SPINK1 c.−253T>C

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
Common polymorphism, ~20%

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

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
6 affected, 10 unaffected

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
6 affected, 43 unaffected

Bernardino AL, Guarita DR, Mott CB, Pedroso MR, Machado MC, Laudanna AA, Tani CM, Almeida FL, Zatz M. (2003) **CFTR, PRSS1 and SPINK1 mutations in the development of pancreatitis in Brazilian patients.** JOP 4, 169-177
20 affected, 20 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
11 affected, 16 unaffected (3 homozygous)

Patuzzo C, Castellani C, Sagramoso C, Gomez-Lira M, Bonamini D, Belpinati F, Dechecchi MC, Assael BM, Pignatti PF. (2003) **Cationic trypsinogen and pancreatic secretory trypsin inhibitor gene mutations in neonatal hypertrypsinaemia.** Eur J Hum Genet 11, 93-96
16 unaffected; with hypertrypsinemia

Chandak GR, Idris MM, Reddy DN, Mani KR, Bhaskar S, Rao GV, Singh L. (2004) **Absence of PRSS1 mutations and association of SPINK1 trypsin inhibitor mutations in hereditary and non-hereditary chronic pancreatitis.** Gut 53, 723-728
Common polymorphism, 3.5% patients, 27.9% controls

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
15 affected

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
4 affected, 5 unaffected

Tomaiuolo AC, Sofia VM, Surace C, Majo F, Genovese S, Petrocchi S, Grotta S, Alghisi F, Lucidi V, Angioni A. (2015) **Relationship between *CFTR* and *CTRC* variants and the clinical phenotype in late-onset cystic fibrosis disease with chronic pancreatitis.** J Mol Diagn 17, 171-178
1 unaffected; CF patient

Hegyi E, Geisz A, Sahin-Tóth M, Derikx M, Németh BC, Balázs A, Hritz I, Izbéki F, Halász A, Párniczky A, Takács T, Kelemen D, Sarlós P, Hegyi P, Czakó L. (2016) ***SPINK1* promoter variants in chronic pancreatitis.** Pancreas 45, 148-153
33 affected (5 homozygous), 20 unaffected

Functional studies:

Derikx MH, Geisz A, Kereszturi É, Sahin-Tóth M. (2015) **Functional significance of *SPINK1* promoter variants in chronic pancreatitis.** Am J Physiol Gastrointest Liver Physiol 308, G779-784

Hegyi E, Geisz A, Sahin-Tóth M, Derikx M, Németh BC, Balázs A, Hritz I, Izbéki F, Halász A, Párniczky A, Takács T, Kelemen D, Sarlós P, Hegyi P, Czakó L. (2016) ***SPINK1* promoter variants in chronic pancreatitis.** Pancreas 45, 148-153