Mutational Analysis of ATP8B1 in Patients with Chronic Pancreatitis.

Abstract:
Mutations in genes encoding cationic trypsinogen (PRSS1), pancreatic secretory trypsin inhibitor (SPINK1) and chymotrypsinogen C (CTRC) are associated with chronic pancreatitis. However, in many patients with a familial chronic pancreatitis pattern suggesting a genetic cause, no mutations in either of these genes can be found, indicating that other, still unknown, associated genes exist. In this respect ATP8B1 is an interesting candidate due to its strong expression in the pancreas, its supposed general function in membrane organization and the higher incidence of pancreatitis in patients with ATP8B1 deficiency. We analyzed all 27 ATP8B1 coding exons and adjacent non-coding sequences of 507 chronic pancreatitis patients by direct sequencing. Exons that harbored possible relevant variations were subsequently sequenced in 1,027 healthy controls. In the exonic regions, 5 novel non-synonymous alterations were detected as well as 14 previously described alterations of which some were associated with ATP8B1 deficiency. However, allele frequencies for any of these variations did not significantly differ between patients and controls. Furthermore, several non-synonymous variants were exclusively detected in control subjects and multiple variants in the non-coding sequence were identified with similar frequencies in both groups. We did not find an association between heterozygous ATP8B1
variants and chronic pancreatitis in our cohort of patients with hereditary and idiopathic chronic pancreatitis.

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