

***PRSS1* c.410C>T p.T137M**

Citations: Recommended primary citations

- Keiles S, Kammesheidt A. (2006) **Identification of *CFTR*, *PRSS1*, and *SPINK1* mutations in 381 patients with pancreatitis.** *Pancreas* 33, 221-227
- Chang YT, Wei SC, L PC, Tien YW, Jan IS, Su YN, Wong JM, Chang MC. (2009) **Association and differential role of *PRSS1* and *SPINK1* mutation in early-onset and late-onset idiopathic chronic pancreatitis in Chinese subjects.** *Gut* 58, 885
Note that variant was erroneously reported at the nucleotide level as c.137C>T
- Zeng K, Liu QC, Lin JH, Lin XH, Zhuang ZH, Gao F, Ou QS. (2011) **Novel mutations of *PRSS1* gene in patients with pancreatic cancer among Han population.** *Chin Med J (Engl)* 124, 2065-2067
Variant was found in a subject with pancreatic cancer. Note that subject also carried the c.403A>G (p.T135A) variant; possibly on a different allele
Note that variant was erroneously reported at the nucleotide level as c.416 C>T
- 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
Variant was incorrectly reported at the nucleotide level as c137C>T
- Yi Q, Dong F, Lin L, Liu Q, Chen S, Gao F, He Q. (2016) ***PRSS1* mutations and the proteinase/antiproteinase imbalance in the pathogenesis of pancreatic cancer.** *Tumour Biol* 37, 5805-5810
Variant was found in five subjects with pancreatic cancer
- Zhiping W, Quwen L, Hai Z, Jian Z, Peiyi G. (2016) **Application of molecular imaging combined with genetic screening in diagnosing MELAS, diabetes and recurrent pancreatitis.** *Folia Neuropathol* 54, 66-71
Unclear how many family members were positive for the *PRSS1* mutation

Functional studies:

- Schnúr A, Beer S, Witt H, Hegyi P, Sahin-Tóth M. (2014) **Functional effects of 13 rare *PRSS1* variants presumed to cause chronic pancreatitis.** *Gut* 63, 337–343