

SPINK1

c.194+90A>T

IVS3+90A>T

Variant is in linkage with c.36G>C (p.L12F) African variant

Citations:

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

8 affected, 1 also carried c.-22C>T, 1 also carried *CFTR* 296+28A>G, 1 also carried *CFTR* 4375-20A>G, 2 also carried *PRSSI* p.R122H

Text described 7 affected; tables indicated 8

Functional studies:

Zou WB, Boulling A, Masson E, Cooper DN, Liao Z, Li ZS, Férec C, Chen JM. (2016) **Clarifying the clinical relevance of *SPINK1* intronic variants in chronic pancreatitis.** *Gut* 65, 884-886