**Question 39 – May 4**

A 43 year old female presents because she is worried about her risk of developing colon cancer. She tells you that several of her family members have died of cancer some as young as 40. She has never had a colonoscopy before. You perform an upper endoscopy and colonoscopy. The colonoscopy reveals several small (less then 1 cm) polyps. Pathology reveals all of them to be either hamartomatous or ganglioneuromas. The upper endoscopy multiple hamartomatous polyps and ganglioneromas in the stomach and duodenum. Biopsy of multiple small esophageal nodules reveals glycogenic acanthosis. Genetic testing of this patient is likely to reveal a mutation in which of the following genes?

A. APC
B. MLH-1
C. PTEN
D. STK11
E. SMAD4

Answer: C

This patient has Cowden syndrome. Cowden syndrome is an autosomal dominant hamartoma tumor syndrome. Patients are at increased risk of developing tumors of the colon, small bowel, breast, and thyroid cancer. The mutation responsible for this is found in the PTEN gene.

Reference:

Germline mutations of the PTEN gene in Cowden disease, an inherited breast and thyroid cancer syndrome. Liaw D, Marsh DJ, Li J, Dahia PL, Wang SI, Zheng Z, Bose S, Call KM, Tsou HC, Peacocke M, Eng C, Parsons R. Nat Genet. 1997 May;16(1):64-7.