

CTRC c.514A>G p.K172E

dbSNP [rs34949635](#)

Citations:

Recommended primary citations

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

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- 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](#)
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- Werlin S, Konikoff FM, Halpern Z, Barkay O, Yerushalmi B, Broide E, Santo E, Shamir R, Shaoul R, Shteyer E, Yaakov Y, Cohen M, Kerem E, Ruzzniewski P, Masson E, Férec C, Wilschanski M. (2015) **Genetic and electrophysiological characteristics of recurrent acute pancreatitis.** [J Pediatr Gastroenterol Nutr 60, 675-679](#)
Variant was described at the protein level only
- Sofia VM, Da Sacco L, Surace C, Tomaiuolo AC, Genovese S, Grotta S, Gnazzo M, Petrocchi S, Ciocca L, Alghisi F, Montemitto E, Martemucci L, Elce A, Lucidi V,

Castaldo G, Angioni A. (2016) **Extensive molecular analysis suggested the strong genetic heterogeneity of idiopathic chronic pancreatitis.** Mol Med 2016 May 26. [Epub ahead of print]

Functional studies:

- 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](#)