

**CTRC**    **c.746C>T**    **p.P249L**

dbSNP [rs142560329](#)

Citations:

- Rosendahl J, Witt H, Szmola R, Bhatia E, Ózsvári B, Landt O, Schulz HU, Gress TM, Pfützer R, Löhr M, Kovacs P, Blüher M, Stumvoll M, Choudhuri G, Hegyi P, te Morsche RH, Drenth JP, Truninger K, Macek M Jr, Puhl G, Witt U, Schmidt H, Büning C, Ockenga J, Kage A, Groneberg DA, Nickel R, Berg T, Wiedenmann B, Bödeker H, Keim V, Mössner J, Teich N, Sahin-Tóth M. (2008) **Chymotrypsin C (CTRC) variants that diminish activity or secretion are associated with chronic pancreatitis.** [Nat Genet 40, 78-82](#)
- Cho SM, Shin S, Lee KA. (2016) **PRSSI, SPINK1, CFTR, and CTRC pathogenic variants in Korean patients with idiopathic pancreatitis.** *Ann Lab Med* 36, 555-560

Functional studies:

- Beer S, Zhou J, Szabó A, Keiles S, Chandak GR, Witt H, Sahin-Tóth M. (2013) **Comprehensive functional analysis of chymotrypsin C (CTRC) variants reveals distinct loss-of-function mechanisms associated with pancreatitis risk.** [Gut 62, 1616-1624. Epub 2012 September 1.](#)