Hereditary Pancreatitis is caused by mutations in:

A. The pancreatic secretory trypsin inhibitor  
B. The Cystic Fibrosis Transmembrane Conductance Regulator (CFTR)  
C. Chymotrysinogen  
D. Cationic trypsinogen  
E. Procarboxypeptidase A1

The recommended response is D.

Hereditary pancreatitis is distinct clinical entity inherited as an autosomal dominant trait with incomplete penetrance and caused by mutations in cationic trypsinogen (PRSS1). While specific trypsinogen mutations cause disease, the disease mechanism remains unclear. Classic Cystic Fibrosis (CF) is associated with pancreatic insufficiency and chronic pancreatitis. Minor mutations in CFTR that do not cause classic CF can occasionally cause chronic pancreatitis. Mutations in the pancreatic trypsin inhibitor (SPINK1) may increase the risk of chronic pancreatitis in susceptible individuals (e.g. those with minor CFTR mutations), but do not appear themselves to cause chronic pancreatitis.


Chapter 2: Training in Biliary Tract Diseases and Pancreatic Disorders

DDSEP Chapter 8: Question 2

Which of the following is true regarding gallbladder motility?

A. A gallbladder ejection fraction of less than 50% indicates dyskinesia
B. Patients with gallbladder dysmotility commonly have histologic changes of chronic cholecystitis
C. Contractile function is mediated by secretin
D. Short-chain fatty acids are the most potent stimulators of intestinal CCK release
E. Somatostatin release is associated with accelerated gallbladder emptying time

The recommended response is B.

Biliary dysmotility is usually associated with specific conditions such as gallstones or choledocholithiasis (Hyperlink to Gallbladder Dysmotility section). Biliary dyskinesia and sphincter of Oddi dysfunction are the two main categories of biliary motility disorders. Most patients present with unexplained chronic abdominal pain which has some features of biliary-type pain. The duration of pain, however, is often long-standing and may resemble luminal gastrointestinal disorders such as irritable bowel syndrome or functional dyspepsia.

Radionuclide scintigraphy (e.g. HIDA scan) enables the calculation of a gallbladder ejection fraction to objectively assess the severity of dysmotility. Biliary dyskinesia is defined as a gallbladder ejection fraction of less than 35% in the absence of cholelithiasis. Conflicting evidence exists regarding the effectiveness of cholecystectomy for the treatment of biliary dyskinesia. Relief of symptoms has been reported in 50-85% of cases. These patients have histologic changes of chronic cholecystitis in most cases.

Gallbladder contractility is regulated by a neurohumoral axis stimulated by the fat content of ingested food, vagal nerve innervation, and release of CCK. Long-chain fatty acids are the most potent stimulus for the intestinal release of CCK which mediates postprandial gallbladder contraction and sphincter of Oddi relaxation. Somatostatin acts as a physiological inhibitor of gallbladder contraction.

Which one of the following is not a poor prognostic sign for pancreatic cancer at presentation:

A. Weight loss  
B. Back pain  
C. **Jaundice**  
D. Depression  
E. Fatigue

The recommended response is C.

The fact that most symptoms are so non-specific can lead to significant delays in diagnosis of pancreatic cancer. Obstructive jaundice from a pancreatic head mass usually prompts a work-up and may pick up early, resectable lesions, while weight loss, depression, and fatigue are usually indicative of high tumor burden and or disseminated disease and back pain may indicate local invasion.