

SPINK1

c.195-606G>A

IVS3-606G>A

Variant is in linkage with variants c.56-37T>C, c.87+268A>G, c.101A>G (p.N34S), and c.195-66_65insTTTT

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

18 affected (6 homozygous)

Variant was incorrectly reported as IVS3-604G>A

Functional studies:

Masamune A, Kume K, Takagi Y, Kikuta K, Satoh K, Satoh A, Shimosegawa T. (2007) **N34S mutation in the *SPINK1* gene is not associated with alternative splicing.** Pancreas 34, 423-428

Kereszturi E, Király O, Sahin-Tóth M. (2009) **Minigene analysis of intronic variants in common *SPINK1* haplotypes associated with chronic pancreatitis.** Gut 58, 545-549

Boulling A, Chen JM, Callebaut I, Férec C. (2012) **Is the *SPINK1* p.Asn34Ser missense mutation per se the true culprit within its associated haplotype?** WebmedCentral GENETICS 2012; 3, WMC003084