3/2/2020
A 40-year-old female presents with history of attention deficit hyperactivity disorder, autism, and history of glycogen acanthosis. She has several subcutaneous lipomas and a history of thyroid cancer. She also mentions difficulty finding hat sizes that fit. She presents with rectal bleeding. She describes passing brown stool mixed with red blood for the past week. Her chemistries and blood counts are unremarkable. What do you recommend next:

A. Reassurance. Her blood counts are normal, and presentation is consistent with bleeding hemorrhoids
B. Colonoscopy
C. Sitz baths, increased fiber intake, and preparation H if the bleeding continues
D. Repeat blood counts in one week

Answer: B
Rationale: This individual likely has PTEN-hamartoma / Cowden’s syndrome. Syndrome features include head size >97th percentile (difficulty finding a hat that fits), glycogen acanthosis, lipomas, & thyroid cancer. Individuals develop polyps throughout their gastrointestinal tract, particularly in their stomachs and colons. The prevalence of colon polyps reaches 90% and the risk of colon cancer ranges from 9-13%. Her bleeding is suspicious for a lower rectal source, and in addition to hemorrhoids, the differential should also include a bleeding polyp or colorectal cancer. Thus, choice B is the correct.