

***SPINK1* c.-41G>A**

Variant might be in linkage with c.174C>T (p.C58=)

rs79438751

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 c.174C>T (p.C58=) was also reported but unclear whether in same subject

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

3 unaffected

Audrezet 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); did not count

Linkage with c.174C>T (p.C58=) reported in same subject

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.174C>T (p.C58=) was also reported but unclear whether in same subject

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

8 affected (2 homozygous; also carried either *PRSSI* p.R122C or *CFTR* p.D1445N); 1 hetero also carried *CFTR* p.D1270N and p.R74W

Functional studies:

Boulling A, Witt H, Chandak GR, Masson E, Paliwal S, Bhaskar S, Reddy DN, Cooper DN, Chen JM, Férec C. (2011) **Assessing the pathological relevance of *SPINK1* promoter variants.** Eur J Hum Genet 19, 1066-1073

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