

***SPINK1* c.160T>C p.Y54H**

Citations:

Schneider A, Suman A, Rossi L, Barmada MM, Beglinger C, Parvin S, Sattar S, Ali L, Khan AK, Gyr N, Whitcomb DC. (2002) ***SPINK1/PSTI* mutations are associated with tropical pancreatitis and type II diabetes mellitus in Bangladesh.** Gastroenterology 123, 1026-1030
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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