

***SPINK1***

**c.190A>G**

**p.N64D**

Citations:

Sultan M, Werlin S, Venkatasubramani N. (2012) **Genetic prevalence and characteristics in children with recurrent pancreatitis.** J Pediatr Gastroenterol Nutr 54, 645-650

2 affected

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

1 affected; likely the same as in Sultan et al. (2012); both studies used data from Ambry Genetics; did not count

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