A common SLC26A6 haplotype is not associated with chronic pancreatitis.

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Background: Cystic Fibrosis Transmembrane Conductance Regulator (CFTR) mutations are established risk factors for chronic pancreatitis (CP). CFTR variants increase disease risk by causing impairment of pancreatic ductal bicarbonate secretion. However, the role of genetic variations in the bicarbonate secreting SLC26A6 anion transporter has remained largely unexplored so far.

Aims: Our aim was to investigate the role of the SLC26A6 gene in CP.

Patients & methods: 96 subjects with CP (cases) and 99 subjects with no pancreatic disease (controls) were recruited from the Hungarian National Pancreas Registry. In a discovery cohort of 30 idiopathic CP cases the entire SLC26A6 coding sequence, including 21 exons and the exon-intron boundaries were amplified and sequenced. Further genotyping of p.V206M and p.P397P mutations in CP and controls was carried out by RFLP.

Results: Sequencing analysis of the discovery cohort revealed four common mutations: intronic mutations c.23+71_23+103del, c.183-4C>A and c.1134+32C>A; and exonic missense mutation p.V206M. These four mutations were found in linkage disequilibrium indicating a conserved haplotype. We found this haplotype in 18 heterozygous and 2 homozygous cases, and in 24 heterozygous and 2 homozygous controls (allele frequency 11.4% and 14.1% respectively). A synonymous mutation p.P397P was also detected in a single case.

Conclusion: We found a novel, common haplotype in the SLC26A6 gene, which did not show association with CP. Supported by TÁMOP and OTKA.