

**CTRC**    **c.738\_761del24**    **p.K247\_R254del**

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

Note that Witt et al. (2013) reported overlapping subjects with Rosendahl et al

- 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](#)
- Witt H, Beer S, Rosendahl J, Chen JM, Chandak GR, Masamune A, Bence M, Szmola R, Oracz G, Macek M Jr, Bhatia E, Steigenberger S, Lasher D, Bühler F, Delaporte C, Tebbing J, Ludwig M, Pilsak C, Saum K, Bugert P, Masson E, Paliwal S, Bhaskar S, Sobczynska-Tomaszewska A, Bak D, Balascak I, Choudhuri G, Nageshwar Reddy D, Rao GV, Thomas V, Kume K, Nakano E, Kakuta Y, Shimosegawa T, Durko L, Szabó A, Schnúr A, Hegyi P, Rakonczay Z Jr, Pfützer R, Schneider A, Groneberg DA, Braun M, Schmidt H, Witt U, Friess H, Algül H, Landt O, Schuelke M, Krüger R, Wiedenmann B, Schmidt F, Zimmer KP, Kovacs P, Stumvoll M, Blüher M, Müller T, Janecke A, Teich N, Grützmann R, Schulz HU, Mössner J, Keim V, Löhr M, Férec C, Sahin-Tóth M. (2013) **Variants in CPA1 are strongly associated with early onset chronic pancreatitis.** [Nat Genet 45, 1216-1220](#)
- LaRusch J, Lozano-Leon A, Stello K, Moore A, Muddana V, O'Connell M, Diergaard B, Yadav D, Whitcomb DC. (2015) **The common chymotrypsinogen C (CTRC) variant G60G (C.180T) increases risk of chronic pancreatitis but not recurrent acute pancreatitis in a North American population.** [Clin Transl Gastroenterol 6, e68](#)

- Koziel D, Gluszek S, Kowalik A, Chlopek M, Pieciak L. (2015) **Genetic mutations in *SPINK1*, *CFTR*, *CTRC* genes in acute pancreatitis**. *BMC Gastroenterol* 15, 70  
[Unaffected controls described in Discussion only](#)
- Palermo JJ, Lin TK, Hornung L, Valencia CA, Mathur A, Jackson K, Fei L, Abu-El-Haija M. (2016) **Genophenotypic analysis of pediatric patients with acute recurrent and chronic pancreatitis**. *Pancreas*. 2016 May 19. [Epub ahead of print]

#### 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](#)
- Binker MG, Richards D, Gaisano HY, Cosen-Binker LI. (2015) **ER stress-associated *CTRC* mutants decrease stimulated pancreatic zymogen secretion through SIRT2-mediated microtubule dysregulation**. *Biochem Biophys Res Commun* 463, 329-35