

Question 1 – Week of December 5

You are seeing an asymptomatic 15 year old male whose mother is concerned may have familial adenomatous polyposis (FAP). His father has had a total ileo-proctocolectomy after his diagnosis of FAP in his mid-20's. What is the most cost effective initial screening test for this patient?

- A) Colonoscopy
- B) FAP gene analysis
- C) Flexible sigmoidoscopy
- D) 5q loss of heterozygosity DNA testing

Answer: C

FAP is an autosomal dominant hereditary polyposis disease affecting the APC gene causing 100's-1000's of polyps throughout the colon and invariably leading to the development of early colon cancer. This patient has a 50% chance of being affected. An initial screening flexible sigmoidoscopy would be most appropriate to see if he had rectosigmoid polyps. If any polyps were seen then more formal genetic testing would be involved. The father would need to have a known mutation in the FAP gene before genetic testing would be warranted. Genetic testing may reveal mutation in only 80% of affected individuals (Giardiello FM et al, NEJM 1997).