

***PRSSI* c.416G>T p.C139F**

Citations: Recommended primary citations

- Teich N, Bauer N, Mössner J, Keim V. (2002) **Mutational screening of patients with nonalcoholic chronic pancreatitis: identification of further trypsinogen variants.** Am J Gastroenterol 97, 341-346
- Sultan M, Werlin S, Venkatasubramani N. (2012) **Genetic prevalence and characteristics in children with recurrent pancreatitis.** J Pediatr Gastroenterol Nutr 54, 645-650
Note that variant was reported as p.C139F, VS4-, 24C>T 3. Meaning of unconventional nomenclature is unclear; possibly subject carried a variant in intron-4 as well
- Chang MC, Jan IS, Liang PC, Jeng YM, Yang CY, Tien YW, Wong JM, Chang YT. (2014) **Human cationic trypsinogen but not serine peptidase inhibitor, Kazal type 1 variants increase the risk of type 1 autoimmune pancreatitis.** J Gastroenterol Hepatol 29, 2038-2042
- Sofia VM, Da Sacco L, Surace C, Tomaiuolo AC, Genovese S, Grotta S, Gnazzo M, Petrocchi S, Ciocca L, Alghisi F, Montemitro 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:

- Schnúr A, Beer S, Witt H, Hegyi P, Sahin-Tóth M. (2014) **Functional effects of 13 rare *PRSSI* variants presumed to cause chronic pancreatitis.** Gut 63, 337–343. Epub 2013 March 1