

**PRSS1 c.486C>T p.D162= p.D162D**

dbSNP [rs6666](#)

Linked with protective variant c.-204C>A and variants c.-408C>T and c.738C>T

#### Citations

- Gorry MC, Ghabbaizedeh D, Furey W, Gates LK Jr, Preston RA, Aston CE, Zhang Y, Ulrich C, Ehrlich GD, Whitcomb DC. (1997) **Mutations in the cationic trypsinogen gene are associated with recurrent acute and chronic pancreatitis.** Gastroenterology 113, 1063-1068
- Teich N, Mössner J, Keim V. (1998) **Mutations of the cationic trypsinogen in hereditary pancreatitis.** Hum Mutat 12, 39-43  
Note that variant was reported as exon-4 variant nt133807C>T
- Nishimori I, Kamakura M, Fujikawa-Adachi K, Morita M, Onishi S, Yokoyama K, Makino I, Ishida H, Yamamoto M, Watanabe S, Ogawa M. (1999) **Mutations in exons 2 and 3 of the cationic trypsinogen gene in Japanese families with hereditary pancreatitis.** Gut 44, 259-263
- Férec C, Raguénès O, Salomon R, Roche C, Bernard JP, Guillot M, Quéré I, Faure C, Mercier B, Audrézet MP, Guillausseau PJ, Dupont C, Munnich A, Bignon JD, Le Bodic L. (1999) **Mutations in the cationic trypsinogen gene and evidence for genetic heterogeneity in hereditary pancreatitis.** J Med Genet 36, 228-232
- O'Reilly DA, Yang BM, Creighton JE, Demaine AG, Kingsnorth AN. (2001) **Mutations of the cationic trypsinogen gene in hereditary and non-hereditary pancreatitis.** Digestion 64, 54-60  
Note that variant was reported as C133807T
- Gomez Lira M, Patuzzo C, Castellani C, Bovo P, Cavallini G, Mastella G, Pignatti PF. (2001) **CFTR and cationic trypsinogen mutations in idiopathic pancreatitis and neonatal hypertrypsinemia.** Pancreatology 1, 538-542  
Note that variant was reported as nt133807C/T
- Tautermann G, Ruebsamen H, Beck M, Dertinger S, Drexel H, Lohse P. (2001) **R116C mutation of cationic trypsinogen in a Turkish family with recurrent pancreatitis illustrates genetic microheterogeneity of hereditary pancreatitis.** Digestion 64, 226-232
- Chandak GR, Idris MM, Reddy DN, Bhaskar S, Sriram PV, Singh L. (2002) **Mutations in the pancreatic secretory trypsin inhibitor gene (*PSTI/SPINK1*) rather than the cationic trypsinogen gene (*PRSS1*) are significantly associated with tropical calcific pancreatitis.** J Med Genet 39, 347-351

- Patuzzo C, Castellani C, Sagramoso C, Gomez-Lira M, Bonamini D, Belpinati F, Dehecchi MC, Assael BM, Pignatti PF. (2003) **Cationic trypsinogen and pancreatic secretory trypsin inhibitor gene mutations in neonatal hypertrypsinaemia.** [Eur J Hum Genet 11, 93-96](#)  
Note that variant was reported as 133807C/T in exon 4. Subjects likely overlap with Gomez Lira, 2001
- Chandak GR, Idris MM, Reddy DN, Mani KR, Bhaskar S, Rao GV, Singh L. (2004) **Absence of PRSS1 mutations and association of SPINK1 trypsin inhibitor mutations in hereditary and non-hereditary chronic pancreatitis.** [Gut 53, 723-728](#)
- Tzetis M, Kaliakatsos M, Fotoulaki M, Papatheodorou A, Doudounakis S, Tsezou A, Makrythanasis P, Kanavakis E, Nousia-Arvanitakis S. (2007) **Contribution of the CFTR gene, the pancreatic secretory trypsin inhibitor gene (SPINK1) and the cationic trypsinogen gene (PRSS1) to the etiology of recurrent pancreatitis.** [Clin Genet 71, 451-457](#)
- Liu QC, Gao F, Ou QS, Zhuang ZH, Lin SR, Yang B, Cheng ZJ. (2008) **Novel mutation and polymorphism of PRSS1 gene in the Chinese patients with hereditary pancreatitis and chronic pancreatitis.** [Chin Med J \(Engl\) 121, 108-111](#)  
Note that article reported variant incorrectly as c.488 C>T, a new polymorphism
- Liu QC, Zhuang ZH, Zeng K, Cheng ZJ, Gao F, Wang ZQ. (2009) **Prevalence of pancreatic diabetes in patients carrying mutations or polymorphisms of the PRSS1 gene in the Han population.** [Diabetes Technol Ther 11, 799-804](#)
- Dai LN, Chen YW, Yan WH, Lu LN, Tao YJ, Cai W. (2016) **Hereditary pancreatitis of 3 Chinese children: Case report and literature review.** *Medicine (Baltimore)*. 95, e4604  
Mutation not described; identified from electropherograms