

CTRC **c.760C>T** **p.R254W homozygous**

dbSNP [rs121909293](#)

Citations:

- Rosendahl J, Landt O, Bernadova J, Kovacs P, Teich N, Bödeker H, Keim V, Ruffert C, Mössner J, Kage A, Stumvoll M, Groneberg D, Krüger R, Luck W, Treiber M, Becker M, Witt H. (2013) ***CFTR*, *SPINK1*, *CTRC* and *PRSSI* variants in chronic pancreatitis: is the role of mutated *CFTR* overestimated?** [Gut 62, 582-592. Epub 2012 Mar 17.](#)

Functional studies:

- Rosendahl J, Witt H, Szmola R, Bhatia E, Ózsvári B, Landt O, Schulz HU, Gress TM, Pfützner 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.](#)