Question 27 - February 4

A 42-year-old male presents to your clinic due to concern that he may be at an increased risk for developing pancreatic cancer because of a strong personal history. His past medical history is significant for surgical resection of early stage melanomas at ages 29, 33, and 37. Based on this you determine that genetic testing is warranted.

Which gene mutation do you believe that he and his family likely have?

A. PRSS1  
B. SPINK1  
C. BRCA1/BRCA2  
D. P16/CDKN2A  
E. VHL

Answer: D

FAMMM syndrome (familial atypical multiple mole melanoma is characterized by a P16/CDKN2A mutation). Patients get melanoma at an early age, with the average being age 34. They are at an increased risk of developing pancreatic cancer (13-39 fold). PRSS-1 (hereditary pancreatitis), MLH1 (Lynch syndrome), BRCA1/2 (hereditary breast and ovarian cancer), and VHL (Von-Hippel Lindau) are all associated with increased risk of pancreatic cancer but not melanoma. The risk of pancreatic cancer with a SPINK (hereditary pancreatitis) mutation is unknown.