Gardner’s Syndrome with Desmoid Tumor
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Introduction:
• Gardner’s Syndrome (GS) is a variant of Familial Adenomatosis Polyposis (FAP) characterized by the presence of benign and/or malignant extraintestinal lesions in association with colonic polyposis.
• APC gene mutation is responsible for both FAP and GS.
• Desmoid tumors account for 0.03% of all neoplasms, but may be seen in 10-20% of patients with GS.

Case Description:
• A 21-year-old Caucasian female was admitted to our institution for worsening fatigue and hematochezia.
• She reported having 5-6 episodes of bloody diarrhea daily for the past 5 years and a weight loss of 40 pounds over the past one year.
• Family history significant for father with a total colectomy at the age 35 for unknown reasons.

Physical Examination:
• Vital Signs:
  - BP 104/68
  - PR 94/min
  - RR 16/min
  - Temp 98.1 F
  - Pulse Ox 97% on RA
  - BMI 13.1
• General: extremely cachectic
• HEENT: mildly pale, anicteric
• CHEST: clear to auscultation
• ABDOMEN: soft, mild tenderness with deep palpation in the lower abdomen, palpable para umbilical mass, no rebound tenderness or guarding.

Diagnostic Data:
• Hgb 13.0 g/dL (2 wks ago 5.7 g/dL)
• Hct 42.4 %
• MCV 71.4 fl
• WBC 32.9 K/ mcL
• MCV 71.4 fl
• Pre albumin 5.5 mg/dL(18-45 mg/DL)
• HIV negative
• Iron 13 mcg/dL
• Percent iron saturation 10
• TIBC 124 mcg/dL
• Ferritin 33 ng/mL
• Vit B 12 411 Pg/mL
• Creatinine 0.4 mg/dL
• Folate 4.4 ng/mL
• TSH 1.4 mcUnits/mL
• Celiac panel – negative

Clinical Course:
• She underwent colonoscopy which revealed hundreds of single and coalescing polypoid lesions with multiple large mass lesions preventing the colonoscope from reaching the proximal transverse colon. (Figure 1)
• Biopsies of the polypoid lesions revealed adenomatous tissue. (Figure 2)
• Genetic testing confirmed FAP.
• She ultimately underwent a two-stage total proctocolectomy with ileoanal anastomosis. (Figure 3) There were no signs of metastasis.
• A large mesenteric desmoid tumor was also resected during surgery. (Figure 4)
• She is currently doing well.

Discussion:
• Desmoid tumors are locally aggressive tumors that do not metastasize.
• In patients with GS, the most commonly involved sites include the intra-abdominal cavity (mesentery) and abdominal wall.
• These tumors are rarely symptomatic due to their indolent growth, but may present as intestinal obstruction or ischemia from local compression of surrounding organs.
• Desmoid tumors are responsible for death in up to 11% of patients with FAP.
• These tumors may be observed, but surgical resection is indicated when they become symptomatic. Radiation therapy is effective in patients with high surgical risk.
• These tumors have a high rate of recurrence after resection despite negative margins.

References: