

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

rs121909293

Citations:

Recommended primary citations

Note that Rosendahl et al. reported overlapping 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](#)
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- Felderbauer P, Karakas E, Fendrich V, Lebert R, Bartsch DK, Bulut K. (2011) **Multifactorial genesis of pancreatitis in primary hyperparathyroidism: evidence for "protective" (PRSS2) and "destructive" (CTRC) genetic factors.** [Exp Clin Endocrinol Diabetes 119, 26-29](#)
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- 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 PRSS1, 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, Balascek 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, Pfützner 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](#)

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Variant is indicated as T>C in Table 2
Unclear if found in affected or unaffected subject; assigned to affected
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See Table 3
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Functional studies:

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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](#)