

Question 20 – December 12

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

PTEN mutation is seen in Cowden Syndrome. APC gene mutation is associated with FAP and MLH1 and PMS2 can be mutated in Lynch Syndrome.

Reference:

Hampel, Heather, et al. "A practice guideline from the American College of Medical Genetics and Genomics and the National Society of Genetic Counselors: referral indications for cancer predisposition assessment". *Genetics in Medicine* (2014).