

***Assessment of vitamin D receptor (VDR) gene
polymorphisms and bone mineral density (BMD)
in Egyptian rheumatoid arthritis patients***

Thesis

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By

Manar Ragab Senosi

M.B.; B. Ch. Faculty of Medicine, Fayoum University
Ass. Lecturer of Rheumatology and Rehabilitation
Faculty of Medicine, Fayoum University

Supervised by

Prof. Dr. Tamer Mohamed Atef Gheita

Professor of Rheumatology and Rehabilitation
Faculty of Medicine, Cairo University

Dr. Hanan Mohamed Mohamed Fathi

Ass. Prof. of Rheumatology and Rehabilitation
Faculty of Medicine, Fayoum University

Dr. Noha Mahmoud Abdel-Baki

Ass. Prof. of Rheumatology and Rehabilitation
Faculty of Medicine, Cairo University

Dr. Ahmed Mohamed Magdy Mahmoud

Lecturer of Radiodiagnosis
Faculty of Medicine, Fayoum University

Faculty of Medicine

Cairo University

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Abstract

Objective: to assess vitamin D receptor (VDR) gene polymorphisms, bone mineral density and to investigate the possible risk factors of osteoporosis and fracture in rheumatoid arthritis (RA). **Methods:** 97 RA patients and 45 matched controls were enrolled. Serum vitamin D level, VDR genotyping, dual energy X-ray absorptiometry (DEXA) scan, trabecular bone score (TBS) and fracture risk assessment in 10 years (FRAX) were assessed. Disease activity score (DAS28) and modified health assessment questionnaire (MHAQ) were measured. **Results:** Mean age of the patients was 47.9 ± 8.9 years; 85 females, 12 males (F:M 7.1:1) and mean disease duration 9.4 ± 6.2 years. DAS28 was 4.52 ± 1.04 and MHAQ 0.6 ± 0.4 . There was a significant difference between cases and controls as regards DEXA and FRAX ($p < 0.0001$) but the TBS and VDR genotyping were comparable ($p = 0.29$ and $p = 0.12$, respectively). The vitamin D level was comparable with the control (9.3 ± 6.5 vs 10.4 ± 7.5 ng/ml, $p = 0.4$). None of the patients were receiving anti-osteoporotic therapy or biologic therapy. There was a significant association between the presence of osteoporosis and age, disease duration, menopause and rheumatoid factor (RF) positivity. The TBS was significantly lower and FRAX higher in patients with positive RF and anti-CCP. FRAX was significantly related and the TBS inversely with the age, disease duration, serum uric acid, alkaline phosphatase and MHAQ. **Conclusion:** Reduced BMD and increased tendency to fractures are remarkable in RA patients. Vitamin D level was decreased in patients and control and VDR gene polymorphisms were not linked to RA. TBS and FRAX are effective tools to assess osteoporotic fractures in RA.

Keywords: Bone mineral density; VDR gene; FRAX; TBS; rheumatoid arthritis; DAS28