SPINK1 c.194G>A p.R65Q

rs141634296

Citations:

Ockenga J, Dörk T, Stuhrmann M. (2001) **Low prevalence of *SPINK1* gene mutations in adult patients with chronic idiopathic pancreatitis.** J Med Genet 38, 243-244
1 family with 1 affected and 2 unaffected; all carried *CFTR* p.Y1092X

Keiles S, Kammesheidt A. (2006) **Identification of *CFTR, PRSS1,* and *SPINK1* mutations in 381 patients with pancreatitis.** Pancreas 33, 221-227
1 affected; also carried *PRSS1* p.R122C
Variant was described at the protein level only

Rosendahl J, Landt O, Bernadova J, Kovacs P, Teich N, Bödeker H, Keim V, Ruffert C, Mössner J, Kage A, Stumvoll M, Groneberg D, Krüger R, Luck W, Treiber M, Becker M, Witt H. (2013) ***CFTR, SPINK1, CTRC* and *PRSS1* variants in chronic pancreatitis: is the role of mutated *CFTR* overestimated?** Gut 62, 582-592
1 affected

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
1 affected
Variant was erroneously described at the protein level as p.R65P

Masson E, Chen JM, Audrézet MP, Cooper DN, Férec C. (2013) **A conservative assessment of the major genetic causes of idiopathic chronic pancreatitis: data from a comprehensive analysis of *PRSS1, SPINK1, CTRC* and *CFTR* genes in 253 young French patients.** PLoS One 8, e73522
1 affected (homozygous); also carried p.N34S (homozygous)

Tremblay K, Dubois-Bouchard C, Brisson D, Gaudet D. (2014) **Association of *CTRC* and *SPINK1* gene variants with recurrent hospitalizations for pancreatitis or acute abdominal pain in lipoprotein lipase deficiency.** Front Genet 5, 90
Unclear if found in 1 affected or unaffected subject; counted as 1 affected
Variant is indicated as A>G in Table 2

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

Beer S, Sahin-Tóth M. (2014) **Exonic variants affecting pre-mRNA splicing add to genetic burden in chronic pancreatitis.** Gut 63, 860-861