

## ***SPINK1***

**c.88-23A>T**

**IVS2-23A>T**

### 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 affected, 2 unaffected

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

1 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

2 unaffected

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 hypertrypsinemia.** Eur J Hum Genet 11, 93-96

2 unaffected; with hypertrypsinemia

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 affected

Gullo L, Laghi L, Migliori M, Lucrezio L, Bianchi P, Randolph AE, Mantovani V, Bastagli L, Pezzilli R, Malesci A. (2008) ***SPINK1* and *PRSSI* mutations in benign pancreatic hyperenzymemia.** Pancreas 37, 31-35

1 unaffected; with hyperenzymemia

Hegy E, Geisz A, Sahin-Tóth M, Derikx MH, 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

1 affected

### Functional studies:

Zou WB, Boulling A, Masson E, Cooper DN, Liao Z, Li ZS, Férec C, Chen JM. (2016) **Clarifying the clinical relevance of *SPINK1* intronic variants in chronic pancreatitis.** Gut 65, 884-886