

***SPINK1* c.-215G>T**

Citations:

Chandak GR, Idris MM, Reddy DN, Bhaskar S, Sriram PV, Singh L. (2002) **Mutations in the pancreatic secretory trypsin inhibitor gene (*PSTI/SPINK1*) rather than the cationic trypsinogen gene (*PRSSI*) are significantly associated with tropical calcific pancreatitis.** J Med Genet 39, 347-351

3 affected; all carried p.N34S

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

Same subjects reported as in Chandak et al. (2002)

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