

SPINK1 **c.*32C>T** **c.272C>T**

rs11319

Citations:

Witt H, Luck W, Hennies HC, Classen M, Kage A, Lass U, Landt O, Becker M. (2000) **Mutations in the gene encoding the serine protease inhibitor, Kazal type 1 are associated with chronic pancreatitis.** Nat Genet 25, 213-216

Common polymorphism

Variant was reported as 272C>T

Pfützer RH, Barmada MM, Brunskill AP, Finch R, Hart PS, Neoptolemos J, Furey WF, Whitcomb DC. (2000) **SPINK1/PSTI polymorphisms act as disease modifiers in familial and idiopathic chronic pancreatitis.** Gastroenterology 119, 615-623

9 affected, 10 unaffected

Variant was reported as 272C>T

Kaneko K, Nagasaki Y, Furukawa T, Mizutamari H, Sato A, Masamune A, Shimosegawa T, Horii A. (2001) **Analysis of the human pancreatic secretory trypsin inhibitor (PSTI) gene mutations in Japanese patients with chronic pancreatitis.** J Hum Genet 46, 293-297

5 affected, 4 unaffected

Variant was reported as 272C>T

Rossi L, Pfützer RH, Parvin S, Ali L, Sattar S, Kahn AK, Gyr N, Whitcomb DC. (2001) **SPINK1/PSTI mutations are associated with tropical pancreatitis in Bangladesh. A preliminary report.** Pancreatology 1, 242-245

8 affected, all included in Schneider et al. (2002); not counted

Variant was reported as 272C>T

Kuwata K, Hirota M, Sugita H, Kai M, Hayashi N, Nakamura M, Matsuura T, Adachi N, Nishimori I, Ogawa M. (2001) **Genetic mutations in exons 3 and 4 of the pancreatic secretory trypsin inhibitor in patients with pancreatitis.** J Gastroenterol 36, 612-618

6 affected, 5 unaffected

Variant was reported as c.*35C>T (the authors probably included the stop codon in the count)

Schneider A, Suman A, Rossi L, Barmada MM, Beglinger C, Parvin S, Sattar S, Ali L, Khan AK, Gyr N, Whitcomb DC. (2002) **SPINK1/PSTI mutations are associated with tropical pancreatitis and type II diabetes mellitus in Bangladesh.** Gastroenterology 123, 1026-1030

26 affected, 17 unaffected; preliminary report by Rossi et al. (2001); all variants counted here

Variant was reported as 272C>T

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

28 affected, 24 unaffected

Variant was reported as 272C>T

Weiss FU, Simon P, Witt H, Mayerle J, Hlouschek V, Zimmer KP, Schnekenburger J, Domschke W, Neoptolemos JP, Lerch MM. (2003) ***SPINK1* mutations and phenotypic expression in patients with pancreatitis associated with trypsinogen mutations.** J Med Genet 40, e40
3 affected; also carried *PRSSI* p.R122H

Kuwata K, Hirota M, Nishimori I, Otsuki M, Ogawa M. (2003) **Mutational analysis of the pancreatic secretory trypsin inhibitor gene in familial and juvenile pancreatitis in Japan.** J Gastroenterol 38, 365-370
6 affected; possible overlap with Kuwata et al. (2001); not counted
Variant was reported as 272C>T

Hirota M, Kuwata K, Ohmuraya M, Ogawa M. (2003) **From acute to chronic pancreatitis: the role of mutations in the pancreatic secretory trypsin inhibitor gene.** JOP 4, 83-88
6 affected; same subjects as in Kuwata et al. (2003); not counted
Variant was reported as 272C>T

Inoue N, Ito T, Akashi T, Kawabe K, Oono T, Gibo J, Arita Y, Nawata H, Funakoshi A. (2004) **Acute pancreatitis in the early stages of pregnancy associated with a PSTI gene mutation.** Pancreas 29, 242-243
1 affected; with history of acute pancreatitis; sister to the index patient
Index patient was a pregnant woman with p.N34S inherited from father (no c.*32C>T)

Kume K, Masamune A, Mizutamari H, Kaneko K, Kikuta K, Satoh M, Satoh K, Kimura K, Suzuki N, Nagasaki Y, Horii A, Shimosegawa T. (2005) **Mutations in the serine protease inhibitor Kazal Type 1 (*SPINK1*) gene in Japanese patients with pancreatitis.** Pancreatology 5, 354-360
19 affected
Variant was reported as 272C>T

Tzetis M, Kaliakatsos M, Fotoulaki M, Papatheodorou A, Doudounakis S, Tsezou A, Makrythanasis P, Kanavakis E, Nousia-Arvanitakis S. (2007) **Contribution of the *CFTR* gene, the pancreatic secretory trypsin inhibitor gene (*SPINK1*) and the cationic trypsinogen gene (*PRSSI*) to the etiology of recurrent pancreatitis.** Clin Genet 71, 451-457
2 affected
Variant was reported as 272C>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.*34C>T (reported as 274C>T) on the same allele
Variant was reported as 272C>T

Gullo L, Laghi L, Migliori M, Lucrezio L, Bianchi P, Randolph AE, Mantovani V, Bastagli L, Pezzilli R, Malesci A. (2008) ***SPINK1* and *PRSSI* mutations in benign pancreatic hyperenzymemia.** Pancreas 37, 31-35
1 unaffected; with hyperenzymemia

Chang YT, Wei SC, L PC, Tien YW, Jan IS, Su YN, Wong JM, Chang MC. (2009) **Association and differential role of PRSS1 and SPINK1 mutation in early-onset and late-onset idiopathic chronic pancreatitis in Chinese subjects.** Gut 58, 885

6 affected, 18 unaffected

Variant was reported as 272C>T

Tremblay K, Dubois-Bouchard C, Brisson D, Gaudet D. (2014) **Association of *CTRC* and *SPINK1* gene variants with recurrent hospitalizations for pancreatitis or acute abdominal pain in lipoprotein lipase deficiency.** Front Genet 5, 90

5 affected, 6 unaffected

Variant was indicated as T>C in Table 2