

SPINK1

c.195-323C>T

IVS3-323C>T

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 IVS3-321C>T

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 IVS3-321C>T

Hegyi E, Geisz A, Sahin-Tóth M, Derikx MH, Németh BC, Balázs A, Hritz I, Izbéki F, Halász A, Párniczky A, Takács T, Kelemen D, Sarlós P, Hegyi P, Czako L. (2016) ***SPINK1* promoter variants in chronic pancreatitis.** Pancreas 45, 148-153

1 affected

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