

***SPINK1* c.-22C>T**

rs190219062

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

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

Keiles S, Kammesheidt A. (2006) **Identification of *CFTR*, *PRSS1*, and *SPINK1* mutations in 381 patients with pancreatitis.** *Pancreas* 33, 221-227

1 affected; also carried c.36G>C (p.L12F) and c.194>90A>T

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

1 unaffected

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