Research article

**Association between vitamin D receptor genes polymorphisms with systemic lupus erythematosus in children**

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**Abstract**

**Background:** Several vitamin D receptor polymorphisms have been associated with SLE development and clinical manifestations. So, the aim of the study was to evaluate the possible association between the common VDR gene polymorphisms and SLE susceptibility and severity in children. **Patients and methods:** Thirty six lupus children, 29 females (80.6%) and 7 males (19.6 %) were recruited from the Collagen Clinic, Children Hospital, Cairo University between February and November, 2016. Forty three healthy age and sex matched children were included as a control group. Five ml venous blood sample was withdrawn under aseptic conditions from the studied groups. Two ml were put in a sterile tube containing EDTA for genotyping of BsmI (rs1544410), ApaI (rs7975232), TaqI (rs731236) and FokI (rs2228570) polymorphisms by real time PCR. The rest was centrifuged and serum was used for assessment of: serum calcium, vitamin D level, total and intact Parathormone (IPTH) level, and fibroblast growth factor 23 (FGF23). All were done by ELISA. **Results:** The serum calcium, vitamin D level and IPTH were significantly lower in patients than that in controls (p=0.001, p=0.001 & p=0.007) respectively, while FGF23 was significantly higher in the patients (p=0.02). The risk of SLE was significantly higher among patients carrying (Fok1 AA) (OR=2.6, & p =0.04). SLICC values >3 were higher in SLE patients with FokI AA and BsmI CT genotypes (p=0.03 & p=0.1 respectively). GACC haplotype was significantly higher in patients (p=0.02, OR=3.24). **Conclusion:** The VDR gene polymorphisms and haplotypes were associated with SLE. FokI AA and BsmI CT genotypes carry a worse prognosis.