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## Master Thesis Defense

Entitled

*GENETIC VARIATION OF HUMAN LEUKOCYTE ANTIGEN (HLA) ALLELES AMONG RHEUMATOID ARTHRITIS PATIENTS IN FUJAIRAH*

by

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Abstract

Rheumatoid arthritis (RA) is an autoimmune disease characterized by chronic inflammation that affects the joints. It occurs when the immune system attacks the body's own tissues and organs. The precise cause of RA is not fully understood but evidence suggests that genes, hormones and environmental factors could be involved. It has been established that human leukocyte antigen (HLA) loci is a strong risk factor of RA. Specifically, the shared epitope (SE) region of HLA-DRB1 and variations at position 11 and 13 of HLA-DRB1 and position 77 of HLA-A have shown strong association with RA. The hypothesis of this study was that these variations could be associated with RA patients in the UAE. To study this, regions of the HLA-DRB1 and HLA-A genes from RA and control samples from Fujairah Hospital were sequenced and analyzed. Results indicated that there was minimal incidence of high risk HLA-DRB1 SE or variations at position 11 and 13 of HLA-DRB1 in these samples. An asparagine at position 77 (Asn77) appeared to be a much strong biomarker for RA in this population (Odds ratio = 20.52, p-value = 0.0001985). Additionally, missense mutations Arg56 and Glu76 were also observed with significantly higher incidence in RA samples. The relevance of these variations warrant further investigation. In summary, HLA-A appears to be a stronger indicator of RA than HLA-DRB1 in the samples analyzed.

**Keywords:** Rheumatoid arthritis, human leukocyte antigen, shared epitope, HLA-DRB1, HLA-A.