

## Association study of TLR9 gene polymorphism in Egyptian patients with systemic lupus erythematosus and lupus nephritis

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### Abstract

Alterations of the innate immune response is critical in the development of autoimmunity through self-antigens recognition, processing and presentation to initiate an adaptive immune response towards self-antigens. Toll-Like Receptor 9 (TLR9) is an innate immune receptor expressed on the endosomes of antigen presenting cells, and it recognizes microbial DNA. Hence, altered expression of TLR9 can be detrimental in recognizing self-DNA, triggering autoimmune conditions such as Systemic Lupus Erythematosus (SLE) for which the positivity for ANA and anti dsDNA is a hallmark for diagnosis. Although there is a growing interest in the research for using TLR9 agonists for immune stimulation against cancers, the research interest is relatively slim regarding ameliorating autoimmune diseases and glomerulonephritis through targeting TLR9. Our aim was to assess the association between SLE and Lupus Nephritis (LN) with TLR9 rs352140 gene polymorphism. The study was conducted on a group of Egyptian individuals at Mansoura University Hospital, comprising 38 SLE patients without nephritis, 62 LN patients and 100 healthy individuals as a control group.

**Methodology and results:** An informed consent was taken from the subjects before peripheral blood sample was taken. Genomic DNA extraction was done followed by conventional PCR, and the PCR product was digested by Bst<sub>u</sub>I restriction enzyme through the Restriction Fragment Length Polymorphism (RFLP) technique. Out of the 3 genotypes, GA genotype was found to be uniquely associated with the risk for lupus nephritis despite the lack of association with cutaneous and joint involvement. Conversely, there was no significant association between the polymorphism with the risk for developing SLE.

**Conclusion and recommendations:** TLR9 gene polymorphism plays an important role in the pathogenesis of LN. Therefore, TLR9 gene expression in kidney tissue is an attractive target for further comprehensive studies in kidney tissue in LN patients to assess its value as a target for immune-modulatory therapy.

### Biography:

Islam Abdelkader a physician and pathologist who has developed an appetite biological research since graduation from medical school. His field of interest is immunology and molecular genetics. As a postgraduate medical student, he has been trained on various clinical and basic medical sciences.

### Publications

1. Maia ACRG, Porcino GN, Quellis LR, Emídio NB, Marconato DG, Messias WF, Soldati LL, Faria-Pinto P, Capriles PVSZ, Coimbra ES, Marques MJ, Vasconcelos EG (2019) Leishmania infantum amastigote nucleoside triphosphate diphosphohydrolase 1 (NTPDase 1): Its inhibition as a new insight into mode of action of pentamidine. *Experimental Parasitology* 200: 1-6.
2. Maia ACRG, Porcino GN, Faria-Pinto P, Mendes TV, Antinarelli LMR, Coimbra ES, Reis AB, Juliano L, Juliano MA, Marques MJ, Vasconcelos EG (2019) Leishmania infantum nucleoside triphosphate diphosphohydrolase 1 (NTPDase 1) B-domain: Antibody antiproliferative effect on the promastigotes and IgG subclass responses in canine visceral leishmaniasis. *Veterinary Parasitology* 271: 38-44.



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