

**CTRC**    **c.649G>A**    **p.G217S**

dbSNP [rs202058123](#)

Citations:

**Recommended primary citations**

Note that Rosendahl et al. reported the same subjects in their two publications.

Note that Masson et al. reported the same subjects in their two publications.

- 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](#)
- Masson E, Chen JM, Scotet V, Le Maréchal C, Férec C. (2008) **Association of rare chymotrypsinogen C (CTRC) gene variations in patients with idiopathic chronic pancreatitis.** [Hum Genet 123, 83-91](#)
- 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.](#)
- Masson E, Chen JM, Audrézet MP, Cooper DN, Férec C. (2013) **A conservative assessment of the major genetic causes of idiopathic chronic pancreatitis: Data from a comprehensive analysis of PRSSI, SPINK1, CTRC and CFTR Genes in 253 young French patients.** [PLoS One 8, e73522](#)

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.](#)