

Question 18 – December 1

Juvenile polyposis is caused by a mutation in which of the following genes?

- A. SMAD4
- B. PTEN
- C. APC
- D. MLH1
- E. PMS2

Answer: A

Juvenile polyposis is caused by a mutation to the SMAD4 gene. Mutations in PTEN lead to Cowden syndrome. APC mutations are responsible for FAP. MLH1 and PMS2 mutations are part of HNPCC (Lynch syndrome)

References:

1. Nat Clin Pract Gastroenterol Hepatol CME. 2007;4(9):492-502.
2. Guidelines on Genetic Evaluation and Management of Lynch Syndrome: A Consensus Statement by the US Multi-Society Task Force on Colorectal Cancer . American Journal of Gastroenterology. August 2014