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Angiotensin converting enzyme gene polymorphism in patients with systemic lupus erythematosus

Thesis Submitted for the fulfillment of the Master degree in
Clinical and Chemical Pathology

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Abstract

Back ground:

Systemic lupus erythematosus is a chronic multisystemic autoimmune inflammatory disease, polygenic inheritance and many environmental factors play a role in its development. ACE gene insertion / deletion (I/D) polymorphism within intron 16 has been shown to encode the ACE enzyme which is responsible for the different clinical manifestations of SLE .

Method:

This study was conducted on 50 SLE patients diagnosed according to the ACR criteria and 29 apparently healthy volunteers as control group , all subjects were subjected to full history taking, clinical examination , Radiological and laboratory investigation (CBC, ESR serum urea and creatinine 24 hours urinary protein) and genetic study of the ACE gene I/D using the PCR method.

Results:

The frequency of DD genotype was statistically significantly increased in SLE patients compared to control group , $p = 0,009$, and also the DD genotype was associated with higher serum ACE level when compared to DI and II genotypes On comparing the frequency of DD and DI genotypes between SLE patients with and without vasculitis and those with and without nephritis, a statistically significant increase was observed in DD genotype.

Conclusion:

The associations observed in the current work highlight the potentially important role of ACE genotype and subsequently serum ACE in the pathophysiology of SLE.

Key Words:

ACE, SLE , lupus nephritis ,vasculitis , I/D polymorphism.

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List of abbreviations

<i>Abbreviation</i>	
ACE	Angiotensin converting enzyme
ACR	American College of Rheumatology
ANAs	Antinuclear antibodies
Anti- Sm	Anti Smith
AP-1	Activator protein 1
APCs	Antigen presenting cells
ARBs	Angiotensin Receptor Blockers
ATI	Angiotensin I receptors
BLK	B lymphocyte Kinase
C receptor	Complement receptors
CBC	Complete blood count
CDKN1A	Cyclin dependent kinase inhibitor 1
CI	Confidence interval
CMV	Cytomegalo virus
CNS	Central nervous system
CVD	Cardiovascular disease
DMARDs	Disease modifying antirheumatic drugs
dNTPs	Deoxynucleotide triphosphate
Ds DNA	Double stranded deoxynucleic acid
EBV	Epstein-Barr virus