PRSS1 c.738C>T p.N246= p.N246N

dbSNP rs6667

Linked with protective variant c.-204C>A and variants c.-408C>T and c.486C>T

Citations: Recommended primary citation(s)

- 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 nt134309C>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

- 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 erroneously reported as nt134359C/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

- Bernardino AL, Guarita DR, Mott CB, Pedroso MR, Machado MC, Laudanna AA, Tani CM, Almeida FL, Zatz M. (2003) **CFTR, PRSS1 and SPINK1 mutations in the development of pancreatitis in Brazilian patients.** JOP 4, 169-177

- Patuzzo C, Castellani C, Sagramoso C, Gomez-Lira M, Bonamini D, Belpinati F, Dechecchi 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 134359C/T in exon 5. 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

- Whitcomb DC, LaRusch J, Krasinskas AM, Klei L, Smith JP, Brand RE, Neoptolemos JP, Lerch MM, Tector M, Sandhu BS, Guda NM, Orlichenko L; Alzheimer's Disease Genetics Consortium, Alkaade S, Amann ST, Anderson MA, Baillie J, Banks PA, Conwell D, Coté GA, Cotton PB, DiSario J, Farrer LA, Forsmark CE, Johnstone M, Gardner TB, Gelrud A, Greenhalf W, Haines JL, Hartman DJ, Hawes RA, Lawrence C, Lewis M, Mayerle J, Mayeux R, Melhem NM, Money ME, Muniraj T, Papachristou GI, Pericak-Vance MA, Romagnuolo J, Schellenberg GD, Sherman S, Simon P, Singh VP, Slivka A, Stolz D, Sutton R, Weiss FU, Wilcox CM, Zarnescu NO, Wisniewski SR, O'Connell MR, Kienholz ML, Roeder K, Barmada MM, Yadav D, Devlin B. (2012) **Common genetic variants in the *CLDN2* and *PRSS1-PRSS2* loci alter risk for alcohol-related and sporadic pancreatitis.** Nat Genet 44, 1349-1354

- 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