

**CTRC**    **c.703G>A**    **p.V235I homozygous**

dbSNP [rs140993290](#)

Citations:

- Paliwal S, Bhaskar S, Mani KR, Reddy DN, Rao GV, Singh SP, Thomas V, Chandak GR. (2013) **Comprehensive screening of chymotrypsin C (CTRC) gene in tropical calcific pancreatitis identifies novel variants.** [Gut 62, 1602-1606.](#) Epub 2012 May 12.

Functional studies:

- 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](#)
- 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.