SPINK1 c.27delC p.S10VfsX5

rs193922659

Citations:

Le Maréchal C, Chen JM, Le Gall C, Plessis G, Chipponi J, Chuzhanova NA, Raguénès O, Férec C. (2004) **Two novel severe mutations in the pancreatic secretory trypsin inhibitor gene (*SPINK1*) cause familial and/or hereditary pancreatitis.** Hum Mutat 23, 205
2 families with 5 affected and 5 unaffected

LaRusch J, Barmada MM, Solomon S, Whitcomb DC. (2012) **Whole exome sequencing identifies multiple, complex etiologies in an idiopathic hereditary pancreatitis kindred.** JOP 13, 258-262
Text indicates 14 mutation carriers in the family; pedigree shows 12; the sample IDs sequenced also contradict between text and figure
1 family with 5 affected (including 1 obligate carrier) and 7 unaffected; plus 1 unrelated affected

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

Masson E, Chen JM, Férec C. (2015) **Report of 2 *CTRC* intronic mutations associated with acute or chronic pancreatitis and delineation of their pathogenic molecular mechanisms.** Pancreas 44, 999-1001
1 affected