Emergency Total Proctocolectomy in an Uninsured Hispanic Man with Colorectal Adenocarcinoma Secondary to Familial Adenomatous Polyposis

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Background:

FAP is a rare genetic disorder classically inherited in an autosomal dominant pattern, which affects about 1 in 8 300 individuals. The Hispanic population has limited data regarding the spectrum of FAP mutation and clinical manifestation, although there is significant anecdotal evidence that the prevalence might be higher, with one only known Hispanic familial cancer registry in Puerto Rico.

Case Presentation:

We are reporting the case of a 25-year-old Hispanic gentleman with a strong family history of Familial Adenomatous Polyposis (FAP) and Colorectal Cancer (CRC) who presented for evaluation of abdominal pain, recurrent bloody stools, and profound weight loss. Initial Hb was 7.2 g/dL, and abdominal examination showed generalized rigidity and tenderness worse in the left lower quadrant. Colonoscopy revealed multiple large, non-bleeding polyps in the entire colon and up to the dentate line. The pathology report was positive for tubulovillous adenoma, while the surgical pathology report showed grade 2 moderately differentiated adenocarcinoma. Immunohistochemical stains were positive for the expression of MLH1, PMS2, MSH2, and MSH6 mismatch repair proteins. The patient subsequently had laparoscopic-assisted proctocolectomy with Brooke ileostomy. His hospital course was uneventful, and he was discharged home to follow up with medical oncology, surgery, and primary care.

Conclusion:

To reduce the associated healthcare costs and morbidity and mortality of cancer in general, especially those with associated strong risk factors such as FAP and CRC, early genetic counseling, timely screening, appropriate risk-reducing medical and surgical interventions, and regular lifetime follow-up of index cases are crucial. It is also imperative to promote health literacy, especially in communities with low socio-economic status who are often at a disadvantage. Lastly, there is a need for continued research on FAP, especially in minority populations, with increased promotion and use of familial cancer registries to reduce the overall burden.

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