

SPINK1 **c.*131G>A**

Citations:

Weiss FU, Simon P, Witt H, Mayerle J, Hlouschek V, Zimmer KP, Schnekenburger J, Domschke W, Neoptolemos JP, Lerch MM. (2003) ***SPINK1* mutations and phenotypic expression in patients with pancreatitis associated with trypsinogen mutations.** J Med Genet 40, e40

Variant was described as 371G>A in the 3' flanking region

One unaffected subject; also carried *PRSSI* p.R122H

Tomaiuolo AC, Sofia VM, Surace C, Majo F, Genovese S, Petrocchi S, Grotta S, Alghisi F, Lucidi V, Angioni A. (2015) **Relationship between *CFTR* and *CTRC* variants and the clinical phenotype in late-onset cystic fibrosis disease with chronic pancreatitis.** J Mol Diagn 17, 171-178

One unaffected subject; CF patient