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

Nik Amin Sahid¹, Firdaus Hayati¹, Ikhwan Sani ², Zaidi Zakaria²

¹Surgery Department, Faculty of Medicine and Health Sciences, Universiti Malaysia Sabah, 88800 Kota Kinabalu, Sabah, Malaysia
²Surgery Department, School of Medical Sciences, Hospital Universiti Sains Malaysia,16150, Kubang Kerian, Kelantan, Malaysia

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’ın özellikle gastrik ve duodenal polipozlu hastada FAP’nin ekstrakolonik yönetimini hakkındaki en son önerilerle ilgili literatür gözden geçirilmiştir. Çoğu uzman, duodenal polipozun Spigelman evrelemesi dayanarak yönetimini gerektiği konusunda hemfikirdir. Evre 1 ve Evre 2 için, gözetim için önerir. Evre 3 için, Dzięki veya endoskopik tedavi gerektirirken Evre 4, 35 yaşın üzerindeki hasta yaşlanında cerrahi şekeşen 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). Characterized 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 have polyposis elsewhere still controversial in terms of extent 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 history of FAP who presented with complained of altered bowel habit for almost 2 years. Further workup 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.

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 managed 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 only 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 to 100% 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 1:4 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 a standardized 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.
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