gastric Body (n=33)
Normal	20 (57%)	15 (45%)
Lymphocytic gastritis	2 (6%)	2 (6%)
Chronic gastritis	7 (20%)	9 (27%)
PPI use	0 (0%)	10 (30%)
H. Pylori	n/a	1 (3%)

PPI: proton-pump inhibitor

Esophagus

	Esophagus (n=34)
Normal	24 (71%)
Reflux esophagitis	10 (29%)

Summary of Abnormal Findings

	Duodenum (n=49)	Stomach (n=39)	Esophagus (n=34)
Normal	14 (49%)	15 (38%)	24 (71%)
Abnormal	25 (51%)	24 (62%)	10 (29%)

It was noted that two out of four cases who demonstrated gastric heterotopia also showed histological changes due to proton-pump inhibitor (PPI) use.

Conclusion

Our results are in keeping with the literature, with CD and reflux esophagitis being prevalent in T21.
In addition, our study is the first to describe GH as a potential additional GI finding associated with T21.
The etiology of GH identified in our T21 patients may be related to PPI use or be congenital.