Question 13 – Week of March 5

Hereditary pancreatitis is a syndrome of recurrent acute pancreatitis often leading to chronic pancreatitis where the pancreatitis phenotype appears inherited in an autosomal dominant pattern. What is the most common cause?

A. SPINK 1 mutation
B. CFTR mutation
C. PRSS1 mutation
D. SBDS gene mutation
E. None of the above

Answer C:

Hereditary pancreatitis denotes a specific familial pancreatitis , which is defined as a pancreatitis of any cause that predisposes an individual from a family to develop pancreatitis more than chance alone. The most common causes of familial pancreatitis are mutations in CFTR and SPINK 1, but these are autosomal recessive disorders linked with inherited heterozygous and compound heterozygous mutations in these genes. SBDS gene mutations cause Shcwachman Diamond syndrome and is autosomal recessive. Mutations in the cationic trypsinogen gene or PRSS1 cause an autosomal dominant inheritance of hereditary pancreatitis.

Reference:

Feldman: Sleisenger and Fordtran's Gastrointestinal and Liver Disease, 9th ed. Chapter 57. Hereditary, familial and Genetic Disorders Affecting the Pancreas