



جامعة الإمارات العربية المتحدة
United Arab Emirates University

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Master Thesis Defense

Entitled

*A STUDY ON VITAMIN D METABOLIC GENES SINGLE NUCLEOTIDE POLYMORPHISMS AND
THEIR LINKAGE IN ADULT ASTHMATIC EMIRATIS*

by

Amneh Mohammad Ahmad Al Mousa

Faculty Advisor

Dr. Youssef Abouzaid, Department of Biology

College of Science

Date & Venue

12:30 PM

Thursday, 18 April 2019

Room 234, F3 Building

Abstract

Vitamin D normal levels are vital to many biological processes including its classical role in calcium and phosphate homeostasis, and regulation of immune and non-immune cells. Vitamin D mediates its actions via the vitamin D receptor (VDR) which is expressed in most tissues of the body including the airway smooth muscle. Asthma is a syndrome of chronic inflammatory airway disease. We aim to investigate whether asthma and its severity were associated with single nucleotide polymorphisms (SNPs) in vitamin D metabolic genes as well as vitamin D levels in asthmatic adult Emiratis. We conducted a study that included 132 adult asthmatic patients and 164 non-asthmatic controls of both sexes. Four SNPs in VDR gene (rs731236, rs7975232, rs1544410, and rs2228570), one SNP in vitamin D 25-hydroxylase gene (CYP2R1; rs12794714) and two SNPs in vitamin D binding protein gene (GC; rs4588 and GC; rs7041) were genotyped using TaqMan PCR genotyping techniques. In conclusion, VDR; rs7975232 is significantly associated with asthma severity, and the CC genotype increases the risk of severe asthma by 2.7-fold. Moreover, the ACG haplotype of three SNPs in the VDR gene showed a significant association with asthma severity. VDR rs7975232 polymorphism may be considered as a biomarker for asthma severity and possibly play a role in the management of asthma in adult Emirati patients.

Keywords: Asthma, Vitamin D, Single nucleotide polymorphisms, Haplotype.