PRSS1 c.235G>A p.E79K

dbSNP rs111033564

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

- Chen JM, Piepoli Bis A, Le Bodic L, Ruszniewski P, Robaszkiewicz M, Deprez PH, Raguénès O, Quéré I, Andriulli A, Férec C. (2001) **Mutational screening of the cationic trypsinogen gene in a large cohort of subjects with idiopathic chronic pancreatitis.** Clin Genet 59, 189-193

- 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

- Teich N, Le Maréchal C, Kukor Z, Caca K, Witzigmann H, Chen JM, Tóth M, Mössner J, Keim V, Férec C, Sahin-Tóth M. (2004) **Interaction between trypsinogen isoforms in genetically determined pancreatitis: mutation E79K in cationic trypsin (PRSS1) causes increased transactivation of anionic trypsinogen (PRSS2).** Hum Mutat 23, 22-31

- Keiles S, Kammesheidt A. (2006) **Identification of *CFTR*, *PRSS1*, and *SPINK1* mutations in 381 patients with pancreatitis.** Pancreas 33, 221-227

- 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

- Sultan M, Werlin S, Venkatasubramani N. (2012) **Genetic prevalence and characteristics in children with recurrent pancreatitis.** J Pediatr Gastroenterol Nutr 54, 645-650

- Hamoir C, Pepermans X, Piessevaux H, Jouret-Mourin A, Weynand B, Habyalimana JB, Leal T, Geubel A, Gigot JF, Deprez PH. (2013) **Clinical and morphological characteristics of sporadic genetically determined pancreatitis as compared to idiopathic pancreatitis: higher risk of pancreatic cancer in CFTR variants.** Digestion 87, 229-239

- Oracz G, Kolodziejczyk E, Sobczynska-Tomaszewska A, Wejnarska K, Dadalski M, Grabarczyk AM, Kierkus J, Woynarowski M, Wertheim-Tysarowska K, Ryzko J, Bal J, Rygiel AM. (2016) **The clinical course of hereditary pancreatitis in children - A comprehensive analysis of 41 cases.** Pancreatology 2016 Apr 26. [Epub ahead of print]

Functional studies:

- Teich N, Le Maréchal C, Kukor Z, Caca K, Witzigmann H, Chen JM, Tóth M, Mössner J, Keim V, Férec C, Sahin-Tóth M. (2004) **Interaction between trypsinogen isoforms in genetically determined pancreatitis: mutation E79K in cationic trypsin (PRSS1) causes increased transactivation of anionic trypsinogen (PRSS2).** Hum Mutat 23, 22-31

- 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, 575-582