

SPINK1 **c.203A>G** **p.Q68R**

Citations:

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

I affected; also carried *PRSSI* p.N29I

Variant was described at the protein level only

Functional studies:

Boulling A, Keiles S, Masson E, Chen JM, Férec C. (2012) **Functional analysis of eight missense mutations in the *SPINK1* gene.** *Pancreas* 41, 329-330