البحث الرابع وعنوانه:

Tumor necrosis factor (TNF)-α- 308 G/A gene polymorphism (rs1800629) in Egyptian patients with alopecia areata and vitiligo, a laboratory and in silico analysis

عامل النخر الورم (TNF) -α- 308 G / A تعدد الأشكال الجيني (rs1800629) في مرضى الثعلبة البقعية والبهاق المصريين ، تحليل معملى وسيليكو

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Purpose and methods: Several single-nucleotide polymorphisms (SNPs) in the promoter region of the TNF-α gene can cause variations in the gene regulatory sites and act as risk factors for some autoimmune disorders as alopecia areata (AA) and vitiligo. This study aimed to detect the serum TNF-α (sTNF) level (by ELISA) and the rs1800629 (by real-time PCR) among AA and vitiligo Egyptian patients and to determine their relation with disease duration and severity. In silico analysis of this SNP to study the molecular regulation of the mutant genotypes was also done.

Results: In AA patients, no risk was associated with the mutant genotypes vs. the normal genotype, or with A allele vs. G allele. The risk of vitiligo was significantly higher with the G/A and A/A genotypes compared with HCs ($p = 0.011$). Similarly, a significantly increased risk was noted in patients with A allele vs. G allele ($p<0.0001$). In AA and vitiligo patients, a significant increase in sTNF-α levels was noted in the mutant G/A genotypes vs. the normal G/G genotype ($p<0.0001$) and in the A allele vs the G allele ($p<0.0001$). According to the in silico analysis, this SNP could mainly affect the SP1 transcription factor binding site with subsequent effect on TNF-α expression.

Conclusion: According to results of the laboratory and the in silico study, the mutant TNF-α (308) genotypes were risk factors that conferred susceptibility to vitiligo among Egyptian patients but had no effect on the susceptibility to AA.