

SPINK1 **c.150T>G** **p.D50E**

Citations:

Pfützer RH, Barmada MM, Brunskill AP, Finch R, Hart PS, Neoptolemos J, Furey WF, Whitcomb DC. (2000) **SPINK1/PSTI polymorphisms act as disease modifiers in familial and idiopathic chronic pancreatitis.** *Gastroenterology* 119, 615-623
I affected; also carried p.N34S

Functional studies:

Király O, Wartmann T, Sahin-Tóth M. (2007) **Missense mutations in pancreatic secretory trypsin inhibitor (SPINK1) cause intracellular retention and degradation.** *Gut* 56, 1433-1438

Boulling A, Le Maréchal C, Trouvé P, Raguénès O, Chen JM, Férec C. (2007) **Functional analysis of pancreatitis-associated missense mutations in the pancreatic secretory trypsin inhibitor (*SPINK1*) gene.** *Eur J Hum Genet* 15, 936-942

Mokmak W, Chunsrivirod S, Assawamakin A, Choowongkamon K, Tongsimma S. (2013) **Molecular dynamics simulations reveal structural instability of human trypsin inhibitor upon D50E and Y54H mutations.** *J Mol Model* 19, 521-528