SPINK1 c.56-37T>C IVS1-37T>C

Variant is in linkage with variants c.87+268A>G, c.101A>G (p.N34S), c.195-606G>A, and c.195-66_65insTTTT

Citations:

Chen JM, Mercier B, Audrezet MP, Ferec C. (2000) **Mutational analysis of the human pancreatic secretory trypsin inhibitor (*PSTI*) gene in hereditary and sporadic chronic pancreatitis.** J Med Genet 37, 67-69
1 family with unspecified number of affected and 2 unaffected; counted as 1 affected

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
18 affected (6 homozygous)

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
29 affected (7 homozygous), 1 unaffected

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

Chandak GR, Idris MM, Reddy DN, Bhaskar S, Sriram PV, Singh L. (2002) **Mutations in the pancreatic secretory trypsin inhibitor gene (*PSTI/SPINK1*) rather than the cationic trypsinogen gene (*PRSS1*) are significantly associated with tropical calcific pancreatitis.** J Med Genet 39, 347-351
31 affected (8 homozygous), 3 unaffected

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
20 affected (1 homozygous), 1 unaffected; 5 affected overlaps with Rossi et al. (2001); all counted

Truninger K, Witt H, Köck J, Kage A, Seifert B, Ammann RW, Blum HE, Becker M. (2002) **Mutations of the serine protease inhibitor, Kazal type 1 gene, in patients with idiopathic chronic pancreatitis.** Am J Gastroenterol 97, 1133-1137
6 affected (1 homozygous), 4 unaffected

Gomez-Lira M, Bonamini D, Castellani C, Unis L, Cavallini G, Assael BM, Pignatti PF. (2003) **Mutations in the *SPINK1* gene in idiopathic pancreatitis Italian patients.** Eur J Hum Genet 11, 543-546
3 affected (1 homozygous), 1 heterozygous also carried *CFTR* p.L997F

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
9 affected, overlap with Kuwata et al. (2003); homozygotes not specified; 4 affected counted

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
7 affected (2 homozygous)

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
9 affected (1 homozygous), 1 unaffected

Keiles S, Kammesheidt A. (2006) **Identification of *CFTR, PRSS1,* and *SPINK1* mutations in 381 patients with pancreatitis.** Pancreas 33, 221-227
5 affected

Shimosegawa T, Kume K, Masamune A. (2006) ***SPINK1* gene mutations and pancreatitis in Japan.** J Gastroenterol Hepatol 21 Suppl 3, S47-51
11 affected (1 homozygous), 1 unaffected; possible overlap with Kume et al. (2005); 2 affected counted

Masamune A, Kume K, Takagi Y, Kikuta K, Satoh K, Satoh A, Shimosegawa T. (2007) **N34S mutation in the *SPINK1* gene is not associated with alternative splicing.** Pancreas 34, 423-428
2 affected (homozygous)

Masamune A, Kume K, Shimosegawa T. (2007) **Differential roles of the *SPINK1* gene mutations in alcoholic and nonalcoholic chronic pancreatitis.** J Gastroenterol 42 Suppl 17, 135-140
11 affected (1 homozygous), 1 unaffected, likely overlap with Shimosegawa et al. (2006); not counted

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 (*PRSS1*) to the etiology of recurrent pancreatitis.** Clin Genet 71, 451-457
2 affected

Singh S, Choudhuri G, Agarwal S. (2014) **Frequency of CFTR, SPINK1, and cathepsin B gene mutation in North Indian population: connections between genetics and clinical data.** Scientific World Journal, 2014:763195
11 affected, 1 unaffected

Functional studies:

Masamune A, Kume K, Takagi Y, Kikuta K, Satoh K, Satoh A, Shimosegawa T. (2007) **N34S mutation in the *SPINK1* gene is not associated with alternative splicing.** Pancreas 34, 423-428

Kereszturi E, Király O, Sahin-Tóth M. (2009) **Minigene analysis of intronic variants in common *SPINK1* haplotypes associated with chronic pancreatitis.** Gut 58, 545-549

Boulling A, Chen JM, Callebaut I, Férec C. (2012) **Is the SPINK1 p.Asn34Ser missense mutation per se the true culprit within its sssociated haplotype?** WebmedCentral GENETICS 2012; 3, WMC003084