

Vitamin D Receptor (VDR) gene polymorphisms and their link to Infectious Disease Susceptibility.

¹Krugel, C., ¹Bornman, L.

Department of Biochemistry, University of Johannesburg, PO Box 524, Auckland Park, 2006, South Africa.

Background: The vitamin D receptor (VDR) is a transcription factor mediating genomic responses to the biologically active form of vitamin D, 1,25(OH)₂-D₃, a key modulator of the immune system. A number of studies have shown associations between VDR gene (*VDR*) polymorphisms and infectious diseases. Knowledge on how these polymorphisms modulate this endocrine system and confer risk of disease is hindered by the fact that several of the associated allelic variants are located in introns or are synonymous and serve as markers within the haplotype covering disease-causing alleles. **Objectives:** To identify alleles/haplotypes of *VDR* commonly found in an American population and to study their functional impact on DNA and protein level. **Methods:** Cell lines from a number of individuals were established. VDR function is investigated using the expression of cytokines expressed or inhibited by the VDR-complex. **Results:** Preliminary data obtained from genotyping and flow cytometric analysis showed that from 6 individuals genotyped, the majority were heterozygous for the *FokI* SNP. Preliminary results proposed that VDR may play a role in the Th1 immune response as the mean channel fluorescence indicated a higher ratio of VDR in the presence of the less active (f) allele. No significant differences were observed but more individuals need to be included to verify any significance. **Conclusions:** Inconsistencies in association between *VDR* polymorphisms and infectious disease susceptibility might be better understood as the project progress. Early identification of individuals at risk for infectious diseases and appropriate treatment could lead to prevention/delay in manifestation of the disease.