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Degree PhD Title of Thesis

Gene polymorphism in non-alcoholic fatty liver disease in Egyptians

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ABSTRACT

Background: Liver diseases are emerging global health issues, non-alcoholic fatty liver disease (NAFLD) represents the most prevalent severe liver disease worldwide in the 21st century. Besides environmental factors, genetic predisposition triggers NAFLD development.

<u>Aim:</u> This study investigated the relationship of Resistin (rs1862513) gene polymorphism and NAFLD and hepatic fibrosis in NAFLD Egyptian patients.

<u>Methodology:</u> The study was performed on 126 subjects as 63 healthy control subjects and 63 NAFLD patients. Genotyping of resistin gene single nucleotide polymorphism rs1862513 for NAFLD patients and healthy controls were done using Taqman genotyping assay.

Results: The biochemical parameters in NAFLD group showed significantly elevated WBCs count; NFS; ALT; AST; TG; direct bilirubin; FBS; serum insulin and HOMA-IR when compared with control and showed significant decrease in both albumin level and PLT count. There were no significant differences regarding genotypic and allelic frequencies among NAFLD subjects and control individuals. Fibrosis progression estimated by NFS may be accompanied with higher age, PLTs, WBCs count, Hb and TG significantly decreased while INR, AST, total and direct bilirubin, FBS and HOMA-IR increased in correlation with level of fibrosis. Genotype frequencies differed significantly among the different fibrosis stages groups. CG+GG genotype carriers showed significantly higher age and ALT while lower WBCs count with fibrosis progression.

<u>Conclusion:</u> This study provided evidence about absence of association among RETN -420C>G SNP and NAFLD susceptibility within Egyptian population, but

proved association with increased NAFLD associated fibrosis risk. G allele harboring patients reported significantly higher fibrosis score, AST levels, total bilirubin and IR whereas lower Alb than CC wild type bearing patients. Moreover, G allele associations with higher age and ALT activities whereas lower WBCs count with fibrosis progression were indicated.