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Cholesterol 7- Alpha Hydroxylase Gene *(CYP7A1)* Promoter Polymorphism rs3808607 as a Risk Factor for Pulmonary Tuberculosis in Egypt

Short running head: CYP7A1 gene polymorphism in pulmonary tuberculosis

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Abstract:

Background: A major tuberculosis susceptibility locus has been identified by genome wide association study on chromosomal region 8q12-q13. The *CYP7A1* gene coding for cholesterol 7a-hydroxylase enzyme is located in this locus, this enzyme is involved in cholesterol catabolism.

Objective: The aim of the study is to investigate C/A polymorphism rs3808607 at *CYP7A1* gene as a risk factor for pulmonary tuberculosis in Egyptian subjects.

Subjects & Methods: A total number of 180 subjects divided into two groups: group I included 100 newly diagnosed sputum positive pulmonary tuberculosis patients and group II included 80 subjects served as controls. Sputum samples were stained by Ziehl Neelsen method. Determination of serum lipid profile was done. Genotyping of the *CYP7A1* SNP rs3808607 was analyzed by real time PCR technique.

Results: Serum lipid profile was significantly decreased in group I when compared to controls ($p<0.05$). The highest frequency of (AA) genotype was detected in pulmonary tuberculosis patients (OR = 28, 95% CI), while (CC) genotype was the most frequent in controls ($p<0.05$). The allele A was more frequent in group I with increased risk of pulmonary tuberculosis in Egyptian subjects by 7.7 folds than controls while, allele C was dominant in group II ($p<0.05$).