

SPINK1

c.87+1G>A

IVS2+1G>A

Citations:

Le Maréchal C, Chen JM, Le Gall C, Plessis G, Chipponi J, Chuzhanova NA, Raguénès O, Férec C. (2004) **Two novel severe mutations in the pancreatic secretory trypsin inhibitor gene (*SPINK1*) cause familial and/or hereditary pancreatitis.** Hum Mutat 23, 205

1 affected; also carried p.P55S

Masson E, Chen JM, Audrézet MP, Cooper DN, Férec C. (2013) **A conservative assessment of the major genetic causes of idiopathic chronic pancreatitis: data from a comprehensive analysis of *PRSS1*, *SPINK1*, *CTRC* and *CFTR* genes in 253 young French patients.** PLoS One 8, e73522

1 affected; also carried p.P55S; likely the same as in Le Maréchal et al. (2004); did not count

Functional studies:

Zou WB, Boulling A, Masson E, Cooper DN, Liao Z, Li ZS, Férec C, Chen JM. (2016) **Clarifying the clinical relevance of *SPINK1* intronic variants in chronic pancreatitis.** Gut 65, 884-886