

SPINK1

c.2T>C

p.M1T

p.M1?

rs104893938

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

1 family with 2 affected and 1 unaffected

Perri F, Piepoli A, Stanziale P, Merla A, Zelante L, Andriulli A. (2003) **Mutation analysis of the cystic fibrosis transmembrane conductance regulator (*CFTR*) gene, the cationic trypsinogen (*PRSSI*) gene, and the serine protease inhibitor, Kazal type 1 (*SPINK1*) gene in patients with alcoholic chronic pancreatitis.** Eur J Hum Genet 11, 687-692

1 unaffected; with alcoholic liver disease

Rosendahl J, Landt O, Bernadova J, Kovacs P, Teich N, Bödeker H, Keim V, Ruffert C, Mössner J, Kage A, Stumvoll M, Groneberg D, Krüger R, Luck W, Treiber M, Becker M, Witt H. (2013) ***CFTR*, *SPINK1*, *CTRC* and *PRSSI* variants in chronic pancreatitis: is the role of mutated *CFTR* overestimated?** Gut 62, 582-592

1 affected; likely same as in Witt et al. (2000); did not count

Masson E, Chen JM, Audrézet MP, Cooper DN, Férec C. (2013) **A conservative assessment of the major genetic causes of idiopathic chronic pancreatitis: data from a comprehensive analysis of *PRSSI*, *SPINK1*, *CTRC* and *CFTR* genes in 253 young French patients.** PLoS One 8, e73522

Variant was reported only at the protein level as p.M1?

1 affected; also carried p.N34S