

SPINK1

c.-7T>G

Citations:

Bernardino AL, Guarita DR, Mott CB, Pedroso MR, Machado MC, Laudanna AA, Tani CM, Almeida FL, Zatz M. (2003) **CFTR, PRSS1 and SPINK1 mutations in the development of pancreatitis in Brazilian patients.** JOP 4, 169-177
5 affected, 8 unaffected

Functional studies:

Boulling A, Witt H, Chandak GR, Masson E, Paliwal S, Bhaskar S, Reddy DN, Cooper DN, Chen JM, Férec C. (2011) **Assessing the pathological relevance of *SPINK1* promoter variants.** Eur J Hum Genet 19, 1066-1073

Derikx MH, Geisz A, Kereszturi É, Sahin-Tóth M. (2015) **Functional significance of *SPINK1* promoter variants in chronic pancreatitis.** Am J Physiol Gastrointest Liver Physiol 308, G779-784