SPINK1 c.41T>C p.L14P

rs104893939

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

Király O, Boulling A, Witt H, Le Maréchal C, Chen JM, Rosendahl J, Battaggia C, Wartmann T, Sahin-Tóth M, Férec C. (2007) **Signal peptide variants that impair secretion of pancreatic secretory trypsin inhibitor (SPINK1) cause autosomal dominant hereditary pancreatitis.** Hum Mutat 28, 469-47
1 family with 2 affected; index patient same as in Witt et al. (2000); 1 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; likely the same as in Witt et al. (2000); not counted

Functional studies:

Király O, Boulling A, Witt H, Le Maréchal C, Chen JM, Rosendahl J, Battaggia C, Wartmann T, Sahin-Tóth M, Férec C. (2007) **Signal peptide variants that impair secretion of pancreatic secretory trypsin inhibitor (SPINK1) cause autosomal dominant hereditary pancreatitis.** Hum Mutat 28, 469-47

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