Question 42 – Week of April 18

The most common gene mutation in hereditary pancreatitis is:

A. Cationic trypsinogen gene (PRSS1)
B. MLH
C. Mesotrypsin gene (PRSS3)
D. K-ras
E. p53

Answer: A

PRSS1 Hereditary chronic pancreatitis is a rare form of early onset pancreatitis. Mutations in the PRSS1 gene encoding cationic trypsinogen increases autocatalytic conversion of trypsinogen to trypsin, leading to premature intrapancreatic trypsinogen activation. This leads to intrapancreatic imbalance of proteases and their inhibotors. Several other gene mutations such as PRSS2, SPINK1 and CFTR have also been implicated in hereditary pancreatitis.