



Genetic Variation in Cyclin-Dependent Kinase Inhibitor 2A Associated with Increased Pancreatic Cancer Risk

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ABSTRACT

Introduction: Pancreatic ductal adenocarcinoma (PDAC) is associated with an inferior prognosis. Therefore, a focus has been on identifying new biomarkers for early diagnosis and predicting patient survival. Recent studies on the entire human genome have pinpointed prevalent genetic alterations on chromosome 9p21 that heighten the vulnerability to cancer. This study aimed to examine the potential partnership of a genetic variant, rs1333049, in CDKN2A in patients diagnosed with pancreatic cancer. Moreover, genome-wide RNA and microRNA sequencing, bioinformatics, and machine learning approaches were undertaken to identify differentially expressed genes (DEGs), followed by validation in an additional PDAC patient cohort.

Methods and Materials: The process involved extracting DNA samples from these individuals and utilizing TaqMan real-time PCR for genotyping and gene expression analysis. Logistic regression was employed to evaluate the relationship between risk and genotypes, and all prognostic factors of significance identified in the univariate evaluation were incorporated into the multivariate evaluation.

Results: Compared to the patients with PDAC, the control group had a decreased occurrence of a TT genotype at the rs1333049 locus. The latter group of cases with a recessive genetic pattern (GG vs. GC+ CC) showed enhanced susceptibility to promoting PDAC (OR = 1.7; $p = 0.04$).

Conclusion and Discussion: Our findings indicate that genetic variation in CDKN2A was linked to the susceptibility of extending PDAC, suggesting the need for additional research in a broader, multi-center context to approve the possible significance of this gene as a novel indicator for the stratification of PDAC.

Keywords: Adenocarcinoma, Genes, Pancreatic neoplasms