LACK OF *PRSS1* AND *SPINK1* POLYMORPHISMS IN SERBIAN ACUTE PANCREATITIS PATIENTS

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ODSUSTVO *PRSS1* I *SPINK1* POLIMORFIZMA KOD SRPSKIH PACIJENATA OBOLELIH OD AKUTNOG PANKREATITISA

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ABSTRACT

Acute pancreatitis represents an acute nonbacterial inflammation of the pancreas caused by a premature and ectopic activation of pancreatic digestive enzymes. Two of the most important genes in pancreatic autodigestion, PRSS1 and SPINK1, were implicated in the earliest discoveries of the genetic background of pancreatitis. However, the distribution of their variations displays interethnic variability, which could significantly affect the magnitude of their proposed effects on this disease worldwide. The aim of the present study was to investigate the distribution of the most important functional variations of PRSS1 (86A>T and 365G>A) and SPINK1 (101A>G), and their influence on the clinical course of acute pancreatitis in Serbian patients. The study enrolled 81 subjects, the severity of disease course was determined using the Atlanta Classification system, and the genotyping was conducted using a PCR-RFLP method. PRSS1 86A>T and 365G>A SNPs were not observed in the study population, while SPINK1 101A>G was present with the frequency of 0.62% (95% CI: 0.00, 3.83%). Due to extremely low frequencies or absences of examined variations, the proposed effect of these SNPs on the severity of acute pancreatitis could not be confirmed. The results do not support routine genotyping of either PRSS1 or SPINK1 in Serbs.

Keywords: *PRSS1, SPINK1, genetic polymorphism, acute pancreatitis, Serbian*

SAŽETAK

Akutni pankreatitis predstavlja akutno neinfektivno zapaljenje pankreasa, prouzrokovano prevremenom i ektopičnom aktivacijom pankreasnih digestivnih enzima. Enzimi PRSS1 and SPINK1, koji igraju neke od najvažnijih uloga u pankreasnoj autodigestiji, prvi su otkriveni faktori genetske predispozicije za nastanak pankreatitisa. Ipak, zastupljenost njihovih genetskih varijacija varira u zavisnosti od etničke pripadnosti, što u velikoj meri utiče na značaj njihovog učešća u ovoj bolesti širom sveta. Cilj ove studije bio je da ispita distribuciju najznačajnijih funkcionalnih varijacija gena PRSS1 (86A>T i 365G>A) i SPINK1 (101A>G), kao i njihov uticaj na kliničku sliku bolesti, kod Srba obolelih od akutnog pankreatitisa. Istraživanje je uključilo 81 ispitanika, težina bolesti određivana je uz korišćenje Atlanta klasifikacionog sistema, a genotipizacija je sprovedena pomoću PCR-RFLP metode. PRSS1 polimorfizmi 86A>T i 365G>A SNPs nisu detektovani u ispitivanoj populaciji, dok je učestalost SPINK1 101A>G varijacije iznosila 0,62% (95% IP: 0,00; 3,83%). Obzirom na ekstremno nisku učestalost ili potpuno odsustvo ispitivanih varijacija, njihov efekat na težinu i tok akutnog pankreatitisa nije mogao biti potvrđen. Rezultati istraživanja ne preporučuju rutinsku genotipizaciju PRSS1 i SPINK1 kod Srba.

Ključne reči: *PRSS1, SPINK1, genetski polimorfizam, akutni pankreatitis, srpski*

ABBREVIATIONS

PRSS1 - cationic trypsinogen (protease serine type 1);
SPINK1 - pancreatic secretory trypsin inhibitor (serine protease inhibitor Kazal type 1);
SNP - Single nucleotide polymorphism;
EDTA - ethylene diamine tetracetic acid;
PCR - Polymerase chain reaction;
PCR-RFLP - Polymerase chain reaction-restriction fragment length polymorphism

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