

# Role of Autophagy Related Gene 5 (ATG5) Single Nucleotide Polymorphism rs2245214 (C/G) with HBV Susceptibility in North Indian Population

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**Abstract**—Hepatitis B is fatal liver infection caused by Hepatitis B Virus (HBV) that creates acute and chronic disease conditions that may eventually lead to liver cirrhosis and hepatocellular carcinoma (HCC) and cause death. Globally, 240 million people are chronically infected with hepatitis B and 40 million HBV carriers are in India itself. Host genetic factors influence HBV acquisition and play very important role in determining the progression and outcome of HBV infection. The process of autophagy is believed to be utilized by the Hepatitis B virus for its own replication. Autophagy related gene ATG5 is a key autophagy regulator that initiates the formation of autophagosome; a crucial stage of the autophagy process. ATG5 expression levels have also found to be increased in HBV infected patients. Polymorphisms in ATG5 have also been linked to the susceptibility of various diseases in human. Moreover, the SNP rs2245214 in ATG5 is found to be associated with Thyroid carcinoma and Paget disease of bone (PDB). Therefore, in this study we will investigate the role of ATG5 gene polymorphism rs2245214 (C/G) in HBV infection. We have successfully optimized the polymerase chain reaction restriction fragment length polymorphism (PCR-RFLP) conditions using mutated primers containing desired restriction site for genotyping the target SNP. We have genotyped 200 healthy control samples by PCR-RFLP. Genotypic frequencies in healthy individuals were obtained as GG=7.5 % (n=15), GC=46.5% (n=93) and CC=46% (n=92) and allele frequencies were G=30.75% and C=69.25%. Further, we would genotype HBV infected samples to find out any association of this SNP rs2245214 with HBV susceptibility in North Indian population.