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Management of an adolescent with familial adenomatous polyposis: Report of a case

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Familial adenomatous polyposis (FAP) is a syndrome inherited in an autosomal dominant trait which is clinically diagnosed upon identification of >100 colorectal adenomatous polyps. Additional characteristics are several extracolonic manifestations and malignancies. It is a consequence of mutation of a specific tumor supressor gene. Polyps of benign nature begin to appear in childhood or adolescence and if not treated early enough, one or more inevitably progress to cancer. Consequently, identifying and screening for FAP commences in adolescence. Colectomy is unavoidable in this group of patients, yet the decision is not always straightforward. The age and timing at which prophylactic colectomy is performed is not fixed, nor there are evidence to guide us at which point colectomy should be performed based on polyp burden. A 16-year-old male patient with abdominal pain and iron deficient anemia was referred for evaluation at our Clinic. Personal history was unremarkable while family history was positive for FAP syndrome with all second-and third-degree relatives from mother's side being affected. Our patient's mother had subtotal proctolectomy at the age of 36 and subsequent operation for abdominal wall dermatofibrosarcoma. Our patient's 3 siblings were symptom free and unevaluated at the time this article was written. The patient's father was healthy. We performed colonoscopy and detected < 50 polyps throughout the colon with diameter < 1 cm. Endoscopic resection was performed on 15 larger polyps and the histologic finding was consistent with tubular adenoma with low-to high-grade dysplasia. On screening for polyps of the upper gastrointestinal tract (GI) with esophagogastroduodenoscopy no polyps were detected. The finding of high-grade dysplasia would favor a sooner colectomy, still we aimed at postponing the surgery as much as possible given the family history for desmoid disease and patient's age and phenotype. Our management plan included annual surveilance colonoscopies and polypectomies, upper GI endoscopic surveillance and endoscopic resection of adenomas as well as annual abdominal and thyroid ultrasound. His siblings were scheduled for endoscopic evaluation. Family genetic testing was also advised (diagnostic and predictive) but due to technical difficulties it was not completed by the time this article was written.

Keywords: Familial adenomatous polyposis, polyposis, adolescent, colonoscopy, colorectal cancer