SPINK1 c.36G>C p.L12F

variant is in linkage with c.194+90A>T
African variant

rs35877720

Citations:

Keiles S, Kammesheidt A. (2006) **Identification of *CFTR, PRSS1,* and *SPINK1* mutations in 381 patients with pancreatitis.** Pancreas 33, 221-227
Variant was described at the protein level only
Text described 7 affected subjects, tables showed 8; counted as 8 affected
Always found together with c.194+90A>T
1 also carried c.-22C>T, 1 also carried *CFTR* 296+28A>G, 1 also carried *CFTR* 4375-20A>G, 2 also carried *PRSS1* p.R122H

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
15 affected, 33 unaffected; African variant

Gaitch N, Hubert D, Gameiro C, Burgel PR, Houriez F, Martinez B, Honoré I, Chapron J, Kanaan R, Dusser D, Girodon E, Bienvenu T. (2016) **CFTR and/or pancreatitis susceptibility genes mutations as risk factors of pancreatitis in cystic fibrosis patients?** Pancreatology 16, 515-522
2 unaffected (CF patients)

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