

VITAMIN D DEFICIENCY IS NOT MORE FREQUENT IN NF1: A CASE-CONTROL STUDY FROM BRAZIL

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Introduction: Neurofibromatosis type 1 (NF1) is an autosomal dominant disorder caused by mutations in the NF1 gene (17q11.2, neurofibromin). The estimated incidence is approximately 1:3500 newborns and clinical diagnosis is possible in the majority cases. A few studies suggest that patients with NF1 are more likely to have vitamin D deficiency when compared to the general population. Objectives: The goal of the study was to determine the levels of 25-OH-vitamin D [25(OH)D] in individuals with NF1 and disease-unaffected controls and analyze common VDR gene polymorphisms (BsmI and FokI) to verify whether they were associated with lower vitamin D levels. Methods: Case-control study that included 45 NF1 patients from Southern Brazil and 45 healthy controls matched by sex, skin type and age to the cases. Results and conclusions: Overall, 70 (77.8%) of the individuals studied had levels of 25(OH)D below 30ng/mL: vitamin D deficiency was observed in 28 (31.1%) and vitamin D insufficiency in 42 (46.6%) subjects. 25(OH)D deficiency or insufficiency were not more frequent in NF1 patients than in controls ($p=0,074$). We did not observe an association of VDR gene polymorphisms and vitamin D levels suggesting that the insufficient or deficient biochemical phenotypes in the patients studied here are not associated with these genetic variants.