Extracolonic Manifestation of Familial Adenomatous Polyposis in an Asymptomatic Patient; Literature Review On Current Recommendation

Asemptomatik Bir Hastada Ailesel Adenomatöz Polipozun Ekkolonik Manifestasyonu; Güncel Tavsiye Üzerine Literatür Taraması

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ABSTRACT

Extracolonic manifestations of Familial Adenomatous Polyposis often associated with Gardner syndrome. We report a case of patient post panproctocolectomy with gastric and duodenal polyp found during upper GI scope. A rare condition which cause management dilemma as the patient asymptomatic. We review few literature regarding the latest recommendation on the management of extracolonic manifestation of FAP particularly gastric and duodenal polyposis in patient with FAP. Most expert agreed that duodenal polyposis should be manage based on Spigelman staging. For Stage 1 and Stage 2, recommended for surveillance. For Stage 3, require medical or endoscopic treatment while Stage 4, surgical option should be considered in patient age more than 35 years old.

Key Words: Extracolonic FAP management , Gardner syndrome

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ÖZET

Ailesel Adenomatöz Polipozun ekstrakolonik belirtileri Gardner sendromu ile ilişkilidir. Üst GI skopisi sırasında gastrik ve duodenal polip saptanan panproktokolektomi sonrası bir olguyu sunuyoruz. Asemptomatik hasta olarak yönetim ikilemine neden olan nadir bir durumdur. FAP'lı ve özellikle gastrik ve duodenal polipozlu hastada FAP'ın ekstrakolonik yönetimi hakkındaki en son önerilerle ilgili literatür gözden geçirilmiştir. Çoğu uzman, duodenal polipozun Spigelman evrelemesine dayanarak yönetilmesi gerektiği konusunda hemfikirdir. Evre 1 ve Evre 2 için, gözetim için önerilir. Evre 3 için, tıbbi veya endoskopik tedavi gerektirirken Evre 4, 35 yaşın üzerindeki hasta yaşlarında cerrahi seçenek düşünülmelidir.

Anahtar Sözcükler: Ekstrakolonik FAP yaklaşımı, Gardner sendromu

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INTRODUCTION

Familial adenomatous polyposis (FAP) is an autosomal dominant inherited cancer-predisposition syndrome that is causally linked to the adenomatous polyposis coli (APC) gene located on chromosome 5q21 (1). Characterize by diffuse intestinal polyposis. The disease penetrance is up to 100% at the age of 40 years old (2). Thus required prophylactic colectomy which has been a standard practice all around the world. However surgical intervention for those who has polyposis elsewhere still controversial in term of extend of surgery, as further resection may end up with morbidity. We present a case of FAP with extracolonic manifestation (Gastric and Duodenal polyposis) which has been noted during follow up scope post prophylactic pan-proctocolectomy. We review few literature regarding the latest recommendation on the management of extracolonic manifestation of FAP particularly gastric and duodenal polyposis in patient with FAP.

CASE REPORT

Mr RI, a 49-years-old gentleman with strong family of FAP who presented with complained of altered bowel habit for almost 2 years. Further work up showed multiple polyps from caecum to rectum with invasive sigmoid cancer. Panproctocolectomy with ileoanal anastomosis was successfully performed and patient subsequently undergone chemotherapy.
Post-operative follow up OGDS revealed multiple polyps within stomach and duodenum (Figure 1, Figure 2) which were biopsied and showed adenomatous polyps. However patient was not keen for any subsequent surgical intervention.

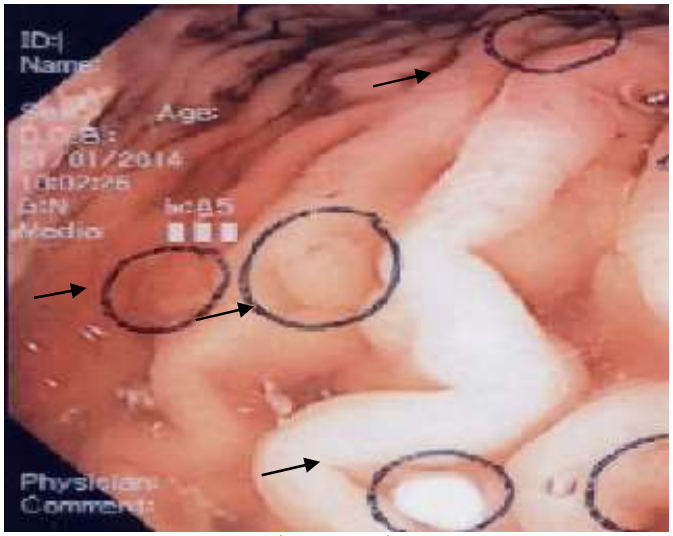

Figure 1: Multiple gastric polyp (black arrow)

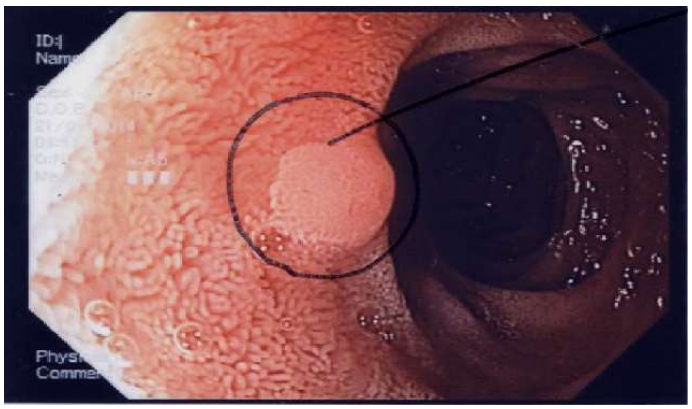

Figure 2: Small polyp on the duodenum.

DISCUSSION

Upper GI polyps (Gastric and duodenal adenoma) are present nearly 90% of FAP patient by the age of 70 years old with 12% discovered during upper scope (3). 2/3 of duodenal polyps occur in papilla or periampullary region, while incidence of gastric polyps estimated around 26%-61% in patient with FAP .(3) Incidence of FAP-associated gastric cancer in a large Korean series is 4.2% while from a Japanese series is 2.1% which was much higher than the incidence reported from Western countries, 0.6% (4).

Most expert agreed that duodenal polyposis should be manage based on Spigelman staging based on villous changes and severity of dysplasia (Table 1) (5,6). However Spigelman stage did not predict risk of ampullary cancer as it ony predict duodenal cancer(7). For Stage 1 and Stage 2, recommended for surveillance. For Stage 3, require medical or endoscopic treatment while Stage 4, surgical option should be considered in patient age more than 35 years old (8). As for surveillance OGDS for duodenal polyposis in FAP patient, recommendations are; stage 0 every 5 years; stage I every 3-5 years; stage II every 3 years; stage III every 6–12 months with consideration for surgery. For stage IV recommended to consider duodenectomy (5,6,8)(Table 2). Surgical options include pylorus-preserving pancreaticoduodenectomy (PPPDR), pancreas-sparing duodenectomy, duodenotomy with surgical polypectomy, and ampullectomy (9). Duodenotomy with polypectomy is the least preferred, as it has been associated with up to100% recurrence of adenomas within 6–36 months (10). As for our case his Spigelman score was 5, he was planned for re scope in 3 years

Table 1 Spigelman scoring system for duodenal adenomas

Variables	1 point	2 points	3 points
Number of polyps	1-4	5-20	>20
Polyp size (mm)	1-4	5-10	>10
Histology	Tubular	Tubulovillous	Villous
Dysplasia	Mild	Moderate	Severe

*The Spigelman score is calculated by adding the scores from each of the 4 categories (polyp number, polyp size, histology, and dysplasia), yielding a score ranging from 0-12

Table 2 Recommendations based on Spigelman stage

Spigelman score	Spigelman stage	Recommendation
0	0	Re-scope in 5 years
1-4	I	Re-scope in 3-5 years
5-6	II	Re-scope in 3 years
7-8	III	Re-scope in 1 year
9-12	IV	Consider duodenectomy vs. re-scope in 6 months

CONCLUSION

This case emphasizes the importance of having the standardize guideline in treating rare case of Gardner Syndrome. It is important to highlight to the patient the need of surveillance scope even though patient was asymptomatic, as further treatment will be decided based on scope findings and its risk of malignant transformation.

Conflict of interest
No conflict of interest was declared by the authors.

REFERENCES

1.Bodmer WF, Bailey CJ, BodmerJ, et al. Localization of the gene for familial adenomatous polyposis on chromosome 5. Nature. 1987; 328:614-6.
2.Bisgaard ML, Fenger K, Bulow S, et al. Familial adenomatous polyposis (FAP): frequency, penetrance, and mutation rate. Hum Mutat. 1994; 3:121-5.
3.B¨ulow S, Bj¨ork J, Christensen IJ, et al. Duodenal adenomatosis in familial adenomatous polyposis. The DAF Study Group. Gut 2004; 53:381–6.
4. Park JC, Park KJ, Ahn YO, et al. Risk of gastric cancer among Korean familial adenomatous polyposis (FAP) patients. Proc Ann Meet Am Sot Clin Oncol 992, 11.
5. Spigelman AD, Williams CB, Talbot IC, Domizio P, Phillips RK. Upper gastrointestinal cancer in patients with familial adenomatous polyposis. Lancet 1989;2:783-785.
6. Brosens LA, Keller JJ, Offerhaus GJ, Goggins M, Giardiello FM. Prevention and management of duodenal polyps in familial adenomatous polyposis. Gut 2005;54:1034-43.
7. Spigelman AD et al. Studies of the foregut in patients with familial adenomatous polyposis (FAP): Clinical problem, management and pathogenesis, MD thesis. Sydney: University of Sydney, 1996
8. LAA Brosens, JJ Keller et al. Prevention and management of duodenal polyps in familial adenomatous polyposis. Gut 2005; 54:1034-43
9. De Vos tot Nederveen Cappel WH, J¨arvinen HJ, Bj¨ork J, et al. Worldwide survey among polyposis registries of surgical management of severe duodenal adenomatosis in familial adenomatous polyposis. Br J Surg 2003; 90:705–10.
10. Penna C, Bataille N, Balladur P, et al. Surgical treatment of severe duodenal polyposis in familial adenomatous polyposis. Br J Surg 1998; 85:665–8.