

## ***SPINK1***

**c.-2C>A**

### Citations:

Audrézet MP, Chen JM, Le Maréchal C, Ruszniewski P, Robaszkiewicz M, Raguénès O, Quéré I, Scotet V, Férec C. (2002) **Determination of the relative contribution of three genes-the cystic fibrosis transmembrane conductance regulator gene, the cationic trypsinogen gene, and the pancreatic secretory trypsin inhibitor gene-to the etiology of idiopathic chronic pancreatitis.** Eur J Hum Genet 10, 100-106

1 affected; also carried *CFTR* p.W1282X

### Functional studies:

Derikx MH, Geisz A, Kereszturi É, Sahin-Tóth M. (2015) **Functional significance of *SPINK1* promoter variants in chronic pancreatitis.** Am J Physiol Gastrointest Liver Physiol 308, G779-784