

SPINK1

c.174C>T

p.C58=

p.C58C

rs35737774

Variant might be in linkage with variant c.-41G>A

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 affected

Variant was described at the protein level as C35C

Variant c.-41G>A was also found; unclear whether from same subject

Audrézet MP, Chen JM, Le Maréchal C, Ruszniewski P, Robaszkiewicz M, Raguénès O, Quéré I, Scotet V, Férec C. (2002) **Determination of the relative contribution of three genes-the cystic fibrosis transmembrane conductance regulator gene, the cationic trypsinogen gene, and the pancreatic secretory trypsin inhibitor gene-to the etiology of idiopathic chronic pancreatitis.** Eur J Hum Genet 10, 100-106

1 affected; likely the same as in Chen et al (2000); not counted

Variant was reported in linkage with c.-41G>A; evidence unclear

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

1 affected; also carried *CFTR* p.F508del and 3878delG

Variant c.-41G>A was also found; unclear whether from same subject

Schneider A, Pfützner RH, Barmada MM, Slivka A, Martin J, Whitcomb DC. (2003) **Limited contribution of the *SPINK1* N34S mutation to the risk and severity of alcoholic chronic pancreatitis: a report from the United States.** Dig Dis Sci 48, 1110-1115

1 affected