

SPINK1

c.194+13T>G

IVS3+13T>G

Citations:

Gomez-Lira M, Bonamini D, Castellani C, Unis L, Cavallini G, Assael BM, Pignatti PF. (2003) **Mutations in the *SPINK1* gene in idiopathic pancreatitis Italian patients.** Eur J Hum Genet 11, 543-546

1 affected

Variant was originally reported as IVS3+12T>G; name was corrected by Zou et al. (2016)

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