

SPINK1

c.194+184T>A

IVS3+184T>A

Citations:

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

5 affected

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

8 affected, 1 also carried *CFTR* p.S1235R; 1 also carried *CFTR* p.F508del

Table 3 indicated 5 affected subjects with IVS+184T>A with no intron number

Kopp BT, Pastore MT, Sturm AC, Holtzlander MJ, Westman JA. (2011) **A novel exon duplication of the cystic fibrosis transmembrane conductance regulator in a patient presenting with adult-onset recurrent pancreatitis**. *Pancreas* 40, 773-777

1 affected, also carried *CFTR* p.G551D and duplication of exon 19

Palermo JJ, Lin TK, Hornung L, Valencia CA, Mathur A, Jackson K, Fei L, Abu-El-Haija M. (2016) **Genophenotypic analysis of pediatric patients with acute recurrent and chronic pancreatitis**. *Pancreas*. 2016 May 19. [Epub ahead of print]

1 affected

Zou WB, Masson E, Boulling A, Cooper DN, Li ZS, Liao Z, Férec C, Chen JM. (2016) **Digging deeper into the intronic sequences of the *SPINK1* gene**. *Gut* 65, 1055-1056

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