SPINK1 c.41T>G p.L14R

rs104893939

Citations:

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
2 families with 5 affected and 1 unaffected

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 overlaps with Király et al. (2007); 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