

## ***SPINK1***            **c.-147A>G**

### Citations:

Keiles S, Kammesheidt A. (2006) **Identification of *CFTR*, *PRSS1*, and *SPINK1* mutations in 381 patients with pancreatitis.** *Pancreas* 33, 221-227  
4 affected; 1 also carried *CFTR* p.W1282X; 1 also carried *SPINK1* p.M1R

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  
3 affected, 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