SPINK1 c.199C>T p.R67C

Citations:

Kuwata K, Hirota M, Sugita H, Kai M, Hayashi N, Nakamura M, Matsuura T, Adachi N, Nishimori I, Ogawa M. (2001) **Genetic mutations in exons 3 and 4 of the pancreatic secretory trypsin inhibitor in patients with pancreatitis.** J Gastroenterol 36, 612-618
1 family with 1 affected and 2 unaffected, all carried p.N34S; 2 other family members were homozygous for p.N34S, 1 affected, 1 unaffected
Table 1 contradicts pedigree and lists family member #16 as idiopathic

Hirota M, Kuwata K, Ohmuraya M, Ogawa M. (2003) **From acute to chronic pancreatitis: the role of mutations in the pancreatic secretory trypsin inhibitor gene.** JOP 4, 83-88
2 affected; likely the same subjects as in Kuwata et al. (2001); not counted

Kuwata K, Hirota M, Nishimori I, Otsuki M, Ogawa M. (2003) **Mutational analysis of the pancreatic secretory trypsin inhibitor gene in familial and juvenile pancreatitis in Japan.** J Gastroenterol 38, 365-370
2 affected; 1 also carried p.N34S and overlaps with Kuwata et al. (2001); 1 affected counted

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 *PRSS1* variants in chronic pancreatitis: is the role of mutated *CFTR* overestimated?** Gut 62, 582-592
1 affected
Incorrectly reported at the protein level only as p.R67Q; confirmed by email from first author on 12-02-2013.

Masamune A. (2014) **Genetics of pancreatitis: the 2014 update.** Tohoku J Exp Med 232, 69-77
1 affected; also carried p.N34S

Functional studies:

Király O, Wartmann T, Sahin-Tóth M.(2007) **Missense mutations in pancreatic secretory trypsin inhibitor (SPINK1) cause intracellular retention and degradation.** Gut 56, 1433-1438

Boulling A, Le Maréchal C, Trouvé P, Raguénès O, Chen JM, Férec C. (2007) **Functional analysis of pancreatitis-associated missense mutations in the pancreatic secretory trypsin inhibitor (*SPINK1*) gene.** Eur J Hum Genet 15, 936-942