Advanced colon cancer before the age of 20 years: A case for extension of the current colonoscopy surveillance guidelines in hereditary nonpolyposis colorectal cancer syndrome

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BACKGROUND: Hereditary nonpolyposis colorectal cancer (HNPCC) currently accounts for between 2% to 6% of all colorectal adenocarcinomas. Controversies exist regarding the current guidelines for colonoscopic screening for colon cancer.

CASE REPORT: A case of colon cancer in a Japanese 19-year-old man with a family history of colon cancer that did not meet the criteria for HNPCC is reported. A malignant pelvic mass discovered shortly before his 20th birthday prompted a colonoscopy. The findings at colonoscopy determined that the patient and his family fulfilled the criteria of HNPCC.

CONCLUSION: Before finding a pelvic mass metastatic from adenocarcinoma of the ascending colon, this patient was clearly outside of the current guidelines for HNPCC screening. It is suggested that in similar patients, even if they do not fulfill all the criteria for HNPCC, it would be appropriate to consider screening well before the recommended lower age.

Key Words: Cancer; Colon; Colonoscopy; Hereditary; Surveillance

CASE PRESENTATION

A 19-year-old Japanese man presented to the emergency department with a complaint of transient self-limited left lower quadrant discomfort, a brief episode of fever and an isolated episode of passing a small amount of blood per rectum on one occasion. There was no previous history of passing blood per rectum and he was otherwise well, except for a 1.8 kg weight loss. His hematological parameters were unremarkable; specifically, his hemoglobin was 143 g/L (normal range 135 g/L to 175 g/L) with a normal mean cell volume. A flexible sigmoidoscopy performed 20 days after his emergency department presentation was unremarkable and he had not had a second episode of rectal bleeding in the intervening time period. His previous symptoms had resolved completely. The only remarkable feature about this patient was that his mother had undergone a right hemicolectomy 12 years previously at the age of 34 years for colon cancer in Japan. She had undergone annual

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The diagnosis of HNPCC is based on guidelines known as the Amsterdam criteria (3), which includes the following:

- At least three relatives with colorectal cancer, one of whom must be a first degree relative of the other two;
- Involvement of two or more generations;
- At least one case diagnosed before the age of 50 years; and
- Familial adenomatous polyposis has been excluded.

It has clearly been established that screening is necessary because of the history of rapid adenoma to carcinoma progression in patients manifesting HNPCC. In one 15 year trial, colonoscopic screening at three-year intervals more than halves the risk of colorectal cancer, prevents deaths due to colorectal cancer, and decreases overall mortality by approximately 65% in HNPCC families (4). A task force organized by the National Human Genome Research Institute, has recommended colonoscopy every one to three years starting at age 25 years for individuals known to have HNPCC-associated mutations (1). Lynch et al (2) have suggested annual colonoscopies starting at the age of 20 years. With our current patient, clearly, there was advanced cancer present before the age of 20 years. This patient’s family history was suggestive of HNPCC but the clinical diagnosis of the syndrome was not fulfilled until he himself was diagnosed with colon cancer. Unfortunately, in this situation there are no current recommendations for screening. This case illustrates the limitations of the current guidelines as it pertains to young people. In this era of quality assurance audits with respect to invasive procedures and medical insurance billing, we suggest that in similar cases, it would be appropriate for both physicians and patients to consider colonoscopic screening well before the lower age limit of current guidelines. In the future, the availability of commercial assays for HNPCC genetic marker screening, as well as sensitive and specific noninvasive colonoscopic imaging, such as three dimensional virtual colonoscopy (5), will make this suggestion easier to accept.

CONTRIBUTIONS: Dr Victor Wong wrote the preliminary discussion and compiled the reference list. Dr Yoshida wrote the preliminary introduction and case presentation section. Dr Salh rewrote the paper for its final presentation. Drs Ryan and Ho reviewed all of the computerized tomography scans and selected Figure 1. They also wrote the legend to Figure 1. There are no competing interests to declare.

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REFERENCES

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Figure 1) Contrast-enhanced transverse computerized tomography image demonstrating a large, heterogeneously enhancing, partially necrotic pelvic mass, a loop of dilated, thick-walled adjacent small bowel (a) and a trace of ascites (b). Normal sigmoid colon is shown posterior to the mass.