

## ***SPINK1* c.-53C>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

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

2 affected; 1 unaffected, 1 affected is likely the same as in Witt et al. (2000); counted 1 affected

Rosendahl J, Landt O, Bernadova J, Kovacs P, Teich N, Bödeker H, Keim V, Ruffert C, Mössner J, Kage A, Stumvoll M, Groneberg D, Krüger R, Luck W, Treiber M, Becker M, Witt H. (2013) ***CFTR*, *SPINK1*, *CTRC* and *PRSSI* variants in chronic pancreatitis: is the role of mutated *CFTR* overestimated?** Gut 62, 582-592

1 affected; likely the same as in Witt et al. (2000); did not count

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

### 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