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### CANCER SCIENCE AND TARGETED THERAPIES

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#### GG genotype of the PNPLA3 RS738409 polymorphisms is associated with NASH in Uzbek population

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**Aim:** The purpose of our research is to investigate the association between a polymorphic variant of PNPLA3 gene (rs738409) and susceptibility to non-alcoholic fatty liver disease (NAFLD) in Uzbekistan.

Materials and Methods: In this case-control study, 73 patients a mean age of 55.1 diagnosed with NAFLD (48 patients with simple steatosis and 25 patients with non-alcoholic steatohepatitis (NASH)) and the age, gender and ethnically matched controls (n=37) were recruited. The diagnosis of NAFLD was verified on the basis of anamnesis, clinical and laboratory tests, and liver ultrasound. Genomic DNA was isolated and SNP genotyping was performed by using the polymerase chain reaction with specific primers followed by restriction fragment length polymorphism analysis.

Result: Our result showed significant association between GG genotype of the PNPLA3 rs738409 polymorphisms and NAFLD (p=0.03,OR = 2.99; 95% CI 1.21–7.42 for the additive model, Cochran-Armitage trend test; p=0.02,OR = 2.99; 95% CI 1.21–7.42 for the recessive model, Pearson's  $\chi 2$  test). Genotype frequencies of PNPLA3 rs738409 polymorphisms in a subset of patients with simple steatosis and NASH compare to the control group. Comparative analysis of resulting genotypes showed a slight increase of CG and GG genotypes among patient with simple steatosis, then among subjects of the control group, but this did not reach statistical significance. However, statistical analysis of genotype distribution between patients with NASH and controls showed a significant association between GG genotype and NASH assuming an additive model (p<0,0001, Cochran-Armitage trend test) and recessive model (p<0,0001, Pearson's  $\chi 2$  test).

**Conclusion:** The present study, we confirm the association of PNPLA3 rs738409 GG genotype with susceptibility to NAFLD. After stratification into the two main subtypes of NAFLD, the risk genotype GG was found to be significantly associated with susceptibility to NASH. We also found that the GG genotype is not associated with simple steatosis in Uzbek population.

#### **Biography**

Guzal Sobirova has been working in the RSSPCTR since the 2002 year. Since 2005 until current time Dr. Sobirova has been holding the position of Chief of Balneology Department. She is also Senior Scientific Researcher of Gastroenterology Department. She involves in the educational process for clinical residents and masters of Tashkent Medical Academy. She possesses 198 scientific works. She regularly takes part in Scientific Practical Conferences and widely introduces and applies gained knowledge in the departments of our Center and other patient care clinic of the Republic of Uzbekistan. Now focus of her interest is a genetic predisposition to gastroenterological disease with propose of the organization of effective preventive measures and medical interventions.

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