

SPINK1

c.*34C>T

c.274C>T

Citations:

Bhatia E, Choudhuri G, Sikora SS, Landt O, Kage A, Becker M, Witt H. (2002) **Tropical calcific pancreatitis: strong association with *SPINK1* trypsin inhibitor mutations.** Gastroenterology 123, 1020-1025

3 affected, 2 unaffected; all carried c.*32C>T (reported as 272C>T) on the same allele; 1 affected also carried p.N34S Variant was reported as 274C>T

O'Reilly DA, Witt H, Rahman SH, Schulz HU, Sargen K, Kage A, Cartmell MT, Landt O, Larvin M, Demaine AG, McMahon MJ, Becker M, Kingsnorth AN. (2008) **The *SPINK1* N34S variant is associated with acute pancreatitis.** Eur J Gastroenterol Hepatol 20, 726-731

1 affected, also carried c.*32C>T (reported as 272C>T) on the same allele Variant was reported as 274C>T; erroneously described as novel variant