

SPINK1 **c.200G>A** **p.R67H**

rs35523678

Citations:

Gullo L, Laghi L, Migliori M, Lucrezio L, Bianchi P, Randolph AE, Mantovani V, Bastagli L, Pezzilli R, Malesci A. (2008) ***SPINK1* and *PRSSI* mutations in benign pancreatic hyperenzymemia.** *Pancreas* 37, 31-35

1 unaffected; with hyperenzymemia

Variant was incorrectly described at the protein level as p.R67G; electropherogram in Figure 2C confirms c.200G>A nucleotide change

Boulling A, Keiles S, Masson E, Chen JM, Férec C. (2012) **Functional analysis of eight missense mutations in the *SPINK1* gene.** *Pancreas* 41, 329-330

11 affected

Werlin S, Konikoff FM, Halpern Z, Barkay O, Yerushalmi B, Broide E, Santo E, Shamir R, Shaoul R, Shteyer E, Yaakov Y, Cohen M, Kerem E, Ruzzniewski P, Masson E, Férec C, Wilschanski M. (2015) **Genetic and electrophysiological characteristics of recurrent acute pancreatitis.** *J Pediatr Gastroenterol Nutr* 60, 675-679]

1 affected

Variant was described at the protein level only

Functional studies:

Boulling A, Keiles S, Masson E, Chen JM, Férec C. (2012) **Functional analysis of eight missense mutations in the *SPINK1* gene.** *Pancreas* 41, 329-330