Editor’s quiz: GI snapshot

Aggressive case of early onset gastric cancer; identifying the knowledge gap

INTRODUCTION
A woman aged 30 years was referred for endoscopy for iron deficiency anaemia. Colonoscopy was normal. Upper endoscopy demonstrated multiple small, carpet-like, sessile polyps in the gastric fundus and a larger 4 cm polyp in the body (Figure 1). The duodenum was normal without polyps. Endoscopic ultrasound demonstrated a 35×36 mm hypoechoic polypoid lesion arising from the mucosa and invading into the submucosa, but with well-defined borders suggesting lack of invasion into adjacent structures. Endoscopic mucosal resection was performed on a portion of this polyp for histology. Pathology showed a focus of high-grade dysplasia characterised by loss of nuclear polarity, micropapillary architecture and large, rounded nuclei (H&E stain, 200× magnification).

QUESTION
What caused this young woman’s aggressive gastric cancer?

See page 852 for answer
Editor’s quiz: GI snapshot

Aggressive case of early onset gastric cancer; identifying the knowledge gap

See page 828 for question

ANSWER

Genetic testing revealed a mutation in the 1B promoter region of the APC gene (c.191T>C) consistent with gastric adenocarcinoma and proximal polyposis of the stomach (GAPPS). Hyperplastic and fundic gland polyps commonly occur in the stomach and are usually sporadic and non-neoplastic. However, GAPPS is an extremely rare syndrome with an autosomal dominant inheritance pattern characterised by extensive polyps carpeting the proximal stomach (fundus and body), often clustering with mass-like features, with antral/lesser curvature sparing and no duodenal/colon polyps or extraintestinal manifestations.¹ These features distinguish GAPPS from sporadic benign polyps, familial adenomatous polyposis, MUTYH-associated polyposis, Peutz-Jeghers syndrome, juvenile polyposis syndrome and Cowden’s syndrome. GAPPS was first reported in 2012² and has since been identified in <20 families worldwide.³ ⁴ There are no published recommendations on screening for family members, endoscopic surveillance, chemoprevention or timing of prophylactic gastrectomy. There are reported cases of individuals with polyps alone or with early cancer who later present with widespread metastases, similar to our patient, that support prophylactic gastrectomy.⁵ We share this case to draw attention to GAPPS and call for prospective registries to follow affected families and fill the knowledge gap with clinical guidelines.

Jennifer M Kolb 1, Stephen Leong, 2 Lindsey M Westbrook, 3 Swati G Patel 1

¹Division of Gastroenterology & Hepatology, University of Colorado Anschutz Medical Campus, Aurora, Colorado, USA
²Division of Medical Oncology, University of Colorado Anschutz Medical Campus, Aurora, Colorado, USA
³Department of Pathology, University of Colorado Anschutz Medical Campus, Aurora, Colorado, USA

Correspondence to Dr Jennifer M Kolb, Division of Gastroenterology & Hepatology, University of Colorado - Anschutz Medical Campus, Aurora, CO 80045, USA; jennifer.m.kolb@cuanschutz.edu

Correction notice This article has been corrected since it published Online First. A typographical error has been corrected in the legend of figure 2.

Twitter Jennifer M Kolb @jenkolbmd

Acknowledgements We thank Ms Allison Richards, Ms Amanda Gould and their family.

Contributors Drafting the manuscript: JK, critical revision: SL and SP.

Funding The authors have not declared a specific grant for this research from any funding agency in the public, commercial or not-for-profit sectors.

Competing interests None declared.

Patient consent for publication Obtained.

Provenance and peer review Not commissioned; externally peer reviewed.

© Author(s) (or their employer(s)) 2021. No commercial re-use. See rights and permissions. Published by BMJ.


Received 11 December 2019
Revised 10 January 2020
Accepted 13 January 2020
Published Online First 11 March 2020

Gut 2021;70:852. doi:10.1136/gutjnl-2019-320453

ORCID iD Jennifer M Kolb http://orcid.org/0000-0002-6352-0797

REFERENCES


