PRSS1 c.86A>C p.N29T *For functional studies click here*

Citations: Recommended primary citations
Note that some authors reported the same subjects in multiple publications

- Applebaum-Shapiro SE, Finch R, Pfützer RH, Hepp LA, Gates L, Amann S, Martin S, Ulrich CD 2nd, Whitcomb DC. (2001) **Hereditary pancreatitis in North America: the Pittsburgh-Midwest Multi-Center Pancreatic Study Group study.** Pancreatology 1, 439-443

- Pfützer R, Myers E, Applebaum-Shapiro S, Finch R, Ellis I, Neoptolemos J, Kant JA, Whitcomb DC. (2002) **Novel cationic trypsinogen (PRSS1) N29T and R122C mutations cause autosomal dominant hereditary pancreatitis.** Gut 50, 271-272

- Ochi F, Fujii M, Sakai T, Sugano M, Oshiro K, Okabayashi Y, Mita M, Kido Y. (2003) **A case of acute pancreatitis associated with cationic trypsinogen N29T mutation.** Pancreas 27, 199-201

- Howes N, Lerch MM, Greenhalf W, Stocken DD, Ellis I, Simon P, Truninger K, Ammann R, Cavallini G, Charnley RM, Uomo G, Delhaye M, Spicak J, Drumm B, Jansen J, Mountford R, Whitcomb DC, Neoptolemos JP; European Registry of Hereditary Pancreatitis and Pancreatic Cancer (EUROPAC). (2004) **Clinical and genetic characteristics of hereditary pancreatitis in Europe.** Clin Gastroenterol Hepatol 2, 252-261

- Lee YJ, Kim KM, Choi JH, Lee BH, Kim GH, Yoo HW. (2011) **High incidence of PRSS1 and SPINK1 mutations in Korean children with acute recurrent and chronic pancreatitis.** J Pediatr Gastroenterol Nutr 52, 478-481

- Rosendahl J, Landt O, Bernadova J, Kovacs P, Teich N, Bödeker H, Keim V, Ruffert C, Mössner J, Kage A, Stumvoll M, Groneberg D, Krüger R, Luck W, Treiber M, Becker M, Witt H. (2013) ***CFTR*, *SPINK1*, *CTRC* and *PRSS1* variants in chronic pancreatitis: is the role of mutated *CFTR* overestimated?** Gut 62, 582-592. Epub 2012 Mar 17

- Dytz MG, Mendes de Melo J, de Castro Santos O, da Silva Santos ID, Rodacki M, Conceição FL, Ortiga-Carvalho TM. (2015) **Hereditary pancreatitis associated With the N29T mutation of the *PRSS1* gene in a Brazilian family: A case-control study.** Medicine (Baltimore) 94, e1508

Functional studies:

- Sahin-Tóth M. (1999) **Hereditary pancreatitis-associated mutation $Asn^{21} \rightarrow Ile$ stabilizes rat trypsinogen in vitro.** J Biol Chem 274, 29699-29704
Note that studies were performed on rat anionic trypsinogen

- Sahin-Tóth M, Gráf L, Tóth M. (1999) **Trypsinogen stabilization by mutation Arg117→His: a unifying pathomechanism for hereditary pancreatitis?** Biochem Biophys Res Commun 264, 505-508
  Note that studies were performed on rat anionic trypsinogen

- Sahin-Tóth M, Tóth M. (2000) **High-affinity Ca^{2+} binding inhibits autoactivation of rat trypsinogen.** Biochem Biophys Res Commun 275, 668-671
  Note that studies were performed on rat anionic trypsinogen

- Sahin-Tóth M. (2000) **Human cationic trypsinogen. Role of Asn-21 in zymogen activation and implications in hereditary pancreatitis.** J Biol Chem 275, 22750-22755

- Kukor Z, Mayerle J, Krüger B, Tóth M, Steed PM, Halangk W, Lerch MM, Sahin-Tóth M. (2002) **Presence of cathepsin B in the human pancreatic secretory pathway and its role in trypsinogen activation during hereditary pancreatitis.** J Biol Chem 277, 21389-21396

- Kereszturi E, Szmola R, Kukor Z, Simon P, Weiss FU, Lerch MM, Sahin-Tóth M. (2009) **Hereditary pancreatitis caused by mutation-induced misfolding of human cationic trypsinogen: a novel disease mechanism.** Hum Mutat 30, 575-382

- Szabó A, Sahin-Tóth M. (2012) **Increased activation of hereditary pancreatitis-associated human cationic trypsinogen mutants in presence of chymotrypsin C.** J. Biol Chem 287, 20701-20710