

PRSS1 c.68A>G p.K23R

dbSNP [rs111033567](#)

Citations: **Recommended primary citations**

- 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
- Rebours V, Boutron-Ruault MC, Schnee M, Férec C, Le Maréchal C, Hentic O, Maire F, Hammel P, Ruszniewski P, Lévy P. (2009) **The natural history of hereditary pancreatitis: a national series.** Gut 58, 97-103
Note that the same patients were reported as in the citation above
- 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, Ruszniewski P, Masson E, Férec C, Wilschanski M. (2015) **Genetic and electrophysiological characteristics of recurrent acute pancreatitis.** J Pediatr Gastroenterol 60, 675-679
Variant was described at the protein level only

Functional studies:

- Teich N, Ockenga J, Hoffmeister A, Manns M, Mössner J, Keim V. (2000) **Chronic pancreatitis associated with an activation peptide mutation that facilitates trypsin activation.** Gastroenterology 119, 461-465
- Teich N, Bödeker H, Keim V. (2002) **Cathepsin B cleavage of the trypsinogen activation peptide.** BMC Gastroenterol 2, 16
- Chen JM, Kukor Z, Le Maréchal C, Tóth M, Tsakiris L, Raguénès O, Férec C, Sahin-Tóth M. (2003) **Evolution of trypsinogen activation peptides.** Mol Biol Evol 20, 1767-1777
- Gaiser S, Ahler A, Gundling F, Kruse ML, Savkovic V, Selig L, Teich N, Tomasini R, Dagorn JC, Mössner J, Keim V, Bödeker H. (2005) **Expression of mutated cationic trypsinogen reduces cellular viability in AR4-2J cells.** Biochem Biophys Res Commun 334, 721-728
- Mikhailova AG, Likhareva VV, Teich N, Rumsh LD. (2007) **The ways of realization of high specificity and efficiency of enteropeptidase.** Protein Pept Lett 14, 227-232

- Kereszturi E, Sahin-Tóth M. (2009) **Intracellular autoactivation of human cationic trypsinogen mutants causes reduced trypsinogen secretion and acinar cell death.** J Biol Chem 284, 33392-33399
- Geisz A, Hegyi P, Sahin-Tóth M. (2013) **Robust autoactivation, chymotrypsin C independence and diminished secretion define a subset of hereditary pancreatitis-associated cationic trypsinogen mutants.** FEBS J 280, 2888-2899