

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

dbSNP [rs140993290](#)

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 subject 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](#)
- Derikx MH, Szmola R, te Morsche RH, Sunderasan S, Chacko A, Drenth JP. (2009) **Tropical calcific pancreatitis and its association with CTRC and SPINK1 (p.N34S) variants.** [Eur J Gastroenterol Hepatol 21, 889-894](#)
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Variant was described at the protein level only
- 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](#)

- Tomaiuolo AC, Sofia VM, Surace C, Majo F, Genovese S, Petrocchi S, Grotta S, Alghisi F, Lucidi V, Angioni A. (2015) **Relationship between *CFTR* and *CTRC* variants and the clinical phenotype in late-onset cystic fibrosis disease with chronic pancreatitis.** *J Mol Diagn* 17, 171-178
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- Gaïtch N, Hubert D, Gameiro C, Burgel PR, Houriez F, Martinez B, Honoré I, Chapron J, Kanaan R, Dusser D, Girodon E, Bienvenu T. (2016) ***CFTR* and/or pancreatitis susceptibility genes mutations as risk factors of pancreatitis in cystic fibrosis patients?** *Pancreatology* 16, 515-522
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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](#)