

SPINK1

c.123G>C

p.K41N

Citations:

Terlizzi V, De Gregorio F, Sepe A, Amato N, Arduino C, Casale A, Majo F, Tomaiuolo R, Castaldo G, Raia V. (2013) **Brand new SPINK1 and CFTR mutations in a child with acute recurrent pancreatitis: a case report.** *Minerva Pediatr* 65, 669-672

1 family with 1 affected child and 1 unaffected parent; child also carried heterozygous *CFTR* p.S1235R inherited from other parent