The SPINK1 N34S variant is associated with acute pancreatitis.

Abstract:

OBJECTIVE: Acute pancreatitis (AP) is a disease whose pathogenesis remains largely obscure. Genetic research has focused attention upon the role of the pancreatic protease/protease inhibitor system. The aim of this study was to investigate the prevalence of genetic variants of the trypsin inhibitor, SPINK1, in acute pancreatitis.

METHODS: We genotyped 468 patients with AP and 1117 healthy controls for SPINK1 alterations by single-strand conformation polymorphism analysis and by melting curve analysis using fluorescence resonance energy transfer probes.

RESULTS: The c.101A>G (p.N34S) variant was detected in 24/936 alleles of patients and in 18/2234 alleles of healthy controls (odds ratio=3.240; 95% confidence interval: 1.766-5.945; PT (p.P55S) variant did not differ between patients and controls.

CONCLUSION: The SPINK1 N34S variant is associated with acute pancreatitis. This supports the importance of premature protease activation in the pathogenesis of AP and suggests that mutated SPINK1 may predispose certain individuals to develop this disease.