

***SPINK1***

**c.195-478T>G**

**IVS3-478T>G**

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

Common polymorphism

Variant was incorrectly reported as IVS3-476T>G

Kalinin VN, Kaifi JT, Schwarzenbach H, Sergeyev AS, Link BC, Bogoevski D, Vashist Y, Izbicki JR, Yekebas EF. (2006) **Association of rare *SPINK1* gene mutation with another base substitution in chronic pancreatitis patients.** World J Gastroenterol 12, 5352-5356

Common polymorphism

Variant was incorrectly reported as IVS3-476T>G

Zou WB, Masson E, Boulling A, Cooper DN, Li ZS, Liao Z, Férec C, Chen JM. (2016) **Digging deeper into the intronic sequences of the *SPINK1* gene.** Gut 65, 1055-1056

2 affected, also carried c.194+1159C>G on the same allele

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

Zou WB, Masson E, Boulling A, Cooper DN, Li ZS, Liao Z, Férec C, Chen JM. (2016) **Digging deeper into the intronic sequences of the *SPINK1* gene.** Gut 65, 1055-1056